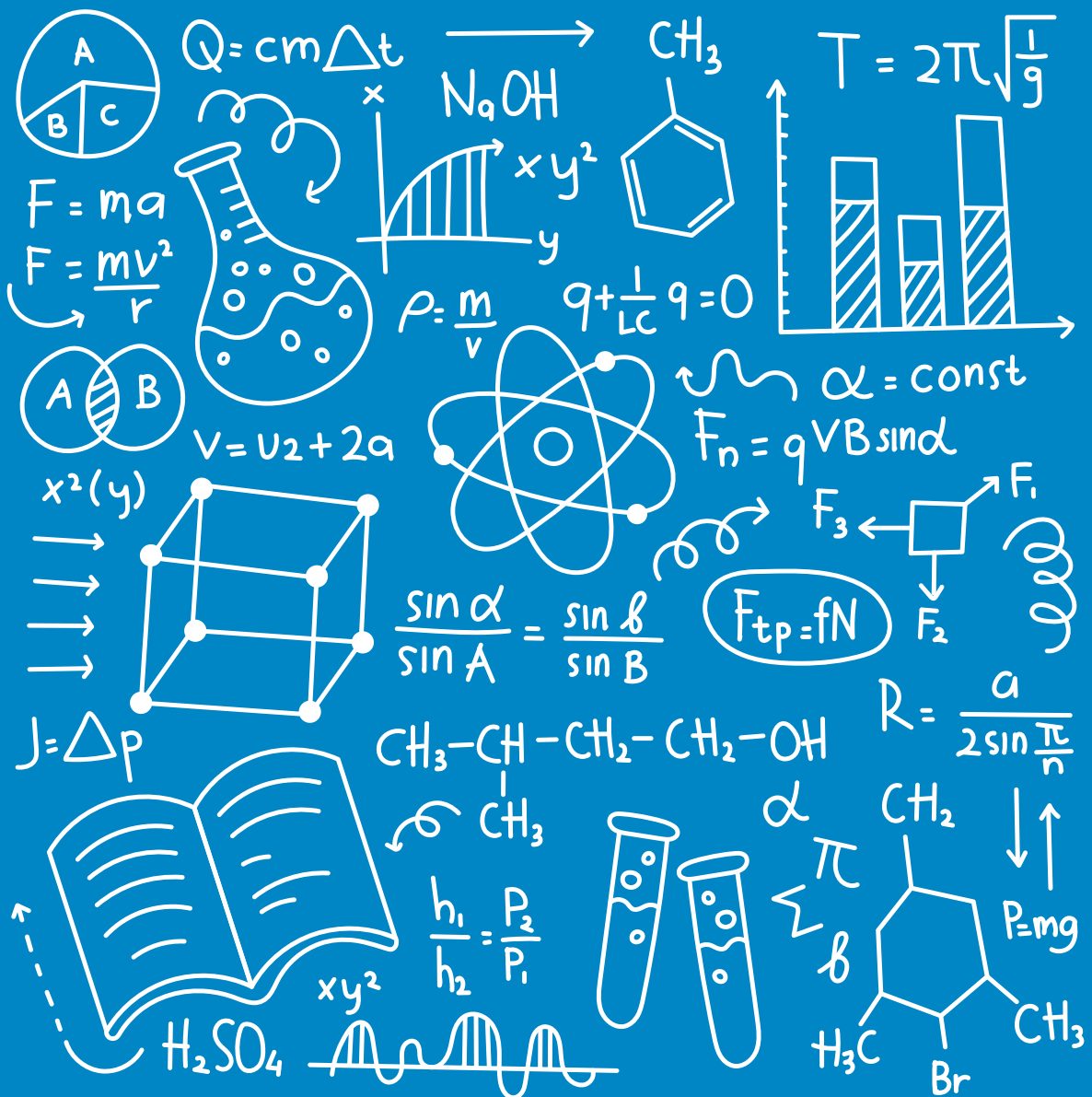


ANNUAL SCIENTIFIC REPORT

2023





SJD

Sant Joan de Déu
Institut de Recerca 

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Sant Joan de Déu, Parc Sanitari Sant
Joan de Déu & Institut de Neurociències ·
Universitat de Barcelona



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1

FOREWORD

Governance and structures
Global numbers 2023
Global numbers 2015-2023
Institutional awards



Governance and structures

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Director of Research, Innovation and Learning, SJD Barcelona Children's Hospital

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Director General of Health Planning. Generalitat de Catalunya

Dra. Laia Arnal Arasa

Director General of the General Directorate of Knowledge Transfer. Generalitat de Catalunya

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Director, Fundació de Recerca Sant Joan de Déu

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Director of IRSJD. Director of Research, Innovation and Learning at the SJD Barcelona Children's Hospital

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Delegate, Institut de Neurociències - Universitat de Barcelona (UBNeuro)

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Director, Fundació de Recerca Sant Joan de Déu

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Scientific Manager, Institut de Recerca Sant Joan de Déu

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Scientific director. European infrastructure for translational medicine (EATRIS)

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Head of Laboratory in Gene Therapy, Inserm, Genethon

Dr. Philippe Courtet

Professor of Psychiatry at the Université de Montpellier and Head of Department of Emergency care Psychiatry at the University Hospital, Montpellier

Dra. Marisol Montolio

Scientific director. Duchenne Parent Project, Spain

Dr. Carlos Rodríguez-Galindo

Executive vice-president. St. Jude Children's Research

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Chairman and Director, Institute for Psychiatric Phenomics and Genomics, LMU Munich and Johns Hopkins University's Department of Psychiatry

Dra. Maithe Tauber

Hospital Paediatric endocrinologist. Institut des Sciences du Cerveau, de la Cognition et du Comportement de Toulouse (ISC3T)

Governance and structures

Research commission

President

Dr. Cecilia Jiménez-Mallebrera

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Dr. Marta Camprubí Camprubí

Dr. Roser Pueyo

Dr. Guerau Fernández Isern

Secretary

Ms. Júlia Ribot Ballabriga



Global numbers 2023

762

RESEARCH STAFF



- 58 Group leaders
- 142 Staff researchers
- 385 Postdoc & associate researchers
- 93 PhD Students
- 84 Assistant researcher & Technicians

And **106** staff in corporate services

484

ACTIVE PROJECTS



- 82 Competitive autonomic
- 210 Competitive national
- 58 Competitive international
- 3 Non Competitive autonomic
- 121 Non Competitive national
- 10 Non Competitive international

129

NEW PROJECTS

- 36 Competitive autonomic
- 38 Competitive national
- 23 Competitive international
- 30 Non Competitive national
- 2 Non Competitive international



19.1 M

BUDGET*



- 66 % Private
- 34 % Public

406

ACTIVE CLINICAL TRIALS & OBSERVATIONAL STUDIES



- 33 Phase I
- 80 Phase II
- 147 Phase III
- 8 Phase IV
- 42 Other
- 96 Observational studies

712

PUBLICATIONS



(Original articles & reviews published in academic journals indexed in Clarivate's Journal Citation Reports 2022)

- 5021 Impact factor
- 7,1 Average impact factor
- 60% Publications in JCR first quartile journals
- 46% Publications with main contributing authors

95

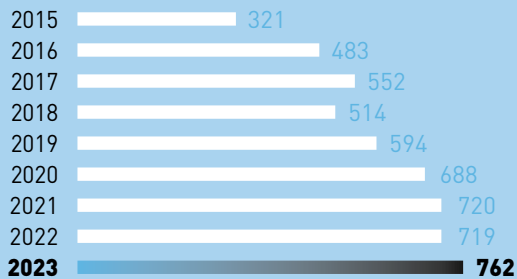
NEW CLINICAL TRIALS & OBSERVATIONAL STUDIES

- 8 Phase I
- 20 Phase II
- 36 Phase III
- 1 Phase IV
- 12 Other
- 18 Observational studies

Global numbers 2015-2023

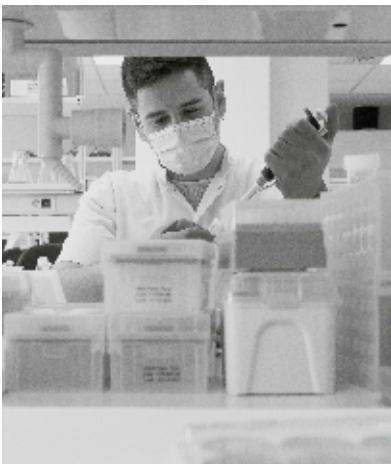
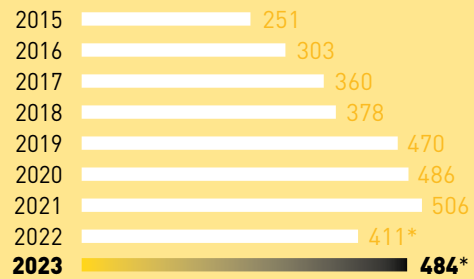
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RESEARCH STAFF



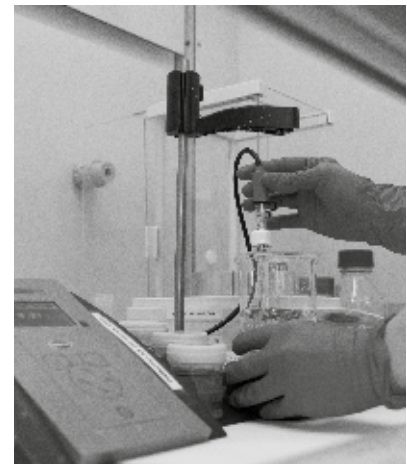
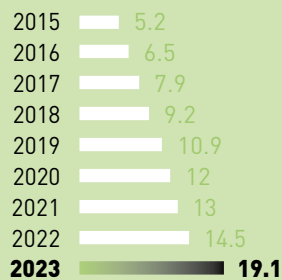
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PROJECTS



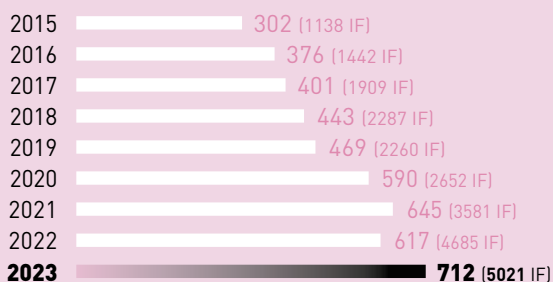
19.1 M

BUDGET**



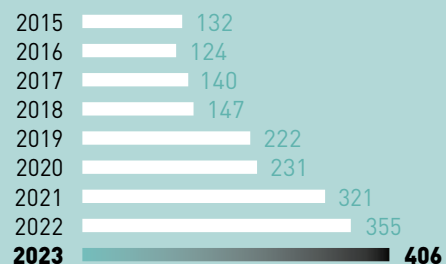
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PUBLICATIONS



406

CLINICAL TRIALS & OBSERVATIONAL STUDIES



* Only the projects managed by the Fundació de Recerca Sant Joan de Déu have been accounted for.
** Millions of euros

Institutional awards

HRS4R award

HRS4R Human Resources Strategy for Researchers

<https://www.irsjd.org/en/research/hrs4r-hr-excellence-research/>

Fundació de Recerca Sant Joan de Déu is awarded with the logo "HR Excellence in research" since 2018. Thus, it has acquired a commitment to implement the European Charter for Researchers and the Code of Conduct for the Recruitment of Researchers (Charter & Code) in their policies and practices.

CERCA

The Institut de Recerca Sant Joan de Déu (IRSJD) has been recognized by the board of trustees of the Institution for Research Centers in Catalonia (CERCA) as a CERCA centre in 2020

This award involves the development of a frontier research aimed at the scientific and economic impact and the improvement of the social and individual wellbeing.

IRSJD is a research center in biomedicine formed by groups from two university hospitals, Sant Joan de Déu Hospital and the Health Park Sant Joan de Déu, and three research institutes, the Institute of Biomedicine (IBUB) and the Institute of Neurosciences (UBNeuro), both from the UB, and thUPC's Research Centre in Biomedical Engineering.

The CERCA institute focuses its actions on the contribution to the international presence of research centres from Catalonia and to promote the cooperation and scientific exchange with the top centres and universities worldwide. Moreover, it promotes the adoption of joint policies in research management, scientific development and knowledge transfer. Also, it works in order to increase and ease knowledge transfer in the business sector and general public, promoting the involvement of these agents in the field of research.





2 STAFF

Research staff



Research staff

**762****243** men (32%) **519** women (68%)**58** GROUP LEADER**29** men (50%) **29** women (50%)**142** STAFF RESEARCHER**27** men (19%) **115** women (81%)**385** POSTDOC ASSOCIATE RESEARCHER**135** men (35%) **250** women (65%)**93** PhD STUDENT**23** men (25%) **70** women (75%)**84** TECHNICIAN AND ASISTANT RESEARCHERS**29** men (35%) **55** women (65%)



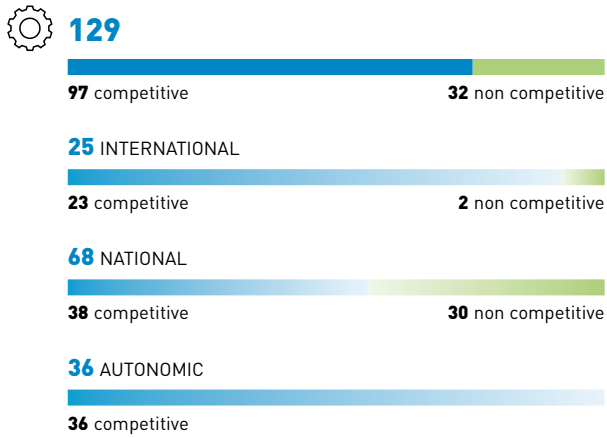
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PROJECTS AND CLINICAL STUDIES

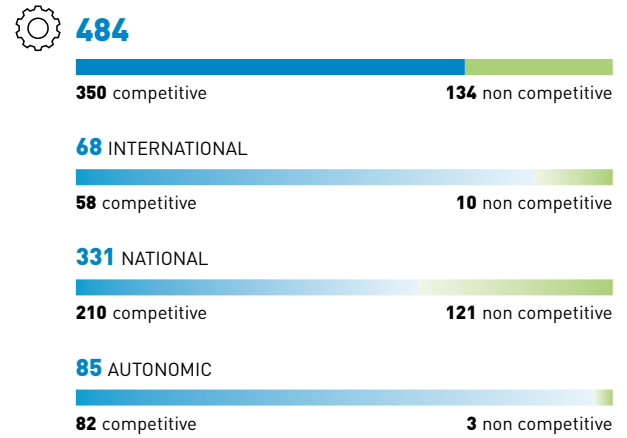
- New projects
- Active projects
- Active clinical studies
- New clinical studies



New projects

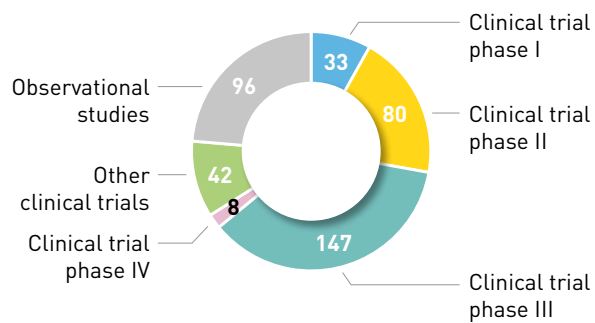


Active projects

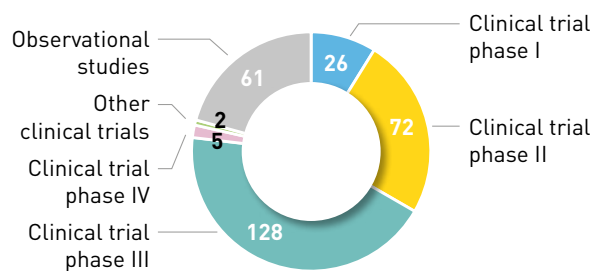


Active clinical studies

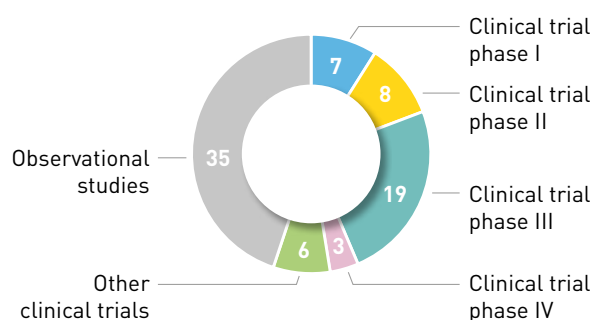
All clinical studies by type



Only commercial clinical studies



Only academic clinical studies



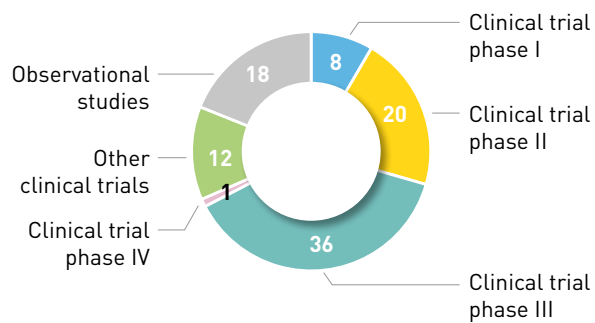
Total active per service



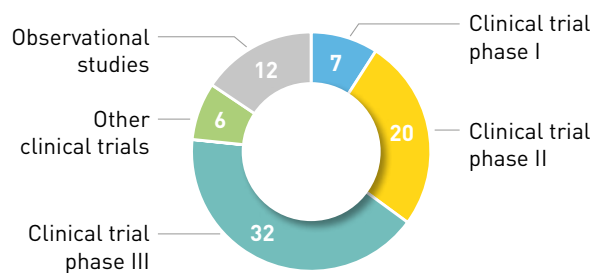
	Clinical Trials (Phase I, II, III, IV)	Clinical Trials (Others)	Observational Studies
Neurology	76	6	13
Oncology	49	2	10
Rheumatology	18	1	9
Dermatology	24	0	5
Infectious diseases	17	1	5
Clinical haematology	11	0	11
Allergy and clinical immunology	12	1	7
Endocrinology	9	0	8
Gastroenterology, hepatology and nutrition	9	3	7
Internal medicine	4	1	5
Gynaecology and obstetrics	8	0	2
Pneumology	9	0	6
Nephrology	8	2	1
Psychiatry and psychology	5	2	1
Cardiology	4	0	0
Intensive care unit	3	2	0
Neonatology	3	2	1
Ophthalmology	0	1	2
Anaesthesia	3	4	1
Orthopaedics and traumatology	1	2	0
Digestive medicine	1	0	0
Emergency room	1	1	0
Rehabilitation and physical medicine	1	1	0
Otorhinolaryngology	0	0	2
Pediatric	0	2	0
Pharmacy	0	0	2
Nursing	0	2	0
TOTAL	276	34	96

New clinical studies

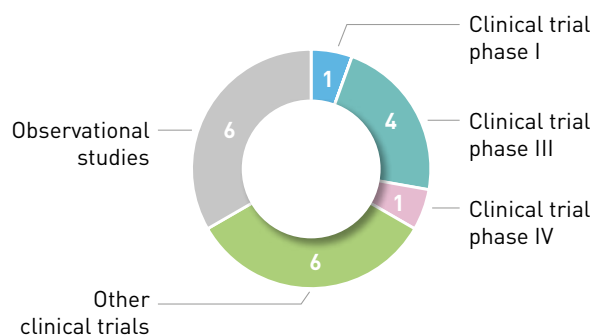
All clinical studies by type



Only commercial clinical studies



Only academic clinical studies



Total new per service

	Clinical Trials (Phase I, II, III, IV)	Clinical Trials (Others)	Observational Studies
Neurology	19	1	3
Oncology	11	1	2
Rheumatology	1	0	3
Dermatology	7	0	1
Infectious diseases	5	0	1
Clinical haematology	2	0	0
Allergy and clinical immunology	4	0	1
Endocrinology	1	0	2
Gastroenterology, hepatology and nutrition	1	1	1
Gynaecology and obstetrics	1	0	1
Pneumology	1	0	0
Nephrology	5	0	2
Psychiatry and psychology	3	0	1
Cardiology	2	1	0
Intensive care unit	1	2	0
Neonatology	0	2	0
Ophthalmology	1	1	0
Anaesthesia	0	1	0
Orthopaedics and traumatology	0	0	1
Digestive medicine	1	0	0
Nursing	0	1	0
Pharmacy	0	0	1
Otorhinolaryngology	1	0	0
TOTAL	66	10	19



4

SCIENTIFIC PRODUCTION AND RESULTS

Scientific publications
High Influence Journals
Clinical guidelines, consensus documents & recommendations
Patents, utility models and spin-offs
Doctoral Theses



Scientific publications

Publications (Original articles & reviews)

Number 712	Impact Factor 5021	Average IF 7,1	% Q1 60%	% D1 25%	% Leadership 46%
% D1 in leadership 18%	% Female leadership 53%	Normalized Impact (2019-2023) 2,35	Highly Cited Papers 10% (2019-2023) 19,3%	% Open Access 70%	% International collaboration 57%

All publications by type

	Number	Impact Factor	Average IF	% Q1	% D1	% Leadership
Original articles	656	4713	7,2	61%	26%	46%
Review	56	308	5,5	55%	25%	50%
Scientific Letters	31	187	6,0	52%	26%	52%
Case reports	2	6	3,0	50%	0%	0%
Editorial material (editorials and letters to the editor)	22	140	6,4	46%	18%	73%
Total*	767	5354	7,0	59%	25%	47%

* Include 19 clinical guidelines, consensus documents and recommendations

Methodology and Definitions

- Except when stated otherwise, indicators take into account original articles & reviews published in academic journals indexed in Clarivate's Journal Citation Reports 2022
- Only publications with at least one IRSJD author (no collaborator)
- No ahead of print publications are taken into account
- Leadership indicators take into account first, last and/or corresponding author
- Open Access takes into account green, gold and hybrid route publications, not bronze, according to the Unpaywall database
- Normalized impact and HCP10% indicators take into account articles and reviews from 2019 to 2023, and are calculated using Clarivate's ESI (Essential Science indicators) baseline citation rates and baseline percentiles from its 09/05/2024 update.
- Citations to calculate the CROWN and HCP10% indicators were downloaded from Clarivate WoS (Web of Science) Core Collection via Fundanet, on 14/05/2024. Citation count is not fractional.
- Normalized impact shows the relative number of citations to publications, compared to the global average of citations to publications of the same type, year of publication and thematic area, being 1 the baseline or world average.
- HCP10% (Highly Cited Papers 10%) shows the publications included in 10% of the world's most cited publications of the same type, year of publication and subject area, being 10% the expected value (higher is better).

High Influence Journals

Original articles & reviews in first decile (D1) journals

Journal	Total articles and reviews	Journal's impact Factor (JCR 2022)
Lancet	1	168,9
N. Engl. J. Med.	2	158,5
Lancet Psychiatry	1	64,3
Science	2	56,9
Lancet Public Health	1	50
Lancet Neurol.	1	48
J. Clin. Oncol.	2	45,4
Lancet Child. Adolesc. Health	1	36,4
Lancet Glob. Health	1	34,3
Lancet Digit. Health	1	30,8
Ann. Rheum. Dis.	1	27,4
J Travel Med	2	25,7
Nat. Cell Biol.	1	21,3
Blood	2	20,3
Circ.Res.	1	20,1
Br. J. Sports Med.	1	18,6
Nat Commun	5	16,6
Angew. Chem.-Int. Edit.	1	16,6
Lancet HIV	1	16,1
Neuro-Oncology	1	15,9
J. Clin. Invest.	1	15,9
J. Exp. Med.	2	15,3
npj Digit. Med.	1	15,2
EClinicalMedicine	5	15,1
Adv. Sci.	1	15,1
Brain	5	14,5
Clin. Microbiol. Infect.	2	14,2
Hepatology	1	14

Journal	Total articles and reviews	Journal's impact Factor (JCR 2022)
JAMA Netw. Open	5	13,8
Trends Pharmacol. Sci.	1	13,8
Trends Mol. Med	1	13,6
Sci Adv	2	13,6
Small	1	13,3
J. Am. Acad. Child Adolesc. Psychiatr.	2	13,3
J Med Virol	10	12,7
Acta Neuropathol.	1	12,7
Clin Gastroenterol Hepatol	1	12,6
Allergy	1	12,4
Genome Med.	1	12,3
Environ. Int.	1	11,8
Clin. Cancer Res.	1	11,5
Redox Biol.	3	11,4
Psychiatry Res	4	11,3
Ann. Neurol.	1	11,2
EMBO Mol Med	2	11,1
EBioMedicine	1	11,1
Mol Psychiatry	1	11
Additive Manufacturing	1	11
Br. J. Psychiatry	2	10,5
Neurology	2	10,1
Adv. Healthc. Mater.	2	10
Crit. Rev. Clin. Lab. Sci.	1	10
Am J Obstet Gynecol	2	9,8
Am. J. Hum. Genet.	1	9,8
Asian J. Psychiatr.	3	9,5

High Influence Journals

Journal	Total articles and reviews	Journal's impact Factor (JCR 2022)
Pharmacol. Res.	2	9,3
Span J Psychiatry Ment Health	2	9,2
J. Cachexia Sarcopenia Muscle	2	8,9
Obes. Rev.	1	8,9
Genet. Med.	1	8,8
Neurol.-Neuroimmunol. Neuroinflammation	1	8,8
World J Pediatr	4	8,7
Int. J. Behav. Nutr. Phys. Act.	1	8,7
JMIR Public Health Surveill	1	8,5
Stroke	1	8,4
Sens. Actuator B-Chem.	1	8,4
Neurosci Biobehav Rev	3	8,2
Rev. Endocr. Metab. Disord.	1	8,2
BMJ Glob. Health	1	8,1
Epidemiol Psychiatr Sci	3	8,1
Int. J. Mental Health Addict.	1	8
Oncogene	1	8
Br J Health Psychol	1	7,9
Transl Res	1	7,8
J Adolesc Health	1	7,6
J. Child Psychol. Psychiatry	2	7,6
Biomed Pharmacother	2	7,5
J. Headache Pain	1	7,4
J Med Internet Res	2	7,4
J. Pathol.	1	7,3
Ultrasound Obstet. Gynecol.	6	7,1
Antioxidants	3	7
Psychol Med	10	6,9
J Invest Dermatol	1	6,5

Journal	Total articles and reviews	Journal's impact Factor (JCR 2022)
Eur Child Adolesc Psychiatry	4	6,4
Am J Obstet Gynecol MFM	2	6,3
Addiction	1	6,1
J. Eur. Ceram. Soc.	1	5,7
Soc. Sci. Med.	1	5,4
J. NeuroEng. Rehabil.	1	5,1
IEEE Trans. Neural Syst. Rehabil. Eng.	1	4,9
Child Abuse Negl.	1	4,8
Pediatr Allergy Immunol	4	4,4
Arch. Dis. Child.-Fetal Neonatal Ed.	1	4,4
Eur J Psychotraumatol	1	4,2
Ear Hear.	1	3,7
Frontiers in Veterinary Science	1	3,2
Brain Struct. Funct.	1	3,1
Med Mycol	1	2,9

High Influence Journals

Original articles & reviews in first decile (D1) journals with author's leadership

Reference	Journal	Journal's impact Factor (JCR 2022)
Myall K, Montero J, Gorczynski P, Kajee N, Syed R, Bernard R et al. Effect of mindfulness-based programmes on elite athlete mental health: a systematic review and meta-analysis. <i>Br. J. Sports Med.</i> 2023;57(2):99-108. IF:18,600. (Q1).	Br. J. Sports Med.	18,6
García AP, Pérez de Diego R, Flores C, Rinchai D, Solé J, Deyà A et al. Humans with inherited MyD88 and IRAK-4 deficiencies are predisposed to hypoxemic COVID-19 pneumonia. <i>J. Exp. Med.</i> 2023;220(5)IF:15,300. (Q1).	J. Exp. Med.	15,3
Živanic M, Espona A, Lin A, Canal C. Current State of Cold Atmospheric Plasma and Cancer-Immunity Cycle: Therapeutic Relevance and Overcoming Clinical Limitations Using Hydrogels. <i>Adv. Sci.</i> 2023;10(8)IF:15,100. (Q1).	Adv. Sci.	15,1
Tangeras T, Ribeiro J, Backe PH, De Oyarzabal AL, Neugebauer J, Weinhold N et al. BCKDK deficiency: a treatable neurodevelopmental disease amenable to newborn screening. <i>Brain.</i> 2023;146(7):3003-3013. IF:14,500. (Q1).	Brain	14,5
Ousingsawat J, Talbi K, Gómez H, Koy A, Fernández A, Tekgül H et al. Broadening the clinical spectrum: molecular mechanisms and new phenotypes of ANO3-dystonia. <i>Brain.</i> 2023;IF:14,500. (Q1).	Brain	14,5
Barroso E, Montori M, Wahli W, Palomer FX, Vazquez M. Striking a gut-liver balance for the antidiabetic effects of metformin. <i>Trends Pharmacol. Sci.</i> 2023;44(7):457-473. IF:13,800. (Q1).	Trends Pharmacol. Sci.	13,8
Crovetto F, Nakaki A, Arranz A, Borrás R, Vellvé K, Paules C et al. Effect of a Mediterranean Diet or Mindfulness-Based Stress Reduction During Pregnancy on Child Neurodevelopment A of the IMPACT BCN Randomized Clinical Trial. <i>JAMA Netw. Open.</i> 2023;6(8)IF:13,800. (Q1).	JAMA Netw. Open	13,8
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Reference	Journal	Journal's impact Factor [JCR 2022]
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High Influence Journals

Reference	Journal	Journal's impact Factor [JCR 2022]
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Zhang M, Barroso E, Ruat M, Peña L, Peyman M, Aguilar D et al. Elafibranor upregulates the EMT-inducer S100A4 via PPAR β /d. <i>Biomed Pharmacother</i> . 2023;167:115623-115623. IF:7,500. (Q1).	Biomed Pharmacother	7,5
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Garin JPL, Sampor C, Grigorovski N, Ceciliano A, Fandiño A, Ossandon D et al. How we approach conservative treatment of retinoblastoma in South America in the era of local ocular treatments: A consensus of the Grupo America Latina de Oncologia Pediátrica (GALOP). <i>Pediatr. Blood Cancer.</i> 2023;70(7)IF:3,200. (Q2).	<i>Pediatr. Blood Cancer</i>
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Patents, utility models and spin-offs

2023

NEW PATENT FAMILIES

5 APPLICATIONS

10 GRANTED

Up to 2023

PATENT FAMILIES

31 APPLICATIONS

31 GRANTED

19 LICENSES

UTILITY MODEL FAMILIES

7 APPLICATIONS

7 GRANTED

1 LICENSES

New patents, license agreements and utility models

Treatment of hepatic steatosis related oligo-ovulation

Date: 29/11/2023

Country: Corea del Sud

Number: KR102608158B1

Inventors: Ibañez Lourdes; De Zegher Francis

Proprietors: Univ Leuven Kath, SJD Barcelona Children's Hospital

Rajah Vignesh; J Møller San-Pedro Claus

Proprietors: Y Mabs Therapeutics Inc [US]; Sant Joan de Déu Hospital [ES]; Memorial Sloan Kettering Cancer Center [US]; Sloan Kettering Inst Cancer Res [US]; Memorial Hospital For The Treat Of Cancer And Allied Diseases [US]

Esteve Arnau [ES]; Pons Odena Martí [ES]; Alaez Vasconcellos Carlos [ES]; Font Vizcarra Lluís [ES]; Bermúdez Castel Adrian [ES]; Pena Pérez Xoel [ES]; Murcia Laguna Marc [ES]
Proprietors: Sant Joan de Déu Hospital [ES]

ANTI-GD2 administration regimen

Date: 29/11/2023

Country: Canada

Number: KR102608158B1

Inventors: Mora Graupera Jaume; Cheung Nai-Kong; Modak Shakeel; Rajah Vignesh; J Møller San-Pedro Claus

Proprietors: Uy Mabs Therapeutics Inc [US]; Sant Joan de Déu Hospital [ES]; Memorial Sloan Kettering Cancer Center [US]; Sloan Kettering Inst Cancer Res [US]; Memorial Hospital For The Treat Of Cancer And Allied Diseases [US]

ANTI-GD2 administration regimen

Date: 14/06/2023

Country: Taiwan

Number: TW202330033

Inventors: Mora Graupera Jaume; Cheung Nai-Kong; Modak Shakeel; Rajah Vignesh; J Møller San-Pedro Claus

Proprietors: Y Mabs Therapeutics Inc [US]; Sant Joan de Déu Hospital [ES]; Memorial Sloan Kettering Cancer Center [US]; Sloan Kettering Inst Cancer Res [US]; Memorial Hospital For The Treat Of Cancer And Allied Diseases [US]

Peptidic conjugates of SN38 useful in the treatment of cancer

Date: 15/08/2023

Country: Australia

Number: AU2021347611

Inventors: Sanchez Navarro Macarena [ES]; Teixidó Turá Meritxell [ES]; Gené Olaciregui Nagore [ES]; Giralt Lledó Ernest [ES]; Montero Carcaboso Angel [ES]

Proprietors: Fundacio Inst De Recerca Biomedica Irb Barcelona [ES]; Univ Barcelona [ES]; Sant Joan de Déu Hospital [ES]

ANTI-GD2 administration regimen

Date: 14/06/2023

Country: Europa

Number: EP4194471

Inventors: Mora Graupera Jaume; Cheung Nai-Kong; Modak Shakeel;

Respirator valve

Date: 30/08/2023

Country: Europa

Number: EP3895752B1

Inventors: Palau Forte Enric [ES]; Valls

SN38 peptide conjugates useful for treatment of cancer

Date: 15/08/2023

Country: China

Number: CN116406301A

Inventors: Sanchez Navarro Macarena [ES]; Teixidó Turá Meritxell [ES]; Gené Olaciregui Nagore [ES]; Giralt Lledó

Patents, utility models and spin-offs

Ernest [ES]; Montero Carcaboso Angel [ES]

Proprietors: Fundacio Inst De Recerca Biomedica Irb Barcelona [ES]; Univ Barcelona [ES]; Sant Joan de Déu Hospital [ES]

Peptidic conjugates of SN38 useful in the treatment of cancer

Date: 02/08/2023

Country: Europa

Number: EP4217007A1

Inventors: Sanchez Navarro Macarena [ES]; Teixidó Turá Meritxell [ES]; Gené Olaciregui Nagore [ES]; Giralt Lledó Ernest [ES]; Montero Carcaboso Angel [ES]

Proprietors: Fundacio Inst De Recerca Biomedica Irb Barcelona [ES]; Univ Barcelona [ES]; Sant Joan de Déu Hospital [ES]

Peptidic conjugates of SN38 useful in the treatment of cancer

Date: 01/05/2023

Country: Israel

Number: IL301558A

Inventor: Sanchez Navarro Macarena [ES]; Teixidó Turá Meritxell [ES]; Gené Olaciregui Nagore [ES]; Giralt Lledó Ernest [ES]; Montero Carcaboso Angel [ES]

Proprietors: Fundacio Inst De Recerca Biomedica Irb Barcelona [ES]; Univ Barcelona [ES]; Sant Joan de Déu Hospital [ES]

Peptidic conjugates of SN38 useful in the treatment of cancer

Date: 06/10/2023

Country: Japan

Number: JP2023542382A

Inventors: Sanchez Navarro Macarena

[ES]; Teixidó Turá Meritxell [ES]; Gené Olaciregui Nagore [ES]; Giralt Lledó Ernest [ES]; Montero Carcaboso Angel [ES]

Proprietors: Fundacio Inst De Recerca Biomedica Irb Barcelona [ES]; Univ Barcelona [ES]; Sant Joan de Déu Hospital [ES]

Portfolio of spin-offs

Virmedex Virtual Experiences S.L.

2022 · <https://virmedex.com/>

Co-funders: Dani Tost, Núria Bonet & Ariel von Barnekow

Virmedex provides the next generation of training platforms for high performance risk-aware professionals. With more than 20 years in the field of serious games and gamification for health and education, they have partnered with hospitals, research centers, institutions and companies. They have designed gamification strategies and serious games for rehabilitation, screening, professional training and other applications.

Vincer.AI

2022 · <https://www.vincer.ai/>

Co-funders: Pol Solà-Santos, Alexandre Perera Lluna & Isidro Pascual.

Vincer. AI assist technological companies to create trusted AI

technologies and therefore help them provide credible AI models and output in a trusted business environment.

Their aim is to build trustworthy AI through auditing, rating, and tracing AI solutions in order to help organizations to improve their decision-making in AI and provide traceability and transparency to their clients.

Ephion Health S.L.

2021 · <https://ephion.health/>

Co-funders: Eurecat, Hospital Sant Joan de Déu and The Collider
Ephion Health is the platform to develop and deploy clinical measures for patient mobility. They assist the complete parametrization of mobility for several diseases and conditions, to create a positive impact on the patient's diagnosis, treatment and quality of life. Ephion's technology allows to save time and resources in clinical care and clinical trials.

Gate2Brain

2020 · <https://gate2brain.com/>

Co-funders: Universidad de Barcelona (UB), Hospital Sant Joan de Déu (HSJD)
Gate2Brain is a biotech company focused on the development of therapeutics that efficiently cross biological barriers such as the blood-brain barrier. We pursue our goals using a radically innovative peptide-based patented technology platform.

OMADRE

2019 · <https://www.omadre.com/>

Dr. Jaume Pérez Payarols (IRSJD)
The company Omadre was created to provide hospitals with the tools required for the efficient management of mother's milk, increasing the safety of existing processes in this way.

Patents, utility models and spin-offs

ABLE Human Motion

2018 ·

<https://www.ablehumanmotion.com/>

Co-funders: Dr. Alfons Carnicero, Dr. Alex García & Dr. Josep Maria Font (IRSJD)

ABLE Human Motion is a medical device spin-off from Universitat Politècnica de Catalunya (UPC) that is developing and will commercialize robotic exoskeletons to enhance mobility and independence of people with lower-limb paralysis. We have created ABLE, the first lightweight, easy-to-use and affordable exoskeleton that allows walking again after a spinal cord injury. It helps palliating health complications while boosting self-confidence and independence.

DBGEN Ocular Genomics

2018 · <https://dbgen.com/>

Co-funders: Dr. Roser González-Duarte and Dr. Gemma Marfany (IRSJD)

DBGen is a company focused on the genetic diagnosis of hereditary eye disorders, providing the highest level of quality, diagnostic effectiveness and close contact with patients, ophthalmologists, and enterprises in need of its services. At DBGEn, we offer high-quality, reliable and accessible genetic diagnosis, allowing the clinical diagnosis to be assured and guidance to be provided to the patient with respect to emerging therapies for genetic disorders of the eyes. Genetic diagnosis is indispensable to identify family members possessing a genetic defect, permitting the most suitable advice to be given in each case.

CEBIOTEX

2012 · <http://www.cebiotex.com/>

Dr. Joan Bertran Llavina (CEO) & Dr. Angel Montero (IRSJD)

Cebiotex has developed and characterised at preclinical level, in vitro and in vivo, nonwoven nanofiber membranes for the local release

of active ingredients in the local treatment of the surgical site after tumour excision surgery. The purpose of the local release of drugs is to achieve high concentrations of active ingredients in the area where the release device is placed, avoiding systemic toxicity and increasing the effectiveness of the drug against possible tumoral remnants. The activity of Cebiotex is based on the interaction of researchers from two Catalan institutions: the Polytechnic University of Catalonia (UPC) and Sant Joan de Déu Barcelona Children's Hospital (HSJD).

BCN Innova

2008 · <http://www.bcninnova.com/>

Dr. Joan Prat (IRSJD)

BCN Innova, a spin-off company of Sant Joan de Déu-Barcelona Children's Hospital, has developed a device (GazeLab) that substantially improves the diagnosis and treatment of strabismus, reducing the number of second operations that are now performed to achieve the expected results. GazeLab offers two main benefits: 1. It allows objective exploration on a revolutionary level of precision, with much better results than the subjective-measurement model that has prevailed until now. 2. It provides a 50% reduction in the number of second operations, obviating those which are surgically unnecessary.

Doctoral Theses

Thesis author	Title	University	Thesis directors	Month of lecture	Research group
Claudia García Mintegui	Bioresorbable Zn-based alloys for biomedical applications	Universidad Politècnica de Catalunya	Marta Pegueroles Neyra & José Luís Cortina Pallás (Universitat Politècnica de Catalunya)	January	Medical technologies: Biomaterials and Tissue Engineering
Carmina Guitart Pardellans	Millora en el diagnòstic i maneig de la pneumònia en el pacient pediàtric crític, mitjançant l'ús de l'ecografia pulmonar i la procalcitonina	Universidad de Barcelona	Mónica Balaguer Gargallo	February	Immune and Respiratory Dysfunction in Critically Ill Children
Carlota Saumell Andreu	Developmental Intergroup Theory of Mind: The protagonist's social group membership as a pragmatic performance factor	Universidad de Barcelona	Ferran Pons Gimeno & Mireia Hernández Pardo (Universitat de Barcelona)	February	Attention, Perception and Language Acquisition (APAL)
Charlotte Martine Juton	Child behavior, feeding practices and health in the context of obesity	Universidad de Barcelona	Carles Lerin Martínez & Helmut Schröder (IMIM)	March	Metabolic diseases of pediatric origin
Paulo Cesar Sarango Granda	Desarrollo de formulaciones de Apremilast para el tratamiento de patologías en piel y mucosas	Universidad de Barcelona	Josefa Badia Palacin, Ana Cristina Calpena Capmany (Universitat de Barcelona) & Lupe Carolina Espinoza Tituana (Universitat de Barcelona)	March	Gut Microbiota
Martina Finocchiaro	Automatic hands-free visualization of a six degrees of freedom agent within a complex anatomical space	Universidad Politècnica de Catalunya	Alicia Casals Gelpi, Albert Hernansanz Prats, Arianna Menciassi (Sant'Anna School of Advanced Studies) & Gastone Ciuti (Sant'Anna School of Advanced Studies)	March	Medical technologies: Robotics and vision
Elia Vila Andreu	Altered molecular pathways in schizophrenia: modulation by therapeutic strategies	Universidad de Barcelona	Belen Ramos Jose Maria	May	Etiopathogenesis and treatment of severe mental disorders (MERITT)
Montserrat Meseguer Meda	Estudi de factors clínics i biològics associats a toxicitat en el tractament de la leucèmia limfoblàstica aguda pediàtrica.	Universidad de Barcelona	Susana Rives Solà & Mireia Camós Guijosa	May	Leukaemia and Paediatric Haematology Disorders

Doctoral Theses

Thesis author	Title	University	Thesis directors	Month of lecture	Research group
Aleix Soler García	Utilitat del QuantiFERON-TB Gold Plus en el diagnòstic de la infecció tuberculosa latent i la malaltia tuberculosa en població pediàtrica	Universitat de Barcelona	Antoni Noguera Julián & Anna Gamell Fullà	May	Infectious diseases and microbiome
Ana Pilar García García	Infecció por SARS-CoV-2 en nens i adults joves amb errors congènits de la immunitat: expressió clínica i resposta immunitària	Universitat de Barcelona	Laia Alsina Manrique de Lara & Àngela Deyà Martínez	June	Cardiovascular Diseases and Child Development / Study of Immune Deficiency Diseases in Paediatrics (GEMDIP)
Georgina Company Se	Temporal and frequency differentiation of healthy and pathological lung tissue through minimally invasive electrical impedance spectroscopy	Universitat Politècnica de Catalunya	Lexa Nescolarde Selva & Ramon Bragós Badia	July	Medical technologies: Instrumentation and eHealth
Xavier Solé Martí	Effects of cold atmospheric plasmas on biomaterials: hydrogels and composites with calcium phosphates	Universitat Politècnica de Catalunya	Cristina Canal Barnils & Maria Pau Ginebra Molins (Universitat Politècnica de Catalunya)	July	Medical technologies: Biomaterials and Tissue Engineering
Ainhoa Pascual Alonso	Study of expression levels in MECP2 related disorders using transcriptomics and proteomics: characterizing Rett syndrome and MECP2 duplication syndrome.	Universitat de Barcelona	Judith Silvia Armstrong Morón	July	Genomics for the diagnosis of rare diseases
Antonio Rodríguez Fernández	Clinical evaluation towards the development of a lower limb exoskeleton for people with spinal cord injury: from gait biomechanics to motor learning	Universitat Politècnica de Catalunya	Josep Maria Font Llagunes & Joan Lobo Prat	July	Medical Technologies: Biomechanics
Ameya Ravindra Pore	Surgical subtask automation for intraluminal procedures using deep reinforcement learning	Universitat Politècnica de Catalunya	Alicia Casals Gelpi & Paolo Fiorini (Kyowa Kirin International plc)	July	Medical technologies: Robotics and vision

Doctoral Theses

Thesis author	Title	University	Thesis directors	Month of lecture	Research group
Daniel Penela Sánchez	Estudio del papel de la microbiota nasofaríngea en la gravedad de la infección por Rino/Enterovirus humano en niños menores de 5 años	Universidad de Barcelona	Carmen Muñoz Almagro & Cristian Launes Montaña	September	Infectious diseases and microbiome / Immune and Respiratory Dysfunction in Critically Ill Children
Marc Josep Cahuana Bartra	Exposició al Diòxid de Nitrògen durant el embaràs i la seva afectació materno-fetal	Universidad de Barcelona	Maria Dolores Gómez Roig & Payam Dadvan (ISGlobal)	September	Environment and complications during pregnancy
Marien Cristina Narvaez Dorado	Crutch gait patterns characterization through spatial and temporal parameters	Universidad Politècnica de Catalunya	Joan Aranda López	September	Medical technologies: Robotics and vision
Moisés Castellà Giner	Involvement of Alternative Splicing Regulation in Adipose Tissue Thermogenic Induction	Universidad de Barcelona	Francesc Villarroya Gombau & Rubén Cereijo Téllez	September	Adipose Tissue Molecular Biology and Gene Regulation and Associated Disorders
Cristina Borràs Novell	Composició macromolecular de la llet materna, influència de factors materns, gestacionals i neonatals i impacte en el creixement del nou-nat prematur	Universidad de Barcelona	Isabel Iglesias Platas & Ana Herranz Barbero	October	Neonatology Research Group (GRen)
Belén González Gómez	Attentional selectivity and attentional control in highly math-anxious individuals	Universidad de Barcelona	Maria Isabel Núñez Peña & Àngels Colomé (Universitat de Barcelona)	October	Cognitive neuroscience
Maria Farahi	US signal and image monitoring and diagnosis using computer vision and deep learning	Universidad Politècnica de Catalunya	Alicia Casals Gelpi & Joan Aranda López	November	Medical technologies: Robotics and vision
Jesús de Miguel Fernández	Control strategies for exoskeleton gait training after stroke: understanding the importance of parameter tuning	Universidad Politècnica de Catalunya	Josep Maria Font Llagunes & Joan Lobo Prat	November	Medical Technologies: Biomechanics

Doctoral Theses

Thesis author	Title	University	Thesis directors	Month of lecture	Research group
Montse Blasco Sierra	Towards the Optimisation of Interventions to Enhance Cognitive Functions and Quality of Life in Children with Cerebral Palsy	Universidad de Barcelona	Roser Pueyo Benito	December	Neuropsychology
Elena Esperanza Cebollada	Estudi de nous biomarcadors en pacients amb leucèmia pediàtrica d'alt risc	Universidad de Barcelona	Mireia Camós Guijosa & Montserrat Torreadell Burriel	December	Leukaemia and Paediatric Haematology Disorders / Paediatric Cancer Treatment
Sánchez L.	Relevant deubiquitinating enzymes in cilium formation and function in the retina: USP48	Universidad de Barcelona	Gemma Marfany Nadal	December	Human Molecular Genetics II
Andrea Valentina Prieto Carruyo	Estudio de la interacción de hCNT3 y QDPR como nexo funcional entre la recuperación de nucleósidos y sus vías biosintéticas	Universidad de Barcelona	Marçal Pastor-Anglada & Sandra Perez-Torras	December	Molecular Pharmacology and Experimental Therapeutics (MPET)
Clara Xiol Viñas	Comprehensive analysis of diagnostic approaches and molecular landscape of Rett syndrome spectrum disorders	Universidad de Barcelona	Judith Silvia Armstrong Morón & Alfonso Luis De Oyarzabal Sanz	December	Genomics for the diagnosis of rare diseases / Paediatric Neurometabolic Disorders: Neural Communication Mechanisms and Personalised Therapies
Xavier Marimon Serra	Caracterització multiescala de dades fisiològiques en la cardiologia translacional	Universidad Politécnica de Catalunya	Raul Benítez Iglesias	December	Medical Technologies: Computer Graphics: Serious Games and Gamification



5

CORE FACILITIES



Core facilities

The Institut de Recerca Sant Joan de Déu makes available to its researchers a set of fundamental services and infrastructures for the performance of their projects. In order to facilitate high-level research, the IRJSD offers its professionals technical, scientific and support services as well as the most suitable infrastructures required to carry out their work:

Scientific core facilities

- Biological Sample Processing and Molecular Analysis
- Confocal Microscopy and Cellular Imaging Unit
- Clinical Trials Unit
- Library
- Methodology and statistics
- Neuroimaging
- Neurological tissues Biobank for research
- Neuropsychological evaluations
- Paediatric Biobank for research
- Patient Engagement in Research Area
- Sequencing Unit
- Animal housing facility
- Unit of Design, Analysis and Interpretation of Omic data (DAIOmics)

Management support services

- Administration and Finance
- People and Values
- Research Promotion and Management
- Knowledge Management and Quality
- Communication
- Clinical Research Ethics Committee





6

TRAINING, COMMUNICATION AND PARTICIPATION

Seminars
Training programme
Communication and public engagement
News



Seminars

14 SEMINARS

Relation of research seminars and IRSJD day of debates

24/1/23

Desenvolupament d'un tractament protector per malalties del neurodesenvolupament

Alfonso de Oyarzabal and Rosanna Mari, IRSJD

13/2/23

ISOLATE: Segmentatlon and claSsification Of vascuLar pATtern symmEtries on cerebral vessels using DL

Gilberto Ochoa, Tecnológico de Monterrey

14/2/23

Malalties del metabolisme de la lisina. Una perspectiva des de la teràpia gènica

Cristina Fillat, IDIBAPS

14/3/23

Patch clamp technique unveils channelopathies pathophysiology and defines personalised therapies

Xavier Altafaj Tardío, Universitat de Barcelona

20/3/23

The impact of breastfeeding duration on gray matter volume: an example of very large sample size neuroimaging

Christian Stephan-Otto, IRSJD

17/4/23

Valoració de la reabsorció òssia fisiològica de l'empelt alveolar òssia secundari mitjançant TAC de baixa dosi en pacients amb fissura labiopalatina unilateral

Marta Gómez Chiari and Martí Engli Rueda, IRSJD

8/5/24

Evaluation of deep learning (DL) algorithms using visual explanation techniques to classify brain Arteriovenous Malformations (AVM) in Arterial Spin Labeling sequences (ASL)

Júlia Romagosa Pérez, IRSJD

23/5/23

El peix zebra com a model per estudiar les GRINpatíes

Sílvia Locubiche Serra, ZeClinics

6/6/23

Models per entendre els trastorns del neurodesenvolupament en el Síndrome de Lamb-Shaffer

Aixa V. Morales García, CSIC

12/6/23

Unitat 3D4H: De la imatge mèdica a impressió 3D i Realitat Virtual

Núria Adell Gómez, IRSJD

19/9/23

Presynapse (dys)function in the Human Central Nervous System: from the Neurotransmitter Release Machinery to Microcircuits

Francisco José López-Murcia, Universitat de Barcelona

24/10/23

Controlling neuronal activity with light: from individual cells to brain waves

Pau Gorostiza, IBEC

7/11/23

Glial-specific regulation of endocytosis controls neuronal function and survival

Tania López-Hernández, Universitat de Barcelona

12/12/23

Role of neural extracellular vesicles in transneuronal communication in health and disease

Júlia Solana and Cristina Malagelada, Universitat de Barcelona

Training programme

23**COURSES IN 2023****395****ATTENDEES IN 2023**

To promote the development of the different competence dimensions, four types of training modalities are established:

- Knowledge and techniques
- Soft Skills
- Organization and Governance
- Institutional training (Values)

Courses**January**

Oportunitats de finançament competitiu

January

Lideratge i Gestió d'equips per Comandaments

March

Becoming a Scientific Writer

March

REDCAP

April

Bones pràctiques en investigació

May

Microscopia Confocal

May

Cómo publicar los resultados de la investigación: impacto científico y social

May

Design Thinking

May

Biobanc

May

Metodologia per projectes de recerca

June

Estadística avançada i analisi de dades

June

Gestores bibliográficos: Mendeley

July

Lean mail. Transforma l'outlook en el teu panell de control

September

Excel: Nivel intermedio

September

Oral Presentations: Beyond Slides

October

Excel: Nivel avanzado

October

Taller de comunicació interpersonal (Bridge)

November

Open Science y Propiedad intelectual

November

Build Mentor-mentee relationships

December

"R", programació bàsica per a estadística

Communication and public engagement

Activities

17
ACTIVITIES ORGANIZED

65
RESEARCHERS

1865
AUDIENCE REACHED

Participation in:

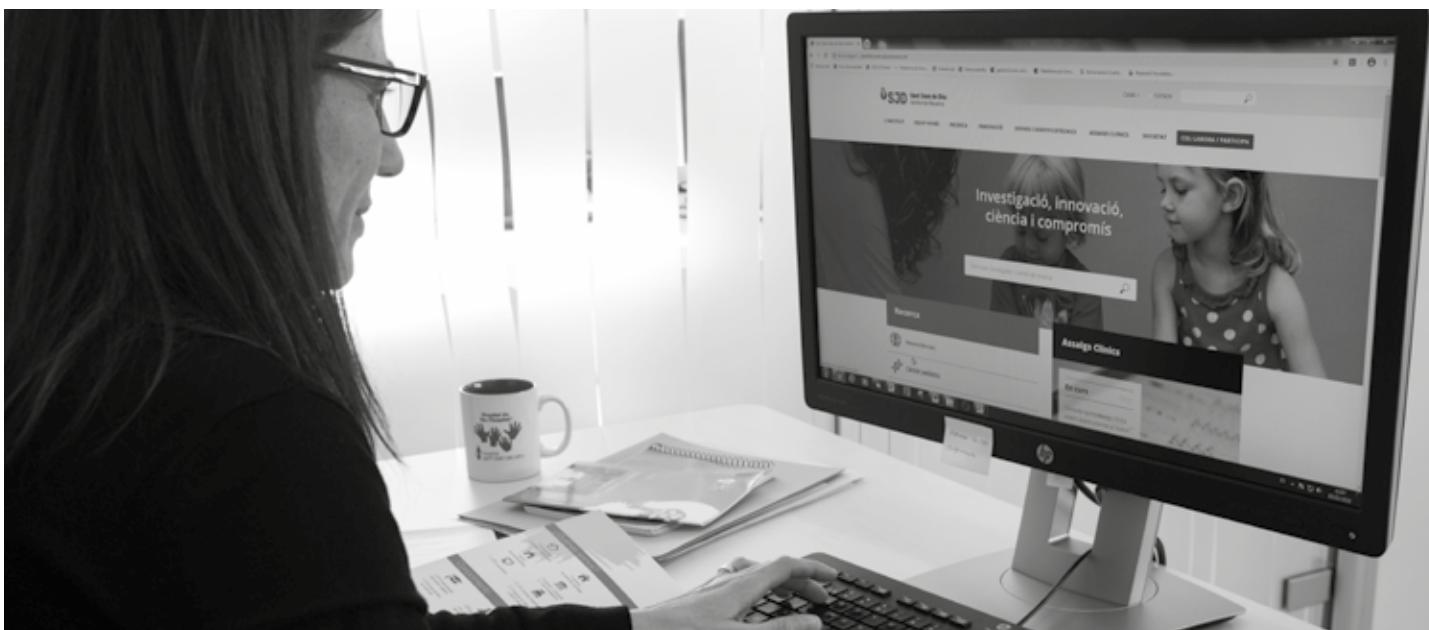
- International Day of Woman and Girl in Science
- Brain Week
- International Day of Clinical Trials
- Festa de la Ciència de Barcelona
- Science week
- European Resercher's Night
- Pint of Science
- Agora de la Ciència
- Escolab
- Visita la recerca de La Marató

Internet and Social media

LINKEDIN
22253 FOLLOWERS

TWITTER
1893 FOLLOWERS

WEBSITE
57333 USERS
188000 NUMBER OF VISITS TO PAGES



News



Pro-inflammatory alterations identified in persons with non-specific low back pain



Researchers at IRSJD develop a method to predict the efficacy of a potent nanomedicine in a severe childhood cancer



An early diagnosis and some diet changes can decrease the risk of severe symptoms in children with BCKDK metabolic deficiency

News



IRSJD researchers have discovered an intergenerational inheritance mechanism in mouse models of childhood obesity



Benefits of listening to music during pregnancy on baby's brain capacity for language sound encoding



Longer breastfeeding duration leads to larger gray matter volume at age 10 years

News



Metformin, key drug in the battle against type 2 Diabetes, reveals its secrets in a new study



Mutation in DOCK11 gene explains rare disorder in immune system regulation

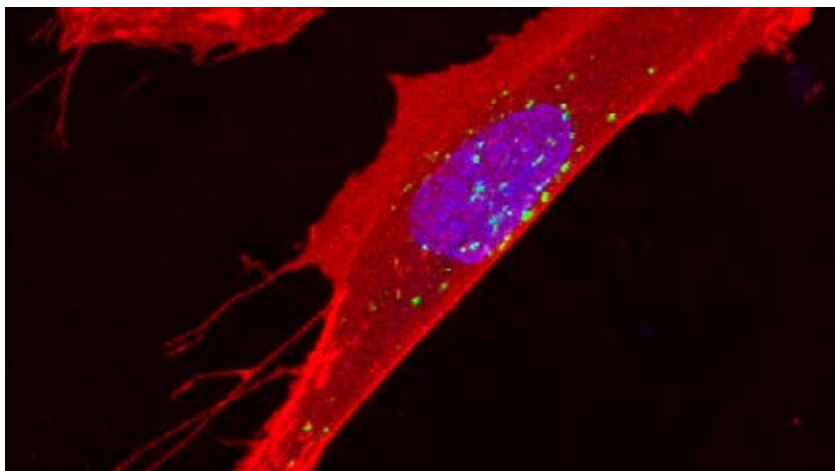


A study has revealed an association between mental disorders and premature mortality in the Catalan population

News



Following a Mediterranean diet reduces stress and anxiety during pregnancy



Characterization of Extracellular Vesicles in Fibroblasts of Collagen-VI Related Muscular Dystrophy



7

SCIENTIFIC ACTIVITY



- 7.1. Medicine and pediatric science area
- 7.2. Mental health area
- 7.2. Mental health area
- 7.3. Transversal program





7.1

MEDICINE AND PEDIATRIC SCIENCE AREA

7.1.1 Neuroscience

Applied Research in Neuromuscular Diseases
Paediatric Neurometabolic Disorders: Neural Communication
Mechanisms and Personalised Therapies
Neonatal Brain
Neuropsychology
Cognitive Neuroscience
Attention, Perception and Language Acquisition (APAL)
Research in Child and Adolescent Mental Health
Epilepsy and Movement Disorders in Pediatrics



Applied Research in Neuromuscular Diseases

Cecilia Jiménez Malebrera, PhD



23
PROJECTS
11 competitive projects
12 non competitive projects



38
CLINICAL TRIALS &
OBSERVATIONAL STUDIES
36 commercial clinical trials
2 academic clinical trials



31
PUBLICATIONS



211
IMPACT FACTOR
6.8 average impact factor



52%
PUBLICATIONS IN 1ST QUARTILE



13%
PUBLICATIONS IN 1ST DECILE



26%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Julita Medina, Irene Beatriz Zschaeck Luzardo, Andrés Nascimento Osorio, Carlos Ignacio Ortez Gonzalez

POSTDOC ASSOCIATED RESEARCHERS

Aristides Lopez Marquez, Selena Trifunov, Daniel Natera de Benito

PHD STUDENTS

Cristina Jou Muñoz

ASSISTANT RESEARCHER

Cristina Puig Ram, Sonia Segovia Simon, Jessica Maria Exposito Escudero, Laura Carrera García, Maria del Carmen Badosa Gallego

TECHNICIAN

Ruth Lavilla Hidalgo, Anna Codina Bergadà

During 2023 we have witnessed important advances in the field of neuromuscular disorders with regards to the development and implementation of novel or recently approved therapies and our group has contributed to those international

efforts at different levels. On one hand the Neuromuscular Unit has participated in more than 10 international multicentric clinical trials for several forms of muscle diseases, mainly Duchenne Muscular Dystrophy, Spinal Muscular Atrophy and Pompe's disease. To highlight our participation in the first clinical trial to apply gene therapy to children with Duchenne Muscular Dystrophy (DMD) under 4 years which until recently was only approved for older children. Also, in DMD we took part in a clinical study to assess the use of an immunomodulator in children with pre-existing immunity to the components of the gene therapy. Treatments for Spinal Muscular Atrophy continue to evolve, and our team participated in two different trials using anti-myostatin inhibitors.

In parallel to this our group is at the forefront of the development of nucleic acid-based therapies for some of the rarer muscle diseases such as Collagen VI related dystrophies. In 2023 we have received private and public funding to progress in this area, not only to conduct the necessary experimental work but also, and thanks to competitive funding to work on the technology transfer aspects of this project. We have been awarded a Llabor grant from Industria del Coneixement and the PERIS program (both from Generalitat de Catalunya) which recognize high impact innovative projects, and which will provide us with training from experts and resources to help us take these potential therapies to patients and to the market. To deliver these drugs to skeletal muscle we are assessing both nanoparticles and viral vectors in collaboration with various national and international experts including collaborators at the Instituto de Ciencias Materiales de Barcelona (ICAMB) and at Genethon-Inserm (Paris).

Pre-clinical research in our laboratory this year has been focused on the deep characterization of a novel mouse model for COL-RD and of more physiological cellular models such as decellularized matrices and 3D models in collaboration with the Institute of Bioengineering of Catalunya (IBEC).

In two collaborative studies published in the first half of 2023 we report

patients with muscular dystrophy and/or high CK with variants in the DTNA gene that encodes α -dystrobrevin, not previously associated with muscle disease and a founder mutation in the TRAPPC11 gene in Roma individuals. Later in the year we have published a study where we demonstrate that extracellular vesicles are key factors in the progression of COL6-RD modulating cell functions such as migration.

Throughout this year we have presented our research at numerous international meetings such as the World Muscle Society, the EVELAM Latinamerican school of Myologie and the prestigious CRISPR/Medicine News webinars but also reaching the wider public via various platforms ("La Ciencia de lo Singular").

SELECTED PUBLICATIONS

- Badosa C, Roldán M, Fernández-Irigoyen J, Santamaria E, Jimenez-Mallebrera C. Proteomic and functional characterisation of extracellular vesicles from collagen VI deficient human fibroblasts reveals a role in cell motility. *Sci Rep.* 2023 Sep 5;13(1):14622. FI:4.6 (Q2)
- Henzi BC, Schmidt S, Nagy S, Rubino-Nacht D, Schaedelin S, Putanackal N, Stimpson G; North Star Consortium; Amthor H, Childs AM, Deconinck N, de Groot I, Horrocks I, Houwen-van Opstal S, Laugel V, Lopez Lobato M, Madruga Garrido M, Nascimento Osorio A, Schara-Schmidt U, Spinty S, von Moers A, Lawrence F, Hafner P, Dorchies OM, Fischer D. Safety and efficacy of tamoxifen in boys with Duchenne muscular dystrophy (TAMDMD): a multicentre, randomised, double-blind, placebo-controlled, phase 3 trial. *Lancet Neurol.* 2023 Oct;22(10):890-899. FI:48 (D1)
- Nascimento A, Bruels CC, Donkervoort S, Foley AR, Codina A, Milisenda JC, Estrella EA, Li C, Pijuan J, Draper I, Hu Y, Stafki SA, Pais LS, Ganesh VS, O'Donnell-Luria A, Syeda SB, Carrera-García L, Expósito-Escudero J, Yubero D, Martorell L, Pinal-Fernandez I, Lidov HGW, Mammen AL, Grau-Junyent JM, Ortez C, Palau F, Ghosh PS, Darras BT, Jou C, Kunkel LM, Hoenicka J, Bönnemann CG, Kang

PB, Natera-de Benito D. Variants in DTNA cause a mild, dominantly inherited muscular dystrophy. *Acta Neuropathol.* 2023 Apr;145(4):479-496. FI:12.7 (D1)

- Goyenvalle A, Jimenez-Mallebrera C, van Roon W, Sewing S, Krieg AM, Arechavala-Gomez V, Andersson P. Considerations in the Preclinical Assessment of the Safety of Antisense Oligonucleotides. *Nucleic Acid Ther.* 2023 Jan;33(1):1-16. FI:4 (Q2)

SELECTED PROJECTS

- Cerrando la brecha terapéutica para Distrofias Musculares Congénitas. Estrategias para mejorar la eficacia, liberación y seguridad de terapias basadas en CRISPR/Cas9 y oligonucleótidos. (PI22/01382). Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD; Instituto de Salud Carlos III (ISCIII); Jiménez. 2023-2025. IP: Cecilia Jiménez Mallebrera.
- CuidAME: Longitudinal Data Collection from Patients with Spinal Muscular Atrophy in Spain in a nationwide Registry. Biogen International GmbH. 2019-2025. IP: Andrés Nascimento Osorio.
- Medicina Personalizada para las Distrofias Musculares Congénitas: Desarrollo de terapias avanzadas, modelos fisiológicos y herramientas diagnósticas precisas. (PI19/00122). Instituto de Salud Carlos III (ISCIII). 2020-2023. IP: Cecilia Jiménez Mallebrera.
- MyoQ: La Plataforma de Nanomedicina para terapias avanzadas contra las Distrofias Musculares Congénitas. (SLT033/23/000032). Generalitat de Catalunya. 2023-2024. IP: Cecilia Jiménez Mallebrera.

SELECTED CLINICAL STUDIES

- Estudio abierto, de dos partes, de administración génica sistémica, para evaluar la seguridad y la expresión de ro7494222 (srp9001) en sujetos menores de cuatro años con distrofia muscular de duchenne. A two-part, open-label systemic gene delivery study to evaluate the safety and expression of ro7494222 (srp-9001) in subjects under the age of four with duchenne muscular dystrophy. (BN43881). Anatomía patológica; Cetir Centre

Diagnòstic Pedralbes S.A; Farmacia; Hospitalización HSJD; Laboratori HSJD; Parexel International (IRL) Ltd.; Roche Diagnostics International Ltd; Servicio de Cardiología; Servicio Rehabilitación; SIMM Molecular S.L.; Unitat de Recerca (URC) FSJD; Vidal. 2022 . IP: Andrés Nascimento Osorio.

- A Randomized, Double-Blind, Placebo-Controlled, Study to Evaluate the Efficacy and Safety of Taldefgrobep Alfa in Ambulatory and Non-Ambulatory Participants with Spinal Muscular Atrophy with Open-Label Extension (RESILIENT). Estudio aleatorizado, doble ciego, controlado con placebo para evaluar la eficacia y la seguridad de taldefgrobep alfa en participantes ambulatorios y no ambulatorios con atrofia muscular espinal con extensión abierta (RESILIENT). (BHV2000-301). Biohaven Pharmaceuticals, Inc; Daimiel; Farmacia; Hospital de día URC; Laboratori HSJD; Roura; Servicio de Cardiología; Servicio Rehabilitación; SIA DOKUMEDS; SIMM Molecular S.L.; Unitat de Recerca (URC) FSJD. 2023. IP: Andrés Nascimento Osorio.
- An Open-Label, Systemic Gene Delivery Study to Evaluate the Safety, Tolerability and Expression of SRP-9001 in Association with Imlifidase in Subjects with Duchenne Muscular Dystrophy with Pre-existing Antibodies to rAAVrh74. (SRP-9001-104). Anatomía patológica; Diagnóstico para la imagen (DXI); Hospitalización HSJD; Laboratori HSJD; Parexel International S.L; Sarepta Therapeutics, Inc.; Servicio de Cardiología; Servicio Rehabilitación; Unitat de Recerca (URC) FSJD; Vallés. 2023. IP: Andrés Nascimento Osorio.
- EFC14462-Baby COMET study: An Open-label, Multinational, Multicenter, Intravenous Infusion Study of the Efficacy, Safety, Pharmacokinetics, and Pharmacodynamics of Avalglucosidase Alfa in Treatment-naïve Pediatric Participants with Infantile-Onset Pompe Disease (IOPD)./ Estudio abierto, multinacional, multicéntrico y de infusión intravenosa sobre la eficacia, seguridad, farmacocinética y farmacodinámica de la avalglucosidasa alfa en

participantes pediátricos sin tratamiento previo con enfermedad de Pompe de inicio en la infancia (EPII)". (EFC14462-Baby COMET). Anglès; Farmacia; Laboratori HSJD; Prats; Sanofi-Aventis recherche & Developpement; Servicio de Cardiología; Servicio Rehabilitación; Unitat de Recerca (URC) FSJD. 2023. IP: Andrés Nascimento Osorio.

Paediatric Neurometabolic Disorders: Neural Communication Mechanisms and Personalised Therapies

M^a Angels García Cazoral, MD, PhD



21
PROJECTS
12 competitive projects
9 non competitive projects



22
CLINICAL TRIALS &
OBSERVATIONAL STUDIES
22 commercial clinical trials
0 academic clinical trials



26
PUBLICATIONS



165
IMPACT FACTOR
6.3 average impact factor



77%
PUBLICATIONS IN 1ST QUARTILE



23%
PUBLICATIONS IN 1ST DECILE



23%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Roser Colomé Roura, Silvia Maria Meavilla Olivas, Alejandra Darling, Jordi Muchart López, M^adel Mar O'Callaghan Gordo, Federico Ramos

POSTDOC ASSOCIATED RESEARCHERS

Alfonso Luis De Oyarzabal Sanz

PHD STUDENTS

Juliana Ribeiro Constante, Uliana Musokhranova Matveeva, Natalia Alexandra Julià Palacios

ASSISTANT RESEARCHER

Júlia Romagosa I Pérez, Jennifer Anticona Huaynate, Leticia Diana Pias Peleteiro, Ariadna Borrás Martínez, Rosanna Marí Vico, Silvia Constanca Liendo Vallejo, Mariya Sigatullina Bondarenko

TECHNICIAN

Anna Serradell Salís, Cristina Grau Páez

SELECTED PUBLICATIONS

- Tangeraas T, Constante JR, Backe PH, Oyarzábal A, Neugebauer J, Weinhold N, Boemer F, Debray FG, Ozturk-Hism B, Evren G, Tuba EF, Ummuhan O, Footitt E, Davison J, Martinez C, Bueno C, Machado I, Rodríguez-Pombo P, Al-Sannaa N, De Los Santos M, López JM, Ozturkmen-Akay H, Karaca M, Tekin M, Pajares S, Ormazabal A, Stoway SD, Artuch R, Dixon M, Mørkrid L, García-Cazorla A. BCKDK deficiency: a treatable neurodevelopmental disease amenable to newborn screening. *Brain*. 2023 Jul 3;146(7):3003-3013. FI: 14.5 [D1]
- Tristán-Noguero A, Fernández-Carasa I, Calatayud C, Bermejo-Casadesús C, Pons-Espinal M, Colini Baldeschi A, Campa L, Artigas F, Bortolozzi A, Domingo-Jiménez R, Ibáñez S, Pineda M, Artuch R, Raya Á, García-Cazorla À, Consiglio A. iPSC-based modeling of THD recapitulates disease phenotypes and reveals neuronal malformation. *EMBO Mol Med*. 2023 Mar 8;15(3):e15847. FI: 11.1 [D1]
- Musokhranova U, Grau C, Vergara C, Rodríguez-Pascau L, Xiol C, Castells AA, Alcántara S, Rodríguez-Pombo P, Pizcueta P, Martinell M, García-Cazorla A, Oyarzábal A. Mitochondrial modulation with leriglitazone as a potential treatment for Rett syndrome. *J Transl Med*. 2023 Oct 26;21(1):756. FI: 7.4 [Q1]
- Stanescu S, Correcher Medina P, Del Castillo FJ, Alonso Luengo O, Arto Millan LM, Belanger Quintana A, Camprodon Gomez M, Diez Langhetée L, Garcia Campos O, Matas Garcia A, Perez-Moreno J, Rubio Gribble B, Visa-Reñé N, Giraldo-Castellano P, O'Callaghan Gordo M. Clinical Outcomes of Patients with Chronic Neuropathic Form of Gaucher Disease in the Spanish Real-World Setting: A Retrospective Study. *Biomedicines*. 2023 Oct 22;11(10):2861. FI: 4.7 [Q1]

SELECTED PROJECTS

- Brain Metabolism: from rare diseases to common biological pathways. (00201794). European Commission. 2023-2023. IP: M^aAngels García Cazoral.
- Entrega de MeCP2 en el cerebro mediante el uso de nanocápsulas para el tratamiento del síndrome

de Rett. (SLT033/23/000097). Generalitat de Catalunya. 2023-2024. IP: Alfonso Luis De Oyarzabal Sanz.

- Development of leriglitazone for Rett Syndrome and related disorders. (CPP2021-008554). Ministerio De Ciencia E Innovacion. 2022-2025. IP: M^aAngels García Cazorla, Alfonso Luis De Oyarzabal Sanz.
- Sinapsis en un ch. IP: medicina personalizada en trastornos genéticos del neurodesarrollo que afectan a la comunicación sináptica. (PI21/00073). Instituto de Salud Carlos III (ISCIII). 2022-2024. IP: M^aAngels García Cazorla.

SELECTED CLINICAL STUDIES

- A Global, Phase 1/2, Open-Label, Dose Optimization Study to Evaluate the Safety, Tolerability, Pharmacodynamics, and Pharmacokinetics of mRNA-3705 in Participants with Isolated Methylmalonic Acidemia Due to Methylmalonyl-CoA Mutase Deficiency. (mRNA-3705-P101). Cañizares; Farmacia; Hospitalización HSJD; Laboratori HSJD; Lancho; ModernaTX, Inc.; PPD Global Limited; Servicio de Cardiología; Servicio de Nutrición; Servicio Neuropsicología; Unitat de Recerca (URC) FSJD. 2023. IP: M^aAngels García Cazorla.
- Estudio abierto de fase III del tratamiento adyuvante con ganaxolona (GNX) en niños y adultos con epilepsia relacionada con el complejo de esclerosis tuberosa (CET) (TrustTSC OLE). (1042-TSC-3002). Casanova; Farmacia; Laboratori HSJD; Marinus Pharmaceuticals; Servicio de Cardiología; Unitat de Recerca (URC) FSJD. 2023. IP: Federico Ramos.
- Estudio multicéntrico, aleatorizado y doble ciego para determinar el efecto de la triheptanoína en comparación con triglicéridos de cadena media (TCM) sobre los episodios clínicos importantes (ECI) en pacientes pediátricos con trastornos de la oxidación de ácidos grasos de cadena larga (LC-FAOD). (UX007-CL302). Farmacia; Laboratori HSJD; Rodriguez; Servicio de Cardiología; Servicio de Nutrición; Ultragenyx Pharmaceutical, Inc.; Unitat de Recerca (URC) FSJD. 2022. IP: M^aAngels García Cazorla.

Neonatal Brain

**Thais Agut Quijano, MD, PhD
& Ana Alarcon Allen, MD, PhD**



5
PROJECTS
5 competitive projects
0 non competitive projects



0
CLINICAL TRIALS &
OBSERVATIONAL STUDIES



10
PUBLICATIONS



202
IMPACT FACTOR
20.2 average impact factor



80%
PUBLICATIONS IN 1ST QUARTILE



50%
PUBLICATIONS IN 1ST DECILE



20%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Sílvia Planas Román, Nuria Herranz Rubia, Maria del Mar Velilla Aparicio

POSTDOC ASSOCIATED RESEARCHERS

Nuria Carreras Blesa

TECHNICIAN

Nerea Liñan Rosa

During 2023, the Neonatal Brain Group continued its main research areas: neonatal hypoxic-ischemic injury, preterm brain maturation, fetal/neonatal infections affecting the developing brain, and care practices for at-risk newborns and their families. We continued to collaborate with other researchers of the IRSJD and other institutions. Thus, our projects have been nourished by multiple approaches (clinical -medical and nursing-, basic science, biomedical engineering, epidemiology), all of them favoring their translational nature. We continued to participate in Working Groups in the field of our research, both national and European, with a

global research and teaching vocation. Our team continued to be awarded a number of competitive and private grants to support our research.

One of the main active projects of the group is the assessment of maturation and serial brain biometries in preterm newborns by imaging, with the aim of detecting anomalies or deviations that alert about possible pathology or deviation of the maturational trajectory. In the same cohort of preterm newborns, researchers of the group are studying the usefulness of both qualitative and quantitative (automated) electroencephalogram parameters in the study of brain maturation/dysmaturation.

Within the line of neonatal infections, the pilot program for universal screening of congenital cytomegalovirus infection was completed. We continued to collaborate with national and international experts with the goal to reduce the burden of this congenital infection, which is the most prevalent and poses a significant risk of long-term complications in affected children. In the line of translational research, the group collaborated with public organizations to implement prevention strategies for fetal cytomegalovirus infection and its sequelae, alongside international efforts to standardize evidence-based management guidelines.

The group continues to support nursing research on aspects of neonatal care that impact the developing brain. Finally, the group has continued to promote predoctoral research, with four doctoral theses in progress.

SELECTED PUBLICATIONS

- Alarcón A, de Vries LS, Parodi A, Arnáez J, Cabañas F, Steggerda SJ, Rebollo M, Ramenghi L, Dorronsoro I, López-Azorín M, Schneider J, Noguera-Julian A, Ríos-Barnés M, Recio M, Bickle-Graz M, Martínez-Biarge M, Fortuny C, García-Alix A, Truttmann AC. Neuroimaging in infants with congenital cytomegalovirus infection and its correlation with outcome: emphasis on white matter abnormalities. Arch Dis Child Fetal Neonatal Ed. 2024 Feb 19;109(2):151-158. FI:4.4 (D1)
- García-Alix A, Arnaez J, Herranz-

Rubia N, Alarcón A, Arca G, Valverde E, Blanco D, Lubian S; Grupo Cerebro Neonatal. Ten years since the introduction of therapeutic hypothermia in neonates with perinatal hypoxic-ischaemic encephalopathy in Spain. Neurologia (Engl Ed). 2020 Sep 25:S0213-4853(20)30227-9. English, Spanish. FI:3.9 (Q3)

- Núñez C, García-Alix A, Arca G, Agut T, Carreras N, Portella MJ, Stephan-Otto C. Breastfeeding duration is associated with larger cortical gray matter volumes in children from the ABCD study. J Child Psychol Psychiatry. 2023 Jul;64(7):1067-1079. FI:7.6 (D1)
- Carreras N, Arnaez J, Valls A, Agut T, Sierra C, Garcia-Alix A. CSF neopterin and beta-2-microglobulin as inflammation biomarkers in newborns with hypoxic-ischemic encephalopathy. Pediatr Res. 2023 Apr;93(5):1328-1335. FI:3.6 (Q1)

SELECTED PROJECTS

- Estudio CRIB (Cribado de citomegalovirus congénito en recién nacidos sanos e ingresados - Barcelona). (PI19/00095). Instituto de Salud Carlos III (ISCIII). 2020-2024. IP: Antoni Noguera Julián, Ana Alarcon Allen.
- Elaboración de un atlas morfométrico normalizado de la maduración cerebral del recién nacido prematuro y valoración del impacto de la patología cerebral en el proceso madurativo. (PI18/00110). Instituto de Salud Carlos III (ISCIII). 2019-2023. IP: Thais Agut Quijano.
- SGR 2022-2024_Grup Cervell Neonatal (GCN). (2021 SGR 01505). Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. 2022-2025. IP: Thais Agut Quijano.
- Evaluation of a new point-of-care risk-stratification tool based on quantification of prognostic biomarkers for the screening of the very sick patient to improve neonatal survival. Sociedad Española de Neonatología. 2023-2024. IP: Ana Alarcon Allen.

Neuropsychology

Roser Pueyo Benito, PhD



0
CLINICAL TRIALS &
OBSERVATIONAL STUDIES



10
PUBLICATIONS



48
IMPACT FACTOR
4.8 average impact factor



60%
PUBLICATIONS IN 1ST QUARTILE



20%
PUBLICATIONS IN 1ST DECILE



70%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Isabel García Garcá, Maria Mataró Serrat, Xavier Caldú Ferrús, Maria Angeles Jurado Luque

POSTDOC ASSOCIATED RESEARCHERS

Cristina Sánchez Castañeda

PHD STUDENTS

Anna Prunell Castañe, Adrià Bermudo Gallaguet, Montse Blasco Sierra, Maria García Galant, Sandra Luís Ruiz, Xavier Prats Soteras, Noemí Lamónja Vicente, Júlia Ballester Plané, Olga Laporta Hoyos, Jonatan Ottino González

ASSISTANT RESEARCHER

Gemma Monté Rubio

In 2023, the group achieved significant progress in advancing the study of neuropsychological functioning in individuals with risk factors like obesity, as well as exploring the neuropsychological consequences of congenital, perinatal, and acquired injuries (including cerebral palsy and stroke) along with their rehabilitation efforts. In addition to ongoing national research projects, the team is actively involved in new national and international initiatives, such as National Academy of Neuropsychology research grant and ERC. The ICREA-Acadèmia program continued its support for one of the team's researchers.

Furthermore, the group continued developing guidelines for the neuropsychological assessment of cognition and autism traits in children with cerebral palsy, securing funding through a national grant and a Marie Skłodowska-Curie Actions Postdoctoral Fellowship. Active projects resulted in publications highlighting the correlation between brain age, lesion volume, and functional outcomes in stroke patients, as well as research on cardiometabolic measures predicting impulsivity and white matter changes in adolescents. Additionally, a systematic review of interventions impacting cognitive functions in cerebral palsy was conducted.

The group's notable publications underscore its adeptness in addressing emerging challenges, such as investigating the neuropsychological sequelae of post-COVID situations.

related to poor executive function in people with post-COVID conditions. *J Neurol.* 2023 May;270(5):2392-2408. FI:6 (Q1)

- Prunell-Castañe A, Jurado MÁ, Ottino-González J, Prats-Soteras X, Sánchez Garre C, Cano Marco N, Salas Gómez-Pablos P, García-García I, Garolera M. Beyond BMI: cardiometabolic measures as predictors of impulsivity and white matter changes in adolescents. *Brain Struct Funct.* 2023 May;228(3-4):751-760. FI:3.1 (D1)
- Blasco M, García-Galant M, Berenguer-González A, Caldú X, Arqué M, Laporta-Hoyos O, Ballester-Plané J, Miralbell J, Jurado MÁ, Roser Pueyo. Interventions with an Impact on Cognitive Functions in Cerebral Palsy: a Systematic Review. *Neuropsychol Rev.* 2023 Jun;33(2):551-577. FI:5.8 (Q1)

SELECTED PUBLICATIONS

- Liew SL, Schweighofer N, Cole JH, Zavaliangos-Petropulu A, Lo BP, Han LKM, Hahn T, Schmaal L, Donnelly MR, Jeong JN, Wang Z, Abdullah A, Kim JH, Hutton A, Barisano G, Borich MR, Boyd LA, Brodtmann A, Bueteftisch CM, Byblow WD, Cassidy JM, Charalambous CC, Ciullo V, Conforto AB, Dacosta-Aguayo R, DiCarlo JA, Domin M, Dula AN, Egorova-Brumley N, Feng W, Geranmayeh F, Gregory CM, Hanlon CA, Hayward K, Holguin JA, Hordacre B, Jahanshad N, Kautz SA, Khlif MS, Kim H, Kuceyeski A, Lin DJ, Liu J, Lotze M, MacIntosh BJ, Margetis JL, Mataro M, Mohamed FB, Olafson ER, Park G, Piras F, Revill KP, Roberts P, Robertson AD, Sanossian N, Schambra HM, Seo NJ, Soekadar SR, Spalletta G, Stinear CM, Taga M, Tang WK, Thielman GT, Vecchio D, Ward NS, Westlye LT, Winstein CJ, Wittenberg GF, Wolf SL, Wong KA, Yu C, Cramer SC, Thompson PM. Association of Brain Age, Lesion Volume, and Functional Outcome in Patients With Stroke. *Neurology.* 2023 May 16;100(20):e2103-e2113. FI: 10.1 (D1)
- Ariza M, Cano N, Segura B, Adan A, Bargalló N, Caldú X, Campabadal A, Jurado MA, Mataró M, Pueyo R, Sala-Llonch R, Barrué C, Bejar J, Cortés CU; NAUTILUS Project Collaborative Group; Garolera M, Junqué C. COVID-19 severity is

SELECTED PROJECTS

- Evaluation of Autism SpEctrum Disorder in children with Cerebral Palsy (EASED-CP). 101066954. Unió Europea. MSCA Postdoctoral Fellowships 2021 (HORIZON-MSCA-2021-PF-01). 2022-2026. IP: Roser Pueyo Benito
- Guías basadas en la evidencia y consensuadas para la evaluación neuropsicológica de las personas con parálisis cerebral grave y parálisis cerebral discinética. PID2020-117163RB-I00. Ministerio de Ciencia e Innovación (MICINN). Convocatòria d'ajuts del Programa Estatal d'Investigació, Desenvolupament i Innovació Orientada als reptes de la Societat. Modalitat Reptes. 2021-2025. IP: Roser Pueyo Benito

Cognitive Neuroscience

Carles Escerca Micó, PhD



0
CLINICAL TRIALS &
OBSERVATIONAL STUDIES



15
PUBLICATIONS



53
IMPACT FACTOR
3.5 average impact factor



53%
PUBLICATIONS IN 1ST QUARTILE



13%
PUBLICATIONS IN 1ST DECILE



80%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Jordi Costa Faidella, Imma Clemente Lapena, Maria José Corral López, Maria Isabel Núñez Peña, Iria San Miguel Insúa, Hans Supèr, Marc Via Garcia

PHD STUDENTS

Sonia Arenillas Alcón, Marta Puertollano Rodríguez, Teresa Ribas Prats, Alejandro Lerer

We have pushed further our research on electrophysiological biomarkers of neurodevelopmental progress and derail, carried out in collaboration with the Foetal Environment and Obstetric Complications research group, led by Dr. Maria Dolores Gómez-Roig. This research capitalizes on a brain response to speech sounds retrieved from the electroencephalogram, the so-called *frequency-following response* (FFR). Five major milestones have been achieved. First, we extended our former findings on newborns with fetal growth restriction to newborns born large for gestational age: both groups show, in comparison to their adequate for gestational age peers, a diminished neural phase-locking to speech stimuli. Second, using neurosonography we have unveiled specific anatomical alterations in brain

regions related to speech processing [tectal length] in fetal growth restriction fetuses, highlighting our previous electrophysiological findings. Third, we were able to characterize the maturational trajectory of the FFR during the first 6 months of life, disclosing a striking fast maturation during the first month of life for the neural encoding of the formant structure of phonemes. Fourth, we showed that musical exposure during the last trimester of the pregnancy enhances the neonatal FFR. All these findings paved the way for our tech transfer strategy, which received support from the Producte program of AGAUR, towards establishing a universal “cognitive” screening at birth based on neurophysiological responses.

SELECTED PUBLICATIONS

- Ribas-Prats T, Cordero G, Lip-Sosa DL, Arenillas-Alcón S, Costa-Faidella J, Gómez-Roig MD, Escera C. Developmental Trajectory of the Frequency-Following Response During the First 6 Months of Life. *J Speech Lang Hear Res.* 2023 Dec 11;66(12):4785-4800. FI:2.6 (Q1)
- Ribas-Prats T, Arenillas-Alcón S, Pérez-Cruz M, Costa-Faidella J, Gómez-Roig MD, Escera C. Speech-Encoding Deficits in Neonates Born Large-for-Gestational Age as Revealed With the Envelope Frequency-Following Response. *Ear Hear.* 2023 Jul-Aug 01;44(4):829-841. FI:3.7 (D1)
- Lip-Sosa DL, Pérez-Cruz M, Ahumada-Droguett P, Ribas-Prats T, Puertollano M, García-Gómez MA, Mazarico E, Eixarch E, Escera C, Gómez-Roig MD. Corpus callosum-fastigium and tectal lengths in late-onset small fetuses. *Ultrasound Obstet Gynecol.* 2023 Aug;62(2):226-233. FI:7.1 (D1)
- Arenillas-Alcón S, Ribas-Prats T, Puertollano M, Mondéjar-Segovia A, Gómez-Roig MD, Costa-Faidella J, Escera C. Prenatal daily musical exposure is associated with enhanced neural representation of speech fundamental frequency: Evidence from neonatal frequency-following responses. *Dev Sci.* 2023 Sep;26(5):e13362. FI: 3.7 (Q1)

SELECTED PROJECTS

- Papel de la sincronía ocular en la sincronía neuronal y en la mejora

de las funciones cognitivas en pacientes con Alzheimer. PID2022-1399680B-I00. Ministerio de Ciencia e Innovación (MICINN). Convocatoria 2022 de ayudas a «Proyectos de Generación de Conocimiento». Modalidad Proyectos de «Investigación Orientada». 2023-2026. IP: Hendrik Anne Super.

- Eina predictiva del grau de neurodesenvolupament en nadons nascuts amb baix pes mitjançant intel·ligència artificial aplicada a registres d'EEG. F2I-PdC_2022-001. Fundació Bosch i Gimpera de la Universitat de Barcelona. Convocatòria Prova de concepte 2022. Fons per a l'Impuls de la Innovació - Fundació Bosch i Gimpera. 2023-2024. IP: Carles Escera Mico.
- Predicción del retraso en el neurodesarrollo en recién nacidos con bajo peso mediante inteligencia artificial aplicada a registros eeg ante sonidos del lenguaje. PDC2022-133044-I00. Ministerio de Ciencia e Innovación. Convocatoria 2022 de ayudas a proyectos de I+D+i para la realización de pruebas de concepto. 2022-2023. IP: Carles Escera Mico.
- Detección de riesgo de la enfermedad de Alzheimer mediante el análisis de movimientos oculares fijacionales registrados por una cámara selfie de un teléfono inteligente. PDC2022-133054-I00. Ministerio de Ciencia e Innovación. Convocatoria 2022 de ayudas a proyectos de I+D+i para la realización de pruebas de concepto. 2022-2023. IP: Hendrik Anne Super.

Attention, Perception and Language Acquisition (APAL)

Laura Bosch Galceran, PhD

0
CLINICAL TRIALS &
OBSERVATIONAL STUDIES

2
PUBLICATIONS

16
IMPACT FACTOR
7.9 average impact factor

100%
PUBLICATIONS IN 1ST QUARTILE

100%
PUBLICATIONS IN 1ST DECILE

50%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Ferran Pons Gimeno, Marta Ramon Casas

POSTDOC ASSOCIATED RESEARCHERS

Joan Birulés Muntané, Clément François

ASSISTANT RESEARCHER

Jessica Sánchez-Galán Frauca

During 2023, we have developed three research lines (MICINN project), focusing on the study of indexical representation, and lexical learning in audiovisual (AV) contexts, involving typically developing infants from monolingual and bilingual families, and also clinical populations. Research on the representation of indexical information has unveiled how early in development infants can consistently associate their language with the face/voice of the speaker. An early associative connection has been found by 4 months of age with no differences between infants in monolingual or bilingual environments (Marcet et al., *Frontiers in Psychology* in press). In a different study, 5-year-old bilingual children differed from

monolinguals in their attention patterns to the mouth of a speaker, revealing the benefit of using AV cues in dual language learning processes (Birulés et al., *Developmental Psychology* 2023). Regarding the research line on lexical learning, a first study has been completed within a broad collaborative research project with ORL, Neuroradiology and Psychology Units involving congenitally deaf infants who received a cochlear implant around 12 months of age, and a control group of hearing participants matched in auditory age and tested at 24 months of age. First analyses revealed the great variability within the implanted group in terms of language outcomes and word learning skills. The focus is now centered in the identification of relevant factors (from biomedical to socio-educative) to better explain the differences in language outcomes after implantation. This research line has benefited from the work of de B. de Diego-Lázaro, expert in deaf and hard-of-hearing infants and children. Regarding the third research line with the UnimTEA Unit (Multidisciplinary Unit of Autism Spectrum Disorder), an eye-tracking pilot study with 9 children diagnosed with ASD (under the age of 30 months) proved feasible in highly affected children, replicating findings from previous research based on the “social motivation hypothesis”.

SELECTED PUBLICATIONS

- Martinez-Alvarez A, Gervain J, Koulaguina E, Pons F, de Diego-Balaguer R. Prosodic cues enhance infants' sensitivity to nonadjacent regularities. *Sci Adv.* 2023 Apr 14;9(15):eade4083. FI:13.6 (D1)
- Ramon-Casas M, Cortés S, Benet A, Lleó C, Bosch L. Connecting perception and production in early Catalan-Spanish bilingual children: language dominance and quality of input effects. *J Child Lang.* 2023 Jan;50(1):155-176. FI:2.2 (Q1)

SELECTED PROJECTS

- Evaluación de nuevas actividades de vesículas secretadas por probióticos y microbiota intestinal y su potencial aplicación como postbióticos en salud humana. Ministerio de Ciencia, Innovación y Universidades. PID2019-107327RB-I00. Convocatòria d'ajuts

del Programa Estatal d'Investigació, Desenvolupament i Innovació Orientada als reptes de la Societat. Modalitat Reptes. 2020-2023. IP: Laura Baldoma Llavines.

- Estrategias postbióticas basadas en el microbioma intestinal: funciones reguladoras de vesículas de microbiota en la comunicación intestino-hígado e intestino-cerebro. Ministerio de Ciencia e Innovación (MICINN). PID2019-107327RB-I00. Convocatoria 2022 de ayudas a «Proyectos de Generación de Conocimiento». Modalidad Proyectos de «Investigación Orientada». 2023-2026. IP: Laura Baldoma

Research in Child and Adolescent Mental Health

Montserrat Dolz Abadia, MD, PhD



19
PROJECTS
9 competitive projects
10 non competitive projects



3
CLINICAL TRIALS &
OBSERVATIONAL STUDIES
2 commercial clinical trials
1 academic clinical trials



26
PUBLICATIONS



142
IMPACT FACTOR
5.5 average impact factor



62%
PUBLICATIONS IN 1ST QUARTILE



31%
PUBLICATIONS IN 1ST DECILE



35%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Alazne Gorrotxategi Aizpitarte, Aritz Aranbarri Paredes, Xavier Estrada Prat, Francisco Villar Cabeza, Laia Mollà Cusí, Esther Via Virgili, Neus Elias Carbonell, José Ángel Alda Díez, Mar Alvarez Segura, Ester Camprodon Rosanas, Maria Diez Juan, Fernando Lacasa Saludes, Ma Ángeles Mairena García de la Torre, Josep Lluís Matalí Costa, Daniel Muñoz Samons, Jordi Navarra Ordoño, Marta Pardo Gallego, Isabel Rueda Barcena, Bernardo Sánchez Fernández, Eduardo Serrano Troncoso, Anna Sintés Estevez

POSTDOC ASSOCIATED RESEARCHERS

Pablo Vidal-Ribas Belil, Inmaculada Insa Pineda, Anna Butjosa Molines

PHD STUDENTS

Laia Villalta Maciá, Ania Perez Racana, Anna Huguet Miguel, Claudia Caprile Elola - Olaso, Laia Portoles, Laura Puigcerver Navarro, Jordina Tor Fabra

ASSISTANT RESEARCHER

Carolina Morales Guerrero, Paula Resina Curado, Berta Sanfeliu Salvà,

Anna Martínez García, Sara Parrilla Carrasco, Ingrid Tortadès Arumí, Marina Fabrega Ribera, Carlota Saumell Andreu, Andrea González Contijoch

COLLABORATOR RESEARCHER

Carme Tello

During 2023, research has mostly focused on recruitment (projects awarded during 2022): a clinical trial using a community-based early intervention in autism (TEA-CARE, Dr Aranbarri, Dr Díez), the study of biorhythms in autism and attention-deficit and hyperactivity disorder (ADHD) (iCASE, Dr Alda, Dr Aranbarri), early treatment based on parent-intervention in children with mental health problems and genetic abnormalities (TheGAP, Dr Villalta), early genetic, psychological and environmental contributors to neurobehavioral and emotional problems (ENGINE, Dr Gascón, Dr Camprodon), neurobiological bases of emotion-regulation alterations in binge eating disorder and emotional eating (PREC-BED, Dr Via). Other European-funded studies are recruiting participants for the treatment of traumatized Children post-COVID-19 (RESCCUE, Dr Lacasa, Dr Alvarez). In the context of a collaboration with the Universitat de Barcelona, a study is evaluating the efficacy of exposure-based therapy using virtual reality in anorexia nervosa (Dr Gutiérrez-Maldonado, Dr Carulla). Part of our research involves the evaluation of innovative clinical programs; for example, during 2023, we have developed a new programme for patients with highly-complex eating disorders (MINERVA), which associates research on efficacy, efficiency, and patient's perception (Dr Serrano).

Two new projects have been awarded to the MIND-Escoles group, to study genetics in ADHD and neurodevelopment in university students (PI23/00040; PI23/00026), and an additional one designed to prevent mental health problems in adolescents (European project 101080934). They also validated into Spanish a new scale on sleep disturbances.

Dr Dolz, PI of the psychosis/risk-for-psychosis group, conducted a research

stage at the Boston's Children Hospital, enhancing expertise in resilience intervention in adolescents at-risk for psychosis. This experience will lead to new clinical and research programs aimed at preventing psychosis development. Recent data from the cohort of at-risk for psychosis adolescents (CAPRIS study, a collaborative study with the Hospital Clinic since 2012) suggested specific cognitive measures that predict, at baseline, the conversion to psychosis.

SELECTED PUBLICATIONS

- Toro-Pérez D, Limonero JT, Guillen M, Bolancé C, Vilarrubí SN, Camprodon-Rosanas E. Evaluating quality of life in pediatric palliative care: a cross-sectional analysis of children's and parents' perspectives. *Eur J Pediatr.* 2024 Mar;183(3):1305-1314. FI:3.6 (Q1)
- Tor J, Baeza I, Sintés-Estevez A, De la Serna E, Puig O, Muñoz-Samons D, Álvarez-Subiela J, Sugranyes G, Dolz M. Cognitive predictors of transition and remission of psychosis risk syndrome in a child and adolescent sample: longitudinal findings from the CAPRIS study. *Eur Child Adolesc Psychiatry.* 2024 Jan;33(1):89-104. FI:6.4 (D1)
- Alcover C, Mairena MÁ, Rodríguez-Medina J, Mezzatesta M, Balañá G, Elias N, Elias M, Arias-Pujol E. Measuring Changes in Social Skills Throughout an Intervention Program for Children with ASD, Contributions from Polar Coordinate Analysis. *J Autism Dev Disord.* 2023 Jun;53(6):2246-2260. FI:3.9 (Q1)
- Via E, Calvo A, de la Serna E, Blázquez A, Lázaro L, Andrés-Perpiñá S, Plana MT, Flamarique I, Martínez E, Pariente J, Moreno E, Bargallo N, Castro-Fornieles J. Longitudinal study in adolescent anorexia nervosa: evaluation of cortico-striatal and default mode network resting-state brain circuits. *Eur Child Adolesc Psychiatry.* 2023 Mar;32(3):513-526. FI:6.4 (D1)

SELECTED PROJECTS

- IMPROVA, H2020: e-Intervention Enhancing Mental Health in Adolescents. European Health and Digital Executive Agency. Project 101080934. C Europea. IP: R. Bosch y M. Casas.
- Trastornos del neurodesarrollo

en estudiantes universitarios: Prevalencia e impacto sobre el abandono académico. PI23/00040. Instituto de Salud Carlos III (ISCIII). IP: M. Pagerols.

- Más allá del riesgo genético para el TDAH: contribución genética a la resiliencia en una muestra poblacional. PI23/00026. Instituto de Salud Carlos III (ISCIII). IP: R. Bosch.
- Bases biológicas de los rasgos autistas: integración de las perspectivas de la medicina genómica, epigenómica y evolutiva (GenAUTIS). PID2022-1397400A-I00. IP: MIND Escolles (M. Casas, R. Bosch, etc.).

SELECTED CLINICAL STUDIES

- Estudio exploratorio a corto plazo para evaluar la seguridad, tolerabilidad y farmacocinética de Seltorexant como terapia adyuvante al tratamiento con antidepresivos en adolescentes con trastorno depresivo mayor que muestran una respuesta inadecuada a los inhibidores de recaptación de serotonina más psicoterapia. A Short-term Exploratory Study to Evaluate Safety, Tolerability, and Pharmacokinetics of Seltorexant as Adjunctive Therapy to Antidepressants in Adolescents with Major Depressive Disorder Who Have an Inadequate Response to an SSRI and Psychotherapy. (42847922MDD1016). Cano; Farmacia; Janssen - Cilag, S.A; Unitat de Recerca (URC) FSJD. 2021. IP: Montserrat Dolz Abadia.
- Ensayo controlado con placebo en sujetos con alto riesgo para la psicosis comparado con ácidos grasos omega-3 en Europa. (PURPOSE / ABR54654). University Medical Center Utrecht. 2016 - . IP: Montserrat Dolz Abadia.
- A Double-blind, Randomized, Psychoactive Placebo-controlled, Study to Evaluate the Efficacy and Safety of 3 Fixed Doses (28 mg, 56 mg and 84 mg) of Intranasal Esketamine in Addition to Comprehensive Standard of Care for the Rapid Reduction of the Symptoms of Major Depressive Disorder, Including Suicidal Ideation, in Pediatric Subjects Assessed to be at Imminent Risk for Suicide.. ESKETINSUI2002. Janssen - Cilag, S.A. 2018 - .. IP: Montserrat Dolz Abadia.

Epilepsy and Movement Disorders in Pediatrics

Alexandros Arzimanoglou, MD, PhD

15
PROJECTS
4 competitive projects
11 non competitive projects

12
CLINICAL TRIALS &
OBSERVATIONAL STUDIES
9 commercial clinical trials
3 academic clinical trials

27
PUBLICATIONS

159
IMPACT FACTOR
5.9 average impact factor

56%
PUBLICATIONS IN 1ST QUARTILE

22%
PUBLICATIONS IN 1ST DECILE

37%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Javier Aparicio, Maria del Carmen Fons Estupiña, Anna López Sala, Juan Darío Ortigoza Escobar

ASSISTANT RESEARCHER

Itziar Alonso Colmenero, Santiago Antonio Candela Cantó, Verónica Delgadillo Chilavert, Andrea Palacio Navarro, Alia Ramírez Camacho

SELECTED PUBLICATIONS

- Ousingsawat J, Talbi K, Gómez-Martín H, Koy A, Fernández-Jaén A, Tekgül H, Serdaroğlu E, Schreiber R, Ortigoza-Escobar JD, Kunzelmann K. Broadening the clinical spectrum: molecular mechanisms and new phenotypes of ANO3-dystonia. Brain. 2023 Dec 11:awad412. FI:14.2 (D1)
- Domínguez-Carral J, Ludlam WG, Junyent Segarra M, Fornaguera Martí M, Balsells S, Muchart J, Čokolić Petrović D, Espinoza I,

Ortigoza-Escobar JD, Martemyanov KA; GNA01-Study Group. Severity of GNA01-Related Disorder Correlates with Changes in G-Protein Function. Ann Neurol. 2023 Nov;94(5):987-1004. FI: 11.2 D1

- Nou-Fontanet L, Martín-Gómez C, Isabel-Gómez R, Bachoud-Lévi AC, Zorzi G, Capuano A; NKX2-1-Related Disorders Guideline Working Group; Blasco-Amaro JA, Ortigoza-Escobar JD. Systematic review of drug therapy for chorea in NKX2-1-related disorders: Efficacy and safety evidence from case studies and series. Eur J Neurol. 2023 Dec;30(12):3928-3948. FI: 5.1 Q1
- Casas-Alba D, Aguilar A, Alonso I, García MT, Cilio MR, Fons C; BIO-KCNQ2-Spanish Study Group. Relationship Between Epileptic Activity and Developmental Outcome in KCNQ2-Related Epilepsy. Pediatr Neurol. 2023 Jul;144:11-15. FI: 3.8 (Q1)

SELECTED PROJECTS

- Safety and therapeutic measures of transcranial cathodal direct current stimulation (tdcs) in patients with refractory focal epilepsy. Medidas de seguridad y terapéuticas de la estimulación con corriente continua catódica transcraneal (tdcs) en pacientes con epilepsia focal resistente al tratamiento. (NE001). Diagnóstico para la imagen (DXI); Ergomedplc; Farmacia; Laboratori HSJD; Neuroelectrics Barcelona S.L; Unitat de Recerca (URC) FSJD. 2023. IP: Maria Alejandra Climent Perin.
- El análisis de la conectividad cerebral como herramienta para evaluar los trastornos neuroconductuales y de regulación emocional (EpileTRE.net). (PI21/01268). Instituto de Salud Carlos III (ISCIII). 2022-2024. IP: Alexis Arzimanoglou.
- Mapas de conectividad funcional cerebral y neurodesarrollo en epilepsias pediátricas de debut precoz (Proyecto EPI-NEUROMAPS). (PI22/01441). Instituto de Salud Carlos III (ISCIII). 2023-2025. IP: Maria del Carmen Fons Estupiña
- PFNR0177_Seguridad y eficacia de la estimulación transcraneal directa de corriente continua (tDCS) en pacientes pediátricos con enfermedades neuropsiquiátricas.

(FSJD-NeuroHoop-2023 (R)). Climent; Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD; Fundació Ramon Areces. 2023-2025. IP: Maria Alejandra Climent Perin

SELECTED CLINICAL STUDIES

- A phase 3, randomized, double-blind, placebo-controlled, fixed-dose, multicenter study to examine the efficacy and safety of ZX008 in subjects with CDKL5 deficiency disorder followed by an open-label extension. [ZX008-2103]. Farmacia; Hospital de dia URC; Laboratori HSJD; Moratinos; Pharmaceutical Research Associates España SAU; Unitat de Recerca (URC) FSJD; Zogenix, Inc. 2023. IP: Alia Ramirez Camacho.
- Estudio aleatorizado, controlado con placebo y doble ciego para investigar la eficacia y la seguridad del carisbamato (YKP509) como tratamiento complementario para las crisis asociadas al síndrome de Lennox-Gastaut en niños y adultos, con extensión abierta opcional. A randomized, double-blind, placebo-controlled study to investigate the efficacy and safety of carisbamato (YKP509) as adjunctive treatment for seizures associated with Lennox-Gastaut Syndrome in children and adults, with optional openlabel extension. [YKP509C003]. Farmacia; Laboratori HSJD; Merino; Pharmaceutical Product Development Spain, S.L.; Servicio de Cardiología; Servicio Neuropsicología; SK Life Science, INC; Unitat de Recerca (URC) FSJD. 2023. IP: Javier Aparicio.
- Estudio de extensión abierto a largo plazo para evaluar la seguridad y tolerabilidad de NBI-827104 en sujetos pediátricos con encefalopatía epiléptica con puntas-ondas continuas durante el sueño. Long-Term, Open-Label Extension Study to Evaluate the Safety and Tolerability of NBI-827104 in Pediatric Subjects with Epileptic Encephalopathy with Continuous Spike-and-Wave During Sleep. [NBI-827104-CSWS2025]. ACCELSIORS; Farmacia; Neurocrine Biosciencias, inc.; Servicio de Cardiología; Servicio Epilepsia; Servicio Neuropsicología; Servicio Oftalmología; Unitat de Recerca (URC) FSJD; Vaquero; Viver. 2022. IP: Alia Ramirez Camacho.
- Estudio de prolongación sin enmascaramiento con XEN496 en niños que padecen encefalopatía epiléptica y del desarrollo por KCNQ2. An Open-Label Extension Study of XEN496 in Pediatric Subjects with KCNQ2 Developmental and Epileptic Encephalopathy. [XPF-009-302]. Farmacia; Icon Clinical Research España, S.L.; Servicio de Cardiología; Servicio Epilepsia; Servicio Neurofisiología; Unitat de Recerca (URC) FSJD; Xenon Pharmaceuticals Inc. 2022. IP: Maria del Carmen Fons Estupiña.



7.1 MEDICINE AND PEDIATRIC SCIENCE AREA

7.1.2 Adaptive Response, Metabolism and Immunobiology

Foetal Medicine and Surgery
Metabolic Endocrinology
Metabolic Diseases of Paediatric origin
Adipose Tissue Molecular Biology and Gene Regulation and
Associated Disorders
Neonatology Research Group (GReN)
Pharmacological Targets in Inflammation and Metabolic Diseases
Allergic Diseases in Childhood and Adolescence
Study of Immune Deficiency Diseases in Paediatrics (GEMDIP)
Infectious Diseases and Microbiome
Gut Microbiota
Environment and Complications during Pregnancy
Cardiovascular Diseases and Child Development
Immune and Respiratory Dysfunction in Critically Ill Children



Foetal Medicine and Surgery

Eduard Gratacós Solsona, MD, PhD



8
PROJECTS
5 competitive projects
3 non competitive projects



1
CLINICAL TRIALS &
OBSERVATIONAL STUDIES
0 commercial clinical trials
1 academic clinical trials



24
PUBLICATIONS



138
IMPACT FACTOR
5.7 average impact factor



71%
PUBLICATIONS IN 1ST QUARTILE



33%
PUBLICATIONS IN 1ST DECILE



50%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

xxx

PHD STUDENTS

xxx

COLLABORATOR RESEARCHER

xxx

In 2023, we continued our research aimed at improving diagnosis and therapy in maternal-fetal medicine. We are working on several projects involving image processing, clinical trials and the development of new medical devices in fetal therapy.

As a highlight of 2023, we published in JAMA Network Open our most recent findings of the follow-up of the IMPACT-BCN clinical trial: the positive consequences of maternal lifestyle interventions during pregnancy based on Mediterranean diet and stress reduction to an improvement of infant neurodevelopment at two years of age. At this moment, we are fully engaged in the development on the PE37 trial, a multinational clinical trial involving

nearly 10,000 participants to prevent late preeclampsia.

Additionally, we have secured funding for several competitive and private projects, Fundació La Caixa has renovated the grant to start the second phase of the CaixaResearch Artificial Placenta project.

At the same time, we remain committed to training the next generation of researchers in the field of maternal-fetal medicine through training fellowship programs.

SELECTED PUBLICATIONS

- Smith ER, Oakley E, Grandner GW, Rukundo G, Farooq F, Ferguson K, Baumann S, Adams Waldorf KM, Afshar Y, Ahlberg M, Ahmadzia H, Akelo V, Aldrovandi G, Bevilacqua E, Bracero N, Brandt JS, Broutet N, Carrillo J, Conry J, Cosmi E, Crispi F, Crovetto F, Del Mar Gil M, Delgado-López C, Divakar H, Driscoll AJ, Favre G, Fernandez Buhigas I, Flaherman V, Gale C, Godwin CL, Gottlieb S, Gratacós E, He S, Hernandez O, Jones S, Joshi S, Kalafat E, Khagayi S, Knight M, Kotloff KL, Lanzone A, Laurita Longo V, Le Doare K, Lees C, Litman E, Lokken EM, Madhi SA, Magee LA, Martinez-Portilla RJ, Metz TD, Miller ES, Money D, Mounghmaithong S, Mullins E, Nachega JB, Nunes MC, Onyango D, Panchaud A, Poon LC, Raiten D, Regan L, Sahota D, Sakowicz A, Sanin-Blair J, Stephansson O, Temmerman M, Thorson A, Thwin SS, Tippet Barr BA, Tolosa JE, Tug N, Valencia-Prado M, Visentin S, von Dadelszen P, Whitehead C, Wood M, Yang H, Zavala R, Tielsch JM. Clinical risk factors of adverse outcomes among women with COVID-19 in the pregnancy and postpartum period: a sequential, prospective meta-analysis. *Am J Obstet Gynecol*. 2023 Feb;228(2):161-177. FI: 9.8 [D1]
- Sanchez-Martinez S, Randanne PC, Hawkins-Villarreal A, Rezaei K, Fucho R, Bobillo-Perez S, Bonet-Carne E, Illa M, Eixarch E, Bijmens B, Crispi F, Gratacós E; Collaborators. Acute fetal cardiovascular adaptation to artificial placenta in sheep model. *Ultrasound Obstet Gynecol*. 2023 Aug;62(2):255-265. FI:7.1 [D1]
- Crovetto F, Nakaki A, Arranz A, Borrás R, Vellvé K, Paules C,

Boutet ML, Castro-Barquero S, Freitas T, Casas R, Martín-Asuero A, Oller Guzmán T, Morilla I, Martínez-Àran A, Camacho A, Pasqual M, Izquierdo Renau M, Pozo ÓJ, Gomez-Gomez A, Estruch R, Vieta E, Crispi F, Gratacós E. Effect of a Mediterranean Diet or Mindfulness-Based Stress Reduction During Pregnancy on Child Neurodevelopment: A Prespecified Analysis of the IMPACT BCN Randomized Clinical Trial. *JAMA Netw Open*. 2023 Aug 1;6(8):e2330255. FI:13.8 [D1]

- Nakaki A, Crovetto F, Urru A, Piella G, Borrás R, Comte V, Vellvé K, Paules C, Segalés L, Dacal M, Gomez Y, Youssef L, Casas R, Castro-Barquero S, Martín-Asuero A, Oller Guzmán T, Morilla I, Martínez-Àran A, Camacho A, Pascual Tutusaus M, Arranz A, Rebollo-Polo M, Gomez-Chiari M, Bargallo N, Pozo ÓJ, Gomez-Gomez A, Izquierdo Renau M, Eixarch E, Vieta E, Estruch R, Crispi F, Gonzalez-Ballester MA, Gratacós E. Effects of Mediterranean diet or mindfulness-based stress reduction on fetal and neonatal brain development: a secondary analysis of a randomized clinical trial. *Am J Obstet Gynecol MFM*. 2023 Dec;5(12):101188. FI:6.3 [D1]

SELECTED PROJECTS


- Cerebra II - Preventing prenatal brain damage with tools for improving risk identification and therapy. Cerebra For Brain Injured Children & Young People. 2020-2024. IP: Eduard Gratacós Solsona.
- Randomized open-label interventional trial of induction of labour at 37 weeks of gestation in women with high sFlt1/PLGF ratio. PE37 TRIAL. Roche Diagnostics, S.L. 2020-2024. IP: Eduard Gratacós Solsona.
- FET-SEAL: Sistema integrado de sellado de membranas fetales para cirugía fetal fetoscópica. (DTS20/00130). Instituto de Salud Carlos III (ISCIII). 2021-2023. IP: Miriam Illa Armengol.
- Fase 2 del Programa Interdisciplinar para el Desarrollo de un Prototipo Experimental de Placenta Artificial y Evaluación de Aplicación Clínica. (PCP00390). Fundació Bancària "La Caixa". 2023-2026. IP: Eduard Gratacós Solsona.


SELECTED CLINICAL STUDIES

- Estudio prospectivo observacional sobre cohorte de fetos con Flutter auricular o taquicardia supraventricular. (FAST REGISTRY). SickKids. 2018. IP: Eduard Gratacós Solsona.


Metabolic Endocrinology

Lourdes Ibáñez Toda, MD, PhD

 **7**
PROJECTS
6 competitive projects
1 non competitive projects

 **5**
CLINICAL TRIALS &
OBSERVATIONAL STUDIES
2 commercial clinical trials
3 academic clinical trials

 **22**
PUBLICATIONS

 **125**
IMPACT FACTOR
5.7 average impact factor

 **77%**
PUBLICATIONS IN 1ST QUARTILE

 **9%**
PUBLICATIONS IN 1ST DECILE

 **50%**
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Silvia Marin Del Barrio, Silvia Xargay Torrent, Paula Casano Sancho, Marta Díaz Silva

PHD STUDENTS

Cristina García Beltran

COLLABORATOR RESEARCHER

Judith Bassols Casadevall, Abel López Bermejo

During 2023, our team has focused on studies mainly related to polycystic ovary syndrome (PCOS), a prevalent entity in adolescent girls and young women, which is the most frequent cause of female subfertility, and which has no approved treatment so far. First, we have further expanded the recruitment campaign to include adolescent girls and young women with PCOS in a multicenter, multinational, randomized, double-blind, and placebo-controlled clinical trial funded by the European Commission, whose objective is to confirm the efficacy and safety of

a first treatment for this disorder. This approach combines a lifestyle intervention program with a pharmacological therapy consisting of a single pill containing low doses of three medications acting on different pathways with different targets. In addition, we have studied a series of molecules, namely, organokines, related to adipose tissue function and to non-alcoholic fatty liver disease (NAFLD), and entity closely associated with PCOS, to ascertain whether some of these molecules could be used as early markers for this disorder. We have also started the recruitment for another clinical trial comparing the effects of a low-dose combination of two insulin sensitizers and one anti-androgen on the outcome of bone age maturation in girls with advanced puberty (breast development between age 8-9 yr) and with a history of an upward mismatch between birth weight and prepubertal body mass index. Finally, we have completed and published the first set of results related to the longitudinal characterization of circulating exosomes in children born appropriate- (AGA) or small-for-gestational-age (SGA). We are currently analyzing the exosomal proteome in the same population. Finally, we have started the recruitment of AGA and SGA patients for the longitudinal assessment of exosomal microRNAs in these populations.

SELECTED PUBLICATIONS

- Díaz M, Casano P, Quesada T, López-Bermejo A, de Zegher F, Villarroya F, Ibáñez L. Circulating exosomes decrease in size and increase in number between birth and age 7: relations to fetal growth and liver fat. *Front Endocrinol (Lausanne)*. 2023 Nov 2;14:1257768. IF:5.2 (Q1)
- de Zegher F, Díaz M, Ibáñez L. Adolescent polycystic ovary syndrome without obesity: HOTAIR rs1443512 genotype relates to fat mass and to the redistribution of fat mass on low-dose pioglitazone. *J Endocrinol Invest*. 2024 Apr;47(4):1037-1043. IF:5.4 (Q1)
- Ibáñez L, de Zegher F. Adolescent PCOS: a postpubertal central obesity syndrome. *Trends Mol Med*. 2023 May;29(5):354-363. IF:14 (D1)
- Garcia-Beltran C, Navarro-Gascon

A, López-Bermejo A, Quesada-López T, de Zegher F, Ibañez L, Villarroya F. Meteorin-like levels are associated with active brown adipose tissue in early infancy. *Front Endocrinol (Lausanne)*. 2023 Mar 2;14:1136245. IF:5.2 (Q1)

SELECTED PROJECTS

- SPIOMET4HEALTH_PCOS in adolescent girls and young women: Toward a treatment guided by pathophysiology. (899671). European Commission. 2021-2026. IP: Lourdes Ibañez Toda.
- SGR 2022-2024_Resposta adaptativa, metabolisme e immunobiologia. *Endocrinologia metabòlica*. (2021 SGR 00659). Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. 2022-2025. IP: Lourdes Ibañez Toda.
- Efectos del tratamiento con spiomet a mitad de dosis en niñas con pubertad adelantada y aceleración de la maduración ósea: estudio multicéntrico, aleatorizado y controlado con placebo. (IC121/00005). Institut d'Investigació Biomèdica de Girona Dr. Josep Trueta. 2022-2025. IP: Lourdes Ibañez Toda.
- Variaciones longitudinales del perfil de expresión de microRNAs exosomales en niños con bajo peso para la edad gestacional con y sin recuperación de peso y talla postnatal. (PI22/01424). Casano; Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD; Instituto de Salud Carlos III (ISCIII). 2023-2025. IP: Paula Casano Sancho, Marta Díaz Silva.

SELECTED CLINICAL STUDIES

- "Identification of circulating microRNAs in short children born small for gestational age: Relationship with pre- and postnatal growth and metabolic parameters". (LOR-HOR-2013-01). López. 2014. IP: Lourdes Ibañez Toda.
- Ensayo clínico de fase II, aleatorizado, multicéntrico y multinacional para evaluar la eficacia, la tolerabilidad y la seguridad de una combinación de dosis fijas de espironolactona, pioglitazona y metformina (SPIOMET) para adolescentes y mujeres adultas jóvenes (AYA) con síndrome de ovario poliquístico

(SOP) A Phase II, randomised, multi-centric, multi-national clinical trial to evaluate the efficacy, tolerability, and safety of a fixed dose combination of Spironolactone, Pioglitazone & Metformin (SPIOMET) for adolescent girls and young adult women (AYAs) with polycystic ovary syndrome (PCOS) / Ensayo clínico de fase II, aleatorizado, multicéntrico y multinacional para evaluar la eficacia, la tolerabilidad y la seguridad de una combinación de dosis fijas de espironolactona, pioglitazona y metformina (SPIOMET) para adolescentes y mujeres adultas jóvenes con síndrome de ovario poliquístico (SOP). (SPIOMET4HEALTH). Carranza; Diagnóstico para la imagen (DXI); Farmacia; Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD; Garcia; Laboratori HSJD; Optimapharm Contract Research Organisation, S.L.U; SIMM Molecular S.L.; Unitat de Recerca (URC) FSJD; Zamfir. 2022. IP: Lourdes Ibañez Toda.

- Efectos del tratamiento con spiomet a mitad de dosis en niñas con pubertad adelantada y aceleración de la maduración ósea: estudio multicéntrico, aleatorizado y controlado con placebo. (Mini-Spiomet). Diagnóstico para la imagen (DXI); Farmacia; Institut d'Investigació Biomèdica de Girona Dr. Josep Trueta; Laboratori HSJD; Serrate; SIMM Molecular S.L.; Unitat de Recerca (URC) FSJD. 2022. IP: Lourdes Ibañez Toda.

Metabolic Diseases of Paediatric origin

Josep Jiménez Chillarón, PhD & Carles Lerin Martínez, PhD

12 PROJECTS
10 competitive projects
2 non competitive projects

6 CLINICAL TRIALS & OBSERVATIONAL STUDIES
5 commercial clinical trials
1 academic clinical trials

11 PUBLICATIONS

62 IMPACT FACTOR
5.6 average impact factor

82% PUBLICATIONS IN 1ST QUARTILE

9% PUBLICATIONS IN 1ST DECILE

55% LEADERSHIP

MEMBERS

STAFF RESEARCHER

Montserrat Amat Bou, Serafin Murillo Garcia, Marina Llobet Garces, Ruben Díaz Naderi, Marta Ramon Krauel

POSTDOC ASSOCIATED RESEARCHERS

Flavio Palmieri, Marcela Catalina Parra Vargas

PHD STUDENTS

Marta Mourin Fernández, Paula Ximena Molina Giraldo, Charlotte Martine Juton

COLLABORATOR RESEARCHER

Ivonne Palacios Marin

ASSISTANT RESEARCHER

Ana Lopez Bataller, Lorena González López

SELECTED PUBLICATIONS

- Bang P, Polak M, Bossowski A, Maghnie M, Argente J, Ramon M et al. Frequency and Predictive Factors of Hypoglycemia in Patients Treated With rhIGF-1: Data From the Eu-IGFD Registry. *J. Clin. Endocrinol.*

- Metab. 2023;109(1):46-56. IF:5.8 (Q1)
- Díaz M, Mestres A, Lerin C, Cereijo R, López A, de Zegher F et al. Circulating progranulin in human infants: relation to prenatal growth and early postnatal nutrition. *Pediatr. Res.* 2023;94(3):1189-1194. IF:3.6 (Q1)
 - Palacios I, Serra D, Jimenez JC, LAURA R, Todorcevic M. Adipose Tissue Dynamics: Cellular and Lipid Turnover in Health and Disease. *Nutrients.* 2023;15(18) IF:5.9 (Q1)
 - Palacios I, Serra D, Jimenez JC, LAURA R, Todorcevic M. Childhood obesity: Implications on adipose tissue dynamics and metabolic health. *Obes. Rev.* 2023;24(12) IF:8.9 (Q1)

SELECTED PROJECTS

- Efecto de una pauta de alimentación de bajo contenido en hidratos de carbono en adolescentes con diabetes tipo 1. Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD; Sociedad Española de Diabetes. 2023-2025. IP: Serafin Murillo Garcia.
- SGR 2022-2024_Malalties metabòliques d'origen pediàtric. (2021 SGR 00661). Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. 2022-2025. IP: Carles Lerin Martinez.
- Modulación de la dieta durante las primeras etapas de la vida como estrategia de prevención de la obesidad infantil. (PDC2021-121816-100). Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD; Lerin; Ministerio de Economía Y Competitividad (MINECO). 2021-2024. IP: Carles Lerin Martinez
- El microbioma intestinal como diana terapéutica en el síndrome de Prader-Willi: efectos metabólicos y en el comportamiento de los pacientes. (PI20/00301). Instituto de Salud Carlos III (ISCIII). 2021-2025. IP: Marta Ramon Krauel.

SELECTED CLINICAL STUDIES

- A multi-country, non-interventional, prospective cohort study to evaluate the long-term safety and effectiveness of Pfizer's long-acting growth hormone (currently in development). Pfizer Registry of Outcomes in Growth hormone REsearch (PROGRES). (C0311015).

- IQVIA Ltd.; Pfizer, S.L.U. 2023. IP: Marta Ramon Krauel.
- Estudio observacional retrospectivo para evaluar el manejo diagnóstico y terapéutico del síndrome de Noonan en España. Retrospective observational study to assess the diagnostic and therapeutic management of Noonan Syndrome in Spain. (U1111-1261-0351). Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD. 2023. IP: Larisa Suarez Ortega, Marta Ramon Krauel
 - A study comparing the effect and safety of once weekly dosing of somapacitan with daily Norditropin® as well as evaluating long-term safety of somapacitan in a basket study design in children with short stature either born small for gestational age or with Turner syndrome, Noonan syndrome, or idiopathic short stature. (INN8640-4467). de Quirós; Diagnóstico para la imagen (DXI); Farmacia; Méndez; Novo Nordisk a/s; Santos; Servicio de Cardiología; Unitat de R A Phase 3, Randomized, Double-Blind, Placebo-Controlled, Four-Arm Parallel-Group Study of Setmelanotide in Patients with Specific Gene Defects in the Melanocortin-4 Receptor Pathway. (RM-493-035). Farmacia; Lopez; Medina; RHYTHM Pharmaceuticals, Inc.; Servicio de Cardiología; Unitat de Recerca (URC) FSJD. 2022. IP: Marta Ramon Krauel. ecerca (URC) FSJD. 2022. IP: Montserrat Amat Bou.

Adipose Tissue Molecular Biology and Gene Regulation and Associated Disorders

Francesc Villarroya Gombau, PhD

0 CLINICAL TRIALS & OBSERVATIONAL STUDIES

13 PUBLICATIONS

80 IMPACT FACTOR
6.2 average impact factor

92% PUBLICATIONS IN 1ST QUARTILE

15% PUBLICATIONS IN 1ST DECILE

46% LEADERSHIP

MEMBERS

STAFF RESEARCHER

Aleix Gavalda Navarro, Joan Villarroya Terrade, Marta Giralt Oms, Roser Iglesias Coll, Teresa Mampel Astals, Anna Planavila Porta

POSTDOC ASSOCIATED RESEARCHERS

Marion Peyrou, Tania Quesada López, Montserrat Cairó Calzada, Rubén Cereijo Téllez

PHD STUDENTS

Laura Campderrós Traver, Gemma Ferrer Curriu, Samantha Morón Ros, Celia Helena Rupérez Gonzalo

TECHNICIAN

Mercedes Morales Rueda, Gemma Moreno Besora, Albert Però Garcia

SELECTED PUBLICATIONS

- Morón-Ros S, Blasco-Roset A, Navarro-Gascon A, Rupérez C, Zamora M, Crispi F, Uriarte I, Fernández-Barrera MG, Avila M, Ferrer-Curriu G, Lupón J, Bayés-Genis A, Villarroya F, Gavalda-Navarro A, Planavila A. A new FGF15/19-mediated gut-to-heart

axis controls cardiac hypertrophy. *J Pathol.* 2023 Nov;261(3):335-348. IF: 7.3 (D1).

- Díaz M, Mestres-Arenas A, Lerin C, Cereijo R, López-Bermejo A, de Zegher F, Villarroya F, Ibáñez L. Circulating progranulin in human infants: relation to prenatal growth and early postnatal nutrition. *Pediatr Res.* 2023 Sep;94(3):1189-1194. IF: 3.6 (Q1).
- Díaz M, Carreras-Badosa G, Villarroya J, Gavaldà-Navarro A, Bassols J, de Zegher F, López-Bermejo A, Villarroya F, Ibáñez L. Circulating GDF15 concentrations in girls with low birth weight: effects of prolonged metformin treatment. *Pediatr Res.* 2023 Mar;93(4):964-968. IF: 3.6 (Q1).
- Castellá M, Blasco-Roset A, Peyrou M, Gavaldà-Navarro A, Villarroya J, Quesada-López T, Lorente-Poch L, Sancho J, Szymczak F, Piron A, Rodríguez-Fernández S, Carobbio S, Goday A, Domingo P, Vidal-Puig A, Giral M, Eizirik DL, Villarroya F, Cereijo R. Adipose tissue plasticity in pheochromocytoma patients suggests a role of the splicing machinery in human adipose browning. *iScience.* 2023 May 9;26(6):106847. IF: 5.8 (Q1).

SELECTED PROJECTS

- La autofagia mediada por chaperonas, un nuevo actor en el control de la plasticidad del tejido adiposo. CNS2022-135516. Ministerio de Ciencia e Innovación (MICINN). Ayudas para Incentivar la Consolidación Investigadora. 2023-2025. IP: Joan Villarroya Terrade.
- Nuevos retos en investigación sobre plasticidad adiposa. RED2022-134313-T. Ministerio de Ciencia e Innovación (MICINN). Convocatòria d'ajuts del Programa Estatal de Foment de la Investigació Científica i Tècnica d'Excel·lència. Subprograma Estatal de generació del coneixement. Accions de dinamització 'Xarxes d'Excel·lència'. 2023-2025. IP: Francesc Villarroya Gombau.
- Modulación de la inflamación y el metabolismo cardíaco para curar el corazón: Nuevas cardioquinas y mecanismos involucrados. PID2021-122941OB-I00. Ministerio de Ciencia e Innovación (MICINN). Convocatoria 2021 de ayudas a «Proyectos de Generación de Conocimiento».

Modalidad Proyectos de «Investigación Orientada». 2022-2025. IP: Ana Planavila Porta.

- Metabolisme molecular i patologies associades. 2021 SGR 00227. Agència de Gestió d'Ajuts Universitaris i de Recerca (AGAUR). Ajuts per donar suport a l'activitat científica dels grups de recerca de Catalunya (SGR-Cat 2021). 2022-2025. IP: Francesc Villarroya Gombau.

Neonatology Research Group (GReN)

Martin Iriondo Sanz, MD, PhD

8 PROJECTS
5 competitive projects
3 non competitive projects

5 CLINICAL TRIALS & OBSERVATIONAL STUDIES
0 commercial clinical trials
5 academic clinical trials

15 PUBLICATIONS

214 IMPACT FACTOR
14.2 average impact factor

40% PUBLICATIONS IN 1ST QUARTILE

20% PUBLICATIONS IN 1ST DECILE

33% LEADERSHIP

MEMBERS

STAFF RESEARCHER

Marta del Olmo Fernández, Cristina Carrasco Carrasco, Isabel Iglesias Platas, Ana Morillo Palomo

POSTDOC ASSOCIATED RESEARCHERS

Marta Camprubi Camprubi, Africa Pertierra Cortada

PHD STUDENTS

Carla Balcells Esponera, Jordi Clotet Caba, Ruth Del Rio Florentino, Beatriz Del Rey Hurtado De Mendoza, Montserrat Izquierdo Renau, Alba Rivas Piorno

ASSISTANT RESEARCHER

Ines Fernanda Medina Rivera

COLLABORATOR RESEARCHER

María Dolores Salvia Roiges, Mattia Bosio, Marta Thió Lluch, Cristina Borràs Novell, Ana Herranz Barbero

SELECTED PUBLICATIONS

- Pertierra Cortada A, Clotet Caba J, Hadley S, Sabrià Bach J, Iriondo Sanz M, Camprubi Camprubi M. Do FETO CDH survivors need the

same follow-up program as non-FETO patients? Eur J Pediatr. 2023 Jul;182(7):3243-3255. FI: 3.6 (Q1)

- Cañizo-Vazquez D, Hadley S, Leonhardt M, Camprubi-Camprubi M, Sanchez-de-Toledo J. Early post-operative neurodevelopment and visual assessment in neonates with congenital heart disease undergoing cardiac surgery. J Perinatol. 2023 Jul;43(7):856-863. FI: 2.9 Q2
- Borràs-Novell C, Herranz Barbero A, Balcells Esponera C, López-Abad M, Aldecoa Bilbao V, Izquierdo Renau M, Iglesias Platas I. Influence of maternal and perinatal factors on macronutrient content of very preterm human milk during the first weeks after birth. J Perinatol. 2023 Jan;43(1):52-59. FI: 2.9 (Q2)
- Herranz Barbero A, Iglesias-Platas I, Prat-Ortells J, Clotet Caba J, Moreno Hernando J, Castañón García-Alix M, Pertierra Cortada Á. Transpyloric Tube Placement Shortens Time to Full Feeding in Left Congenital Diaphragmatic Hernia. J Pediatr Surg. 2023 Nov;58(11):2098-2104. FI: 2.4 (Q2)

SELECTED PROJECTS

- Imágenes biofotónicas del cerebro infantil, el eslabón perdido entre el daño cerebral celular y la unidad neurovascular durante la enfermedad aguda. (FSJD-TinyBrains-2022). Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD. 2023. IP: Marta Camprubi Camprubi
- Eficacia y seguridad de la administración de Budesonida con Surfactante endotraqueal en la prevención de displasia broncopulmonar del recién nacido muy prematuro (BuS). Sociedad Española de Neonatología. 2022-2025. IP: Cristina Carrasco Carrasco.
- Validación prospectiva mediante un estudio multicéntrico estatal (REDSAMID) de modelos predictivos de mortalidad neonatal en prematuros. (PI20/00295). Instituto de Salud Carlos III (ISCIII). 2021-2025. IP: Martin Iriondo Sanz.
- Consecuencias en la infancia del fallo perinatal de crecimiento en Recién Nacidos Muy Prematuros: estudio clínico y metabólico. (PI20/00294). Instituto de Salud Carlos III (ISCIII). 2021-2025. IP: Isabel Iglesias Platas.

SELECTED CLINICAL STUDIES


- EFICACIA Y SEGURIDAD DE LA ADMINISTRACIÓN DE BUDESONIDA CON SURFACTANTE PULMONAR PORCINO ENDOTRAQUEAL EN LA PREVENCIÓN DE DISPLASIA BRONCOPULMONAR DEL RECIÉN NACIDO PREMATURO <32 SEMANAS DE EDAD GESTACIONAL: ENSAYO CLÍNICO ALEATORIZADO MULTICÉNTRICO (BuS). (BuS2020). Fundació de Recerca Clínic Barcelona-Institut d'Investigacions Biomèdiques August Pi i Sunyer. 2023. IP: Cristina Carrasco Carrasco.
- Efecto del Alopurinol en pacientes con encefalopatía hipóxico-isquémica tratados con hipotermia. ALBINO ensayo clínico multicéntrico, aleatorizado, ciego, controlado con placebo. (H2020-PHC-18-2015-667224). Instituto de investigación Sanitaria Hospital La Fe. 2017. IP: Marta Camprubi Camprubi.
- Evaluation of Real Time Continuous Glucose Monitoring in the very preterm infant. (REACT RCT). University of Cambridge. 2017. IP: Isabel Iglesias Platas.
- Recién nacidos con edad gestacional extremadamente baja asignados al azar, a ciegas, para ser resucitados resucitado con un 21% frente al 60% de oxígeno: influencia sobre la mortalidad y las enfermedades crónicas en el período neonatal. (1645-CI-057). Instituto de investigación Sanitaria Hospital La Fe. 2015. IP: Martin Iriondo Sanz.

Pharmacological Targets in Inflammation and Metabolic Diseases


Manuel Vázquez Carrera, PhD

 **0** CLINICAL TRIALS & OBSERVATIONAL STUDIES

 **6** PUBLICATIONS

 **51** IMPACT FACTOR
8.5 average impact factor

 **83%** PUBLICATIONS IN 1ST QUARTILE

 **67%** PUBLICATIONS IN 1ST DECILE

 **100%** LEADERSHIP

MEMBERS

STAFF RESEARCHER

F. Xavier Palomer Tarridas

POSTDOC ASSOCIATED RESEARCHERS

Emma Barroso Fernández, Marta Montori Grau

SELECTED PUBLICATIONS

- Barroso E, Montori-Grau M, Wahli W, Palomer X, Vázquez-Carrera M. Striking a gut-liver balance for the antidiabetic effects of metformin. Trends Pharmacol Sci. 2023 Jul;44(7):457-473. IF: 13.8 (D1).
- Peyman M, Barroso E, Turcu AL, Estrany F Jr, Smith D, Jurado-Aguilar J, Rada P, Morisseau C, Hammock BD, Valverde ÁM, Palomer X, Galdeano C, Vázquez S, Vázquez-Carrera M. Soluble epoxide hydrolase-targeting PROTAC activates AMPK and inhibits endoplasmic reticulum stress. Biomed Pharmacother. 2023 Dec;168:115667. IF: 7.5 (D1).
- Zhang M, Barroso E, Ruat M, Peña L, Peyman M, Aguilar-Recarte D, Montori-Grau M, Rada P, Cugat C, Montironi C, Zarei M, Jurado-Aguilar

J, Camins A, Balsinde J, Valverde ÁM, Wahli W, Palomer X, Vázquez-Carrera M. Elafibranor upregulates the EMT-inducer S100A4 via PPAR δ . Biomed Pharmacother. 2023 Nov;167:115623. IF: 7.5 (D1).

- Aguilar-Recarte D, Barroso E, Zhang M, Rada P, Pizarro-Delgado J, Peña L, Palomer X, Valverde ÁM, Wahli W, Vázquez-Carrera M. A positive feedback loop between AMPK and GDF15 promotes metformin antidiabetic effects. Pharmacol Res. 2023 Jan;187:106578. IF: 9.3 (D1).

SELECTED PROJECTS

- Activación de la vía PPAR β /d-GDF15 para el tratamiento de la resistencia a la insulina y la diabetes mellitus Tipo 2. PID2021-1221160B-I00. Ministerio de Ciencia e Innovación (MICINN). Convocatoria 2021 de ayudas a «Proyectos de Generación de Conocimiento». Modalidad Proyectos de «Investigación Orientada». 2022-2025. IP: Manuel Vazquez Carrera.
- Dianes Farmacològiques en Inflamació i Malalties Metabòliques. 2021 SGR 01106. Agència de Gestió d'Ajuts Universitaris i de Recerca (AGAUR). Ajuts per donar suport a l'activitat científica dels grups de recerca de Catalunya (SGR-Cat 2021). 2022-2025. IP: Manuel Vazquez Carrera.

Allergic Diseases in Childhood and Adolescence

Montserrat Alvaro Lozano, MD, PhD

4
PROJECTS
3 competitive projects
1 non competitive projects

14
CLINICAL TRIALS &
OBSERVATIONAL STUDIES
12 commercial clinical trials
2 academic clinical trials

7
PUBLICATIONS

41
IMPACT FACTOR
5.9 average impact factor

71%
PUBLICATIONS IN 1ST QUARTILE

57%
PUBLICATIONS IN 1ST DECILE

14%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Rosa Jiménez Feijoo, Jaime Lozano Blasco, Adrianna Machinena Spera, Maria del Mar Folqué Giménez, Mónica Piquer Gibert

PHD STUDENTS

Olga Dominguez Sánchez

During 2023 our team has participated in many clinical trials in the area of food allergy, respiratory allergy and chronic urticaria. Most of them are international trials with the participation of children and adolescents. Other studies have been developed by us in the same fields and also in relation with drug allergy and atopic dermatitis.

Our group has also published some works in well known and indexed scientific magazines.

We have been awarded as an "ANACARE Center of Excellence" for

the treatment of children with food allergies and anaphylaxis and as a "UCARE Center of Excellence" for the advances in the treatment of children with chronic urticaria. Both conditions have led us to publications and research in these matters.

We are also conducting some studies for which we have been awarded with national grants.

Our main areas of interest are food allergy and the development of new treatments which are important so our children are protected from anaphylaxis and death. In this field our group is the leader in conducting and participating in studies all through Spain and Europe. Immunotherapy for food allergy and the use of biologics either in the field of food allergy, asthma and chronic urticarial are also in our research interest. In fact one of our members is an author for the most important international guidelines in the diagnosis and treatment of food allergies.

SELECTED PUBLICATIONS

- Santos AF, Riggioni C, Agache I, Akdis CA, Akdis M, Alvarez-Perea A, Alvaro-Lozano M, Ballmer-Weber B, Barni S, Beyer K, Bindslev-Jensen C, Brough HA, Buyuktiryaki B, Chu D, Del Giacco S, Dunn-Galvin A, Eberlein B, Ebisawa M, Eigenmann P, Eiwegger T, Feeney M, Fernandez-Rivas M, Fisher HR, Fleischer DM, Giovannini M, Gray C, Hoffmann-Sommergruber K, Halken S, Hourihane JO, Jones CJ, Jutel M, Knol E, Konstantinou GN, Lack G, Lau S, Marques Mejias A, Marchisotto MJ, Meyer R, Mortz CG, Moya B, Muraro A, Nilsson C, Lopes de Oliveira LC, O'Mahony L, Papadopoulos NG, Perrett K, Peters RL, Podesta M, Poulsen LK, Roberts G, Sampson HA, Schwarze J, Smith P, Tham EH, Untersmayr E, Van Ree R, Venter C, Vickery BP, Vlieg-Boerstra B, Werfel T, Worm M, Du Toit G, Skypala I. EAACI guidelines on the diagnosis of IgE-mediated food allergy. Allergy. 2023 Dec;78(12):3057-3076. FI:12.4 (D1)
- Staubach P, Alvaro-Lozano M, Sekerel BE, Maurer M, Ben-Shoshan M, Porter M, Hua E, Ji Y, Burciu A, Savelieva M, Severin T, Drollmann A, Bieniczak A. Ligelizumab in adolescents with chronic spontaneous urticaria:

Results of a dedicated phase 2b randomized clinical trial supported with pharmacometric analysis. *Pediatr Allergy Immunol.* 2023 Jul;34(7):e13982. FI:4.4 (D1)

- Nieto-Cid M, Garriga-Baraut T, Plaza-Martín AM, Tortajada-Girbés M, Torres-Borrego J, Lozano-Blasco J, Moreno-Galarraga L, Del Mar Folqué-Giménez M, Bosque-García M, Gaboli M, López-Neyra A, Rivas-Juegas C, Caballero-Rabasco MA, Freixa-Benavente A, Valdesoiro-Navarrete L, de Mir-Messa I, Ballester-Asensio E, Penín-Antón M, Romero-García R, Navarro-Morón J, Valenzuela-Soria A, Sánchez-Mateos M, Batlles-Garrido J, Sanz-Santiago V, de Atauri ÁG, Andrés-Martín A, Campos-Alonso E, Gómez-Pastrana D, Vázquez-Rodríguez E, Martínez-Pardo L, Del Río-Camacho G, Mazón-Ramos Á, Nieto-García A. Cost-effectiveness of omalizumab for the treatment of severe pediatric allergic asthma-Results of a real-life study in Spain. *Pediatr Allergy Immunol.* 2023 Apr;34(4):e13942. FI:4.4 (D1)
- Sandoval-Ruballos M, Domínguez O, Ortiz de Landazuri I, Gereda D, Espinoza Cisneros M, Quesada Sequeira F, Ramos Rodríguez SM, Machinena A, Piquer M, Folqué MDM, Lozano J, Jiménez-Feijoo R, Pascal M, Alvaro-Lozano M. Pediatric chronic urticaria: Clinical and laboratory characteristics and factors linked to remission. *Pediatr Allergy Immunol.* 2023 Mar;34(3):e13929. FI:4.4 (D1)

SELECTED PROJECTS

- Desensibilizado versus arreactivo persistente: Análisis a largo plazo de la eficacia de la inmunoterapia oral específica a alimentos en niños con alergia a leche de vaca y huevo. Sociedad Española de Inmunología Clínica, Alergia y Asma Pediátrica. 2018-2024. IP: Montserrat Alvaro Lozano.
- Genetics of Food Allergy Barcelona Montserrat Alvaro. Max Delbrück Centrum für Molekulare medizin Berlin-Buch. 2021-2024. IP: Montserrat Alvaro Lozano.
- Estudio de hipersensibilidad a anti-inflamatorios no esteroideos (AINES) en población pediátrica. Sociedad Española de Inmunología Clínica, Alergia y Asma Pediátrica. 2022-2024. IP: Adrianna Machinena

Spera.

- Estudio de la alergenidad de diferentes proteínas en la alergia alimentaria a pescado en población pediátrica. Sociedad Española de Inmunología Clínica, Alergia y Asma Pediátrica. 2021-2025. IP: Olga Dominguez Sánchez.

SELECTED CLINICAL STUDIES

- Estudio de 52 semanas, aleatorizado, doble ciego, controlado con placebo, de grupos paralelos, multicéntrico sobre la eficacia y seguridad de la terapia adyuvante de GSK3511294 en participantes adultos y adolescentes con asma grave no controlada con un fenotipo eosinofílico. A 52-week, randomised, double-blind, placebo-controlled, parallel-group, multi-centre study of the efficacy and safety of GSK3511294 adjunctive therapy in adult and adolescent participants with severe uncontrolled asthma with an eosinophilic phenotype. (213744). Esteve; Farmacia; Glaxosmithkline, S.A; IQVIA RDS Spain SL; Laboratori HSJD; Servicio de Cardiología; Servicio Pneumología; Unitat de Recerca (URC) FSJD. 2021. IP: Jaime Lozano Blasco.
- A three-year, multi-center, double-blind, extension study to evaluate the long-term safety and efficacy of ligelizumab in patients who completed ligelizumab's Phase III studies in food allergy. (CQGE031G12303B). Balcél; Farmacia; Laboratori HSJD; Novartis Farmacéutica, S.A; Servicio de Cardiología; Servicio Pneumología; Unitat de Recerca (URC) FSJD. 2023. IP: Montserrat Alvaro Lozano.
- A Multicentre, Randomised, Double-blind, Parallel Group, Placebo-controlled, Time-to-first Asthma Exacerbation Phase III Efficacy and Safety Study of Benralizumab in Paediatric Patients with Severe Eosinophilic Asthma (DOMINICA). (D3250C00024). AstraZeneca AB; Farmacia; Laboratori HSJD; León; Parexel International (IRL) Ltd.; Servicio de Cardiología; Unitat de Recerca (URC) FSJD. 2023. IP: Jaime Lozano Blasco.
- Ensayo clínico de eficacia y seguridad, prospectivo, aleatorizado, doble ciego, de doble simulación, controlado con placebo,

multicéntrico, con inmunoterapia en pacientes con asma de leve a moderada controlada y rinitis/ rinoconjuntivitis, alérgicos a Dermatophagoides pteronyssinus y/o Dermatophagoides farinae. (MM09-SIT-040). BioClever 2005, SLU; Escoruela; Farmacia; Inmunotek, .SL.; Laboratori HSJD; Servicio Pneumología; Unitat de Recerca (URC) FSJD. 2022. IP: Adrianna Machinena Spera.

Study of Immune Deficiency Diseases in Paediatrics (GEMDIP)

**Laia Alsina Manrique de Lara, MD, PhD
& Jordi Antón López, MD, PhD**



24
PROJECTS
19 competitive projects
5 non competitive projects



32
CLINICAL TRIALS &
OBSERVATIONAL STUDIES
26 commercial clinical trials
6 academic clinical trials



22
PUBLICATIONS



516
IMPACT FACTOR
23.4 average impact factor



59%
PUBLICATIONS IN 1ST QUARTILE



32%
PUBLICATIONS IN 1ST DECILE



27%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Alessandra Magnani, Violeta Bittermann, Juan Manuel Mosquera Angarita, Ana M^a Plaza Martín, Andrea Zacarias Crovato, Rosa Bou i Torrent, Àngela Deyà Martínez, Estibaliz Iglesias Jimenez, Judith Sánchez Manubens

POSTDOC ASSOCIATED RESEARCHERS

Anna Esteve Solé

PHD STUDENTS

Yiyi Luo, Joan Calzada Hernández

ASSISTANT RESEARCHER

Daniel Acevedo Gómez, Laura Martínez Mifsut, Lucia Rodríguez Díez, Clara Isabel Giménez Roca

COLLABORATOR RESEARCHER

Jordi Yagüe Ribes, Manel Juan Otero, Juan Ignacio Arostegui Gorospe

During 2023 there have been major achievements in the GEMDIP group, designed by AGAUR as consolidated

research group in January 2022. Our team has been awarded several competitive national and international grants, that reinforce our research in the improved diagnosis and novel target treatment identification for our paediatric patients with immune pathology. The group has led significant publications related to MIS-C, Kawasaki, juvenile idiopathic arthritis, childhood uveitis, pediatric systemic sclerosis and autoinflammatory diseases, as well as novel immunomodulatory treatments and their effect, as well as characterization of normal immune development during childhood.

In the area of Rheumatology and autoinflammatory diseases, we are leading research in uveitis unmet needs in the study PAVE (European Joint program in Rare Diseases), and participate in the ongoing international competitive grant IMMUNAID (H2020).

In the area of clinical immunology and primary immunodeficiencies, we are actively participating in a trial for WHIM syndrome, we are pursuing our research in immune dysregulatory disorders (multiple or autoimmunity, lymphoproliferation or inflammation) centered on Treg, as well as the study of newborns born to mother exposed to biologicals during pregnancy.

In the area of advanced therapies we have been included in the Spanish network named CERTERA with SJD's new developed GMP facilities for lentiviral and cellular therapies production.

SELECTED PUBLICATIONS

- Ramanan AV, Quartier P, Okamoto N, Foeldvari I, Spindler A, Fingerhutová Š, Antón J, Wang Z, Meszaros G, Araújo J, Liao R, Keller S, Brunner HI, Ruperto N; JUVE-BASIS investigators; Paediatric Rheumatology International Trials Organisation. Baricitinib in juvenile idiopathic arthritis: an international, phase 3, randomised, double-blind, placebo-controlled, withdrawal, efficacy, and safety trial. *Lancet*. 2023 Aug 12;402(10401):555-570. FI:168.9 (D1)
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- Lee D, Le Pen J, Yatim A, Dong B, Aquino Y, Ogishi M, Pescarmona R, Talouarn E, Rinchai D, Zhang P, Perret M, Liu Z, Jordan I, Elmas Bozdemir S, Bayhan GI, Beaufils C, Bizien L, Bisiaux A, Lei W, Hasan M, Chen J, Gaughan C, Asthana A, Libri V, Luna JM, Jaffré F, Hoffmann HH, Michailidis E, Moreews M, Seeleuthner Y, Bilguver K, Mane S, Flores C, Zhang Y, Arias AA, Bailey R, Schlüter A, Milisavljevic B, Bigio B, Le Voyer T, Materna M, Gervais A, Moncada-Velez M, Pala F, Lazarov T, Levy R, Neehus AL, Rosain J, Peel J, Chan YH, Morin MP, Pino-Ramirez RM, Belkaya S, Lorenzo L, Anton J, Delafontaine S, Toubiana J, Bajolle F, Fumadó V, DeDiego ML, Fidouh N, Rozenberg F, Pérez-Tur J, Chen S, Evans T, Geissmann F, Lebon P, Weiss SR, Bonnet D, Duval X; CoV-Contact Cohort§; COVID Human Genetic Effort¶; Pan-Hammarström Q, Planas AM, Meyts I, Haerynck F, Pujol A, Sancho-Shimizu V, Dalgard CL, Bustamante J, Puel A, Boisson-Dupuis S, Boisson B, Maniatis T, Zhang Q, Bastard P, Notarangelo L, Béziat V, Perez de Diego R, Rodriguez-Gallego C, Su HC, Lifton RP, Jouanguy E, Cobat A, Alsina L, Keles S, Haddad E, Abel L, Belot A, Quintana-Murci L, Rice CM, Silverman RH, Zhang SY, Casanova JL. Inborn errors of OAS-RNase L in SARS-CoV-2-related multisystem inflammatory syndrome in children. *Science*. 2023 Feb 10;379(6632):eabo3627. FI:56.9 (D1)
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Marco FM, Pignata C, Bousfiha A, Turvey SE, Bauer S, Haerynck F, Oejo-Vinyals JG, Lendinez F, Prader S, Naumann-Bartsch N, Pachlopnik Schmid J, Biggs CM, Hildebrand K, Dreesman A, Cárdenes MÁ, Ailal F, Benhsaien I, Giardino G, Molina-Fuentes A, Fortuny C, Madhavarapu S, Conway DH, Prando C, Schidlowski L, Martínez de Saavedra Álvarez MT, Alfaro R, Rodríguez de Castro F; ESID Registry Working Party; COVID Human Genetic Effort; Meyts I, Hauck F, Puel A, Bastard P, Boisson B, Jouanguy E, Abel L, Cobat A, Zhang Q, Casanova JL, Alsina L, Rodríguez-Gallego C. Humans with inherited MyD88 and IRAK-4 deficiencies are predisposed to hypoxemic COVID-19 pneumonia. *J Exp Med.* 2023 May 1;220(5):e20220170. FI:15.3 (D1)

SELECTED PROJECTS

- ImmunAID_Immunome project consortium for AutoInflammatory Disorders. [779295]. European Commission. 2018-2024. IP: Jordi Antón López.
- Estudio del efecto de la exposición intrauterina a fármacos anti-TNF- α en el sistema inmunitario de hijos de madres con enfermedades reumáticas inflamatorias. (PI18/00223). Instituto de Salud Carlos III (ISCIII). 2019-2023. IP: Laia Alsina Manrique de Lara.
- Caracterización de las alteraciones inmunológicas, específicamente en células Treg, implicadas en la desregulación inmune en pacientes pediátricos con errores innatos de inmunidad, hacia terapias dirigidas. (PI21/00211). Instituto de Salud Carlos III (ISCIII). 2022-2024. IP: Laia Alsina Manrique de Lara.
- Producing an Arthritis Value-Framework with Economic Evidence. (AC21_2/00034). Instituto de Salud Carlos III (ISCIII). 2022-2024. IP: Jordi Antón López.

SELECTED CLINICAL STUDIES

- An Open-label Multiple Dose Study to Evaluate the Pharmacokinetics, Safety, and Tolerability of Upadacitinib in Pediatric Subjects with Polyarticular Course Juvenile Idiopathic Arthritis. (M15-340). AbbVie Deutschland GmbH & Co. KG "AbbiVie"; Farmacia; Jurado;

Laboratori HSJD; Sastre; Unitat de Recerca (URC) FSJD. 2019. IP: Jordi Antón López.

- A Phase 3, Randomized, Double-Blind, Placebo-Controlled, Multicenter Study of Mavorixafor in Patients with WHIM Syndrome with Open-Label Extension. (X4P-001-103). Farmacia; González; Hospitalización HSJD; Servicio de Audiovisuales; Servicio de Cardiología; Servicio Dermatología; Servicio Oftalmología; Syneos Health UK Limited; Unitat de Recerca (URC) FSJD; X4 Pharmaceuticals Inc. 2020. IP: Laia Alsina Manrique de Lara.
- Multi-centre, randomised, open-label, blinded endpoint assessed, trial of corticosteroids plus intravenous immunoglobulin (IVIg) and aspirin, versus IVIg and aspirin for prevention of coronary artery aneurysms in Kawasaki disease. (KD-CAAP [C4C]). Farmacia; Laboratori HSJD; Servicio de Cardiología; Unitat de Recerca (URC) FSJD; University College London. 2022. IP: Jordi Antón López.
- ESTUDIO FASE II, MULTICÉNTRICO, ALEATORIZADO, DOBLE CIEGO Y CONTROLADO CON PLACEBO PARA EVALUAR LA EFICACIA, SEGURIDAD Y FARMACOCINÉTICA DE OBINUTUZUMAB EN PACIENTES ADOLESCENTES CON NEFRITIS LÚPICA DE CLASE III O IV ACTIVA. A phase II, randomized, double-blind, placebo-controlled, multicenter study to evaluate the efficacy, safety, and pharmacokinetics of obinutuzumab in adolescent patients with active class III or IV lupus nephritis. (WA42985). Farmacia; Garcia; Roche Farma, S.A; Servicio de Cardiología; Talens; Unitat de Recerca (URC) FSJD. 2021. IP: Jordi Antón López.

Infectious Diseases and Microbiome

Carmen Muñoz Almagro, MD, PhD

17 PROJECTS
10 competitive projects
7 non competitive projects

16 CLINICAL TRIALS & OBSERVATIONAL STUDIES
12 commercial clinical trials
4 academic clinical trials

30 PUBLICATIONS

296 IMPACT FACTOR
9.9 average impact factor

77% PUBLICATIONS IN 1ST QUARTILE

33% PUBLICATIONS IN 1ST DECILE

33% LEADERSHIP

MEMBERS

STAFF RESEARCHER

Clàudia Fortuny Guasch, Maria Goretti López Ramos, Antoni Noguera Julián, Pedro Brotons de los Reyes, Victoria Fumadó, Juan José García García, Cristian Launes Montaña, Silvia Simó Nebot

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TECHNICIAN

Carles Cisneros Camps, Amaresh Pérez Argüello, Jesica Saucedo Verdes

COLLABORATOR RESEARCHER

Emilia Sánchez Ruiz

During 2023, our team has been awarded with six new competitive public and private grants that have contributed to strengthening the three main research lines of our group: 1) improvement of infectious diseases diagnostics; 2) human microbiome; and 3) microbial genomics and pathogen surveillance. With regard to improvement of infectious diseases diagnostics we have been awarded with two projects: a FIS-funded project about the use machine learning algorithms and big data for prediction and rapid detection of sepsis in the Emergency Department and the Pediatric Intensive Care Unit and a CIBER network-funded project about the use of clinical metagenomics for diagnosis of fulminant hepatitis in children. With respect to the human microbiome we have started a new project that analyzes the long- and short-term resilience of the nasopharyngeal microbiota and its implication in the transition from colonization to invasive disease by known and unknown pathobionts bacteria. In relation to microbial genomics and pathogen molecular surveillance we have been granted with two projects: a first project funded by the Precision Personalized Medicine Research Projects of the CIBER network that analyzes the role of the human microbiome in predicting the response to anti-GD2 immunotherapy treatment in pediatric patients with neuroblastoma; and a second project about the role of bacteriophages in the pathogenesis of invasive pneumococcal disease and their potential therapeutic use. Of note, 7 pre- and post-doctoral researchers of the group continue to be contracted thanks to the competitive funds of the research projects awarded during 2023 and previous years.

With respect to dissemination activities, we have published more than 40 publications, participated in 15 clinical trials and led two doctoral theses that have been awarded with the maximum qualification.

SELECTED PUBLICATIONS

- Henares D, Lo SW, Perez-Argüello A, Redin A, Ciruela P, García-García JJ, Brotons P, Yuste J, Sá-Leão R, Muñoz-Almagro C. Comparison of next generation technologies

and bioinformatics pipelines for capsular typing of *Streptococcus pneumoniae*. J Clin Microbiol. 2023 Dec 19;61(12):e0074123. FI:9.4 (Q1)

- Shaw D, Abad R, Amin-Chowdhury Z, Bautista A, Bennett D, Broughton K, Cao B, Casanova C, Choi EH, Chu YW, Claus H, Coelho J, Corcoran M, Cottrell S, Cunney R, Cuypers L, Dalby T, Davies H, de Gouveia L, Deghmane AE, Demczuk W, Desmet S, Domenech M, Drew R, du Plessis M, Duarte C, Erlendsdóttir H, Fry NK, Fuursted K, Hale T, Henares D, Henriques-Normark B, Hilty M, Hoffmann S, Humphreys H, Ip M, Jacobsson S, Johnson C, et al. Trends in invasive bacterial diseases during the first 2 years of the COVID-19 pandemic: analyses of prospective surveillance data from 30 countries and territories in the IRIS Consortium. Lancet Digit Health. 2023 Sep;5(9):e582-e593. FI:30.8 (D1)
- Hernández-García M, Bassat Q, Fumado V, Rodas G, Pi R, Miranda-García M, Girona-Alarcón M, Català M, Alonso S, Alvarez-Lacalle E, López D, Melé-Casas M, Pons-Tomas G, F de Sevilla M, Bonet-Carne E, Fortuny C, García-Miquel A, Jou C, Adroher C, Claverol J, Cubells M, Codina A, Cuadras D, Gratacós E, Brotons P, Muñoz-Almagro C, Prats C, García-García JJ, Jordan I. SARS-CoV-2 transmission in teenagers and young adults in Fútbol Club Barcelona's Multidisciplinary Sports Training Academy. Eur J Pediatr. 2023 May;182(5):2421-2432. FI:3.6 (Q1)
- López-Ramos MG, Vinent J, Aarnoutse R, Colbers A, Velasco-Arnaiz E, Martorell L, Falcón-Neyra L, Neth O, Prieto L, Guillén S, Baquero-Artigao F, Méndez-Echevarría A, Gómez-Pastrana D, Jiménez AB, Lahoz R, Ramos-Amador JT, Soriano-Arandes A, Santiago B, Farré R, Fortuny C, Soy D, Noguera-Julian A. Adequacy of the 10 mg/kg Daily Dose of Antituberculosis Drug Isoniazid in Infants under 6 Months of Age. Antibiotics (Basel). 2023 Jan 30;12(2):272. FI:4.8 (Q1)

SELECTED PROJECTS

- "Estudio de la capacidad de resiliencia a largo y corto plazo de la microbiota nasofaríngea

y su implicación en el paso de la colonización a enfermedad invasiva por bacterias patobiontes". PFIS0385. Instituto de Salud Carlos III (ISCIII). 2024-2026. IP: Carmen Muñoz Almagro.

- Nuevos biomarcadores para el diagnóstico y la monitorización de la respuesta al tratamiento de la tuberculosis pediátrica. PFIS0356. Instituto de Salud Carlos III (ISCIII). 2023-2025. IP: Antoni Noguera Julián.
- Vaccine Effectiveness, Burden and Impact Studies (VEBIS) of COVID-19 and Influenza. PCP00340. European Centre for Disease Prevention and Control (ECDC). 2021-2024. IP: Carmen Muñoz Almagro.
- Laboratorio de soporte para la vigilancia epidemiológica molecular de *Streptococcus pneumoniae*, *Bordetella pertussis* y *Neisseria meningitidis*. PFP00065. Agència de Salut Pública de Catalunya. 2011-. IP: Carme Muñoz Almagro.

SELECTED CLINICAL STUDIES

- Estudio fase III, aleatorizado, doble ciego, controlado con placebo y multicéntrico para demostrar la eficacia de una dosis única de la vacuna materna no adyuvada frente al Virus Respiratorio Sincitial (VRS), administrada por vía IM a gestantes de 18 a 49 años de edad, para la prevención de la Enfermedad vías respiratorias Bajas (EVRB) asociada al VRS en sus hijos hasta los 6 meses de edad. (212171 (RSV MAT-009)). Farmacia; Glaxosmithkline, S.A.; Jiménez; Unitat de Recerca (URC) FSJD. 2021. IP: Victoria Fumadó.
- Estudio de Fase I abierto y de búsqueda de dosis, para evaluar la seguridad, la tolerabilidad e inmunogenicidad y de Fase II/III, controlado con placebo y ciego para el observador, para evaluar la seguridad, tolerabilidad e inmunogenicidad de una vacuna experimental de ARN del SARS-COV-2 contra la COVID-19 en niños. (C4591007). BioNTech SE; Farmacia; ICON Clinical Research, Ltd; Pfizer, S.L.U.; Unitat de Recerca (URC) FSJD. 2021. IP: Clàudia Fortuny Guasch.
- Phase 3, Multicenter, Randomized, Partially Blinded, Palivizumab-Controlled Study to Evaluate the Safety, Efficacy, and

Pharmacokinetics of MK-1654 in Infants and Children at Increased Risk for Severe RSV Disease. (MK-1654-007). Farmacia; Jiménez; Merck, Sharp & Dohme de España. S.A; Unitat de Recerca (URC) FSJD; Ventosa. 2021. IP: Clàudia Fortuny Guasch.

- Ensayo en Fase III, aleatorizado, doble ciego para evaluar la seguridad de una vacuna conjugada antineumococica 20-Valente en lactantes sanos a Phase 3, randomized, partial double-blind trial to evaluate the safety and immunogenicity of 20-valent pneumococcal conjugate vaccine in healthy children ≥ 12 to < 24 months of age with two prior infant doses of Prevenar 13. (B7471027). Farmacia; Gonzalez; ICON Clinical Research, Ltd; Koncalova; Pfizer Inc.; Unitat de Recerca (URC) FSJD. 2022. IP: Antoni Noguera Julián.

Gut Microbiota

Laura Baldomà Llavínés, PhD

0
CLINICAL TRIALS &
OBSERVATIONAL STUDIES

6
PUBLICATIONS

32
IMPACT FACTOR
5.3 average impact factor

67%
PUBLICATIONS IN 1ST QUARTILE

0%
PUBLICATIONS IN 1ST DECILE

33%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Josefa Badia Palacin

The association between gut microbiota and health encourages the design of new microbiome-oriented therapeutic and nutritional strategies to restore intestinal homeostasis. The most recent component of the biotics strategy refers to postbiotics, a term that encompasses bioactive compounds produced by probiotics. EVs from beneficial gut microbes are good candidates to be explored for this potential application. In this context, the main contributions of our group during 2023 were centered on new activities associated to microbiota/probiotic EVs that foresee their potential application in two pathophysiological conditions.

(1) To ameliorate highly prevalent leaky gut-associated diseases, such as IBD. TFF3 is a potential target in IBD. Expression of TFF3 is downregulated in damaged tissue of IBD patients, whereas its regulator miR-7-5p is overexpressed. We have shown that EVs from the probiotic EcN activate TFF3 production and downregulate miR7-5-p in goblet cells. High levels of secreted TFF3 stimulate wound healing and repair of damaged intestinal epithelium.

(2) In the preclinical model of neonatal rats, interventions with EVs from probiotic/microbiota *E. coli* have been revealed as a safe postbiotic strategy to stimulate immunity and intestinal maturation in early life. Among the multiple benefits, EVs: (i) prime the newborn immune system at the cellular and humoral level to better fight against infections, (ii) preserve tolerance against the gut microbiota, (iii) elicit anti-inflammatory effects in the small intestine, (iv) promote intestinal maturation and trophic effects on the developing intestine by increasing the villi area. This is the first preclinical assay evaluating the positive effects of microbiota/probiotic EVs in early life and provides the molecular basis for the design of postbiotic strategies to prevent infections and help intestinal maturation in pre-term newborns, and to ameliorate allergies in infants or later in life. Clinical trials are needed to prove the benefits and safety of microbiota EV-based interventions in human infants.

SELECTED PUBLICATIONS

- Bustos-Salgado P, Domínguez-Villegas V, Andrade-Carrera B, Mallandrich M, Calpena A, Domènech O, Martínez-Ruiz S, Badía J, Baldomà L, Gómez de Aranda I, Blasi J, Garduño-Ramírez ML. PLGA Nanoparticles Containing Natural Flavanones for Ocular Inflammation. *Pharmaceutics*. 2023 Dec 11;15(12):2752. FI:5.4 (Q1)
- Folle C, Marqués AM, Mallandrich M, Suñer-Carbó J, Halbaut L, Sánchez-López E, López-Machado AL, Díaz-Garrido N, Badía J, Baldoma L, Espina M, García ML, Calpena AC. Colloidal hydrogel systems of thymol-loaded PLGA nanoparticles designed for acne treatment. *Colloids Surf B Biointerfaces*. 2024 Feb;234:113678. FI:5.8 (Q1)
- Martínez-Ruiz S, Sáez-Fuertes L, Casanova-Crespo S, Rodríguez-Lagunas MJ, Pérez-Cano FJ, Badía J, Baldoma L. Microbiota-Derived Extracellular Vesicles Promote Immunity and Intestinal Maturation in Suckling Rats. *Nutrients*. 2023 Nov 6;15(21):4701. FI:5.9 (Q1)
- Olivo-Martínez Y, Bosch M, Badía J, Baldomà L. Modulation of the Intestinal Barrier Integrity and Repair by Microbiota Extracellular Vesicles through the Differential


Regulation of Trefoil Factor 3 in LS174T Goblet Cells. *Nutrients*. 2023 May 24;15(11):2437. FI:5.9 (Q1)


SELECTED PROJECTS

- Estrategias postbióticas basadas en el microbioma intestinal: funciones reguladoras de vesículas de microbiota en la comunicación intestino-hígado e intestino-cerebro. PID2022-1371920B-I00. Ministerio de Ciencia e Innovación (MICINN). Convocatoria 2022 de ayudas a «Proyectos de Generación de Conocimiento». Modalidad Proyectos de «Investigación Orientada». 2023-2026. IP: Laura Baldoma Llavines.
- Evaluación de nuevas actividades de vesículas secretadas por probióticos y microbiota intestinal y su potencial aplicación como postbióticos en salud humana. PID2019-107327RB-I00. Ministerio de Ciencia, Innovación y Universidades. Convocatòria d'ajuts del Programa Estatal d'Investigació, Desenvolupament i Innovació Orientada als reptes de la Societat. Modalitat Reptes. 2020-2023. IP: Laura Baldoma Llavines.


Environment and Complications during Pregnancy

Maria Dolores Gómez Roig, MD, PhD


 **6** PROJECTS
5 competitive projects
1 non competitive projects

 **6** CLINICAL TRIALS & OBSERVATIONAL STUDIES
2 commercial clinical trials
4 academic clinical trials

 **17** PUBLICATIONS

 **90** IMPACT FACTOR
5.3 average impact factor

 **77%** PUBLICATIONS IN 1ST QUARTILE

 **35%** PUBLICATIONS IN 1ST DECILE

 **41%** LEADERSHIP

MEMBERS

STAFF RESEARCHER

Eduardo González Bosquet, Núria Lorente Colomé, Rosalia Pascal Capdevila, Silvia - Irena Ferrero Martínez, Joan Sabrià Bach

POSTDOC ASSOCIATED RESEARCHERS

Francesca Crovetto, Edurne Mazarico Gallego, Miriam Pérez Cruz

PHD STUDENTS

Diana Lucia Lip Sosa

ASSISTANT RESEARCHER

Concepción Muñoz Lozano, Natàlia Gorina Careta, Patricia Ferrer Aguilar, Laura Mallén Perez, Marc Josep Cahuana Bartra, Estela Rodriguez Quintero, Laura Almeida Toledano

During 2023, our research team continued the study on alcohol intake during pregnancy and prenatal intervention to assess the possibility of mitigating toxicity in the fetus and after birth. Within this line, we have expanded the study with the project "Influence of alcohol

consumption during pregnancy on placental and fetal genomics, and its impact on placental function and postnatal neurodevelopment," whose objective will be to determine whether prenatal alcohol consumption produces molecular changes in the placenta and/or umbilical cord blood, thus establishing a possible etiopathogenesis or correlation with fetal growth restriction and fetal alcohol spectrum disorders.

We continue with research on air pollution, exposure to harmful chemicals, and/or endocrine disruptors that are toxic factors affecting the fetus, working on identifying and understanding the effects on the baby, with the purpose of intervening and preventing during pregnancy, in the Barcelona Life Study Cohort (BISC), which is currently in the analysis phase.

We have also extended our studies on the effects of the Mediterranean diet and stress reduction during pregnancy in the IMPACT BCN cohort, whose goal is to improve fetal growth, reduce pregnancy complications, and enhance neurodevelopment and maternal-fetal health.

The group's research has led to numerous articles, obtaining scientific results with the aim of implementing acceptable interventions and generating recommendations, policies, and prevention strategies.

SELECTED PUBLICATIONS

- Ribas-Prats T, Cordero G, Lip-Sosa DL, Arenillas-Alcón S, Costa-Faidella J, Gómez-Roig MD, Escera C. Developmental Trajectory of the Frequency-Following Response During the First 6 Months of Life. *J Speech Lang Hear Res*. 2023 Dec 11;66(12):4785-4800. FI:2.6 (Q1)
- Andreu-Fernández V, Serra-Delgado M, Almeida-Toledano L, García-Meseguer À, Vieiros M, Ramos-Triguero A, Muñoz-Lozano C, Navarro-Tapia E, Martínez L, García-Algar Ó, Gómez-Roig MD. Effect of Postnatal Epigallocatechin-Gallate Treatment on Cardiac Function in Mice Prenatally Exposed to Alcohol. *Antioxidants (Basel)*. 2023 May 9;12(5):1067. FI:7 (D1)
- Ribas-Prats T, Arenillas-Alcón S, Pérez-Cruz M, Costa-Faidella J, Gómez-Roig MD, Escera C. Speech-

Encoding Deficits in Neonates Born Large-for-Gestational Age as Revealed With the Envelope Frequency-Following Response. *Ear Hear.* 2023 Jul-Aug 01;44(4):829-841. FI:3.7 (D1)

- Lip-Sosa DL, Pérez-Cruz M, Ahumada-Droguett P, Ribas-Prats T, Puertollano M, García-Gómez MA, Mazarico E, Eixarch E, Escera C, Gómez-Roig MD. Corpus callosum-fastigium and tectal lengths in late-onset small fetuses. *Ultrasound Obstet Gynecol.* 2023 Aug;62(2):226-233. FI:7.1 (D1)

SELECTED PROJECTS

- ATHLETE_Advancing Tools for Human Early Lifecourse Exposome research and Translation. (874583). European Commission. 2020-2025. IP: Maria Dolores Gómez Roig.
- Impacto de un programa de intervención prenatal estructurado, de disminución de consumo de alcohol, en los resultados perinatales y de neurodesarrollo. (PI20/00490). Instituto de Salud Carlos III (ISCIII). 2021-2025. IP: Maria Dolores Gómez Roig.
- Primary care interventions to prevent maternal and child chronic diseases of perinatal and developmental origin. (RD21/0012/0003). Instituto de Salud Carlos III (ISCIII). 2022-2024. IP: Maria Dolores Gómez Roig.
- Consumo de alcohol prenatal, marcadores moleculares, y su repercusión en la función placentaria y el neurodesarrollo. PFIS0387. Instituto de Salud Carlos III (ISCIII). 2024-2026. IP: Maria Dolores Gómez Roig.

SELECTED CLINICAL STUDIES

- Optimización del manejo antenatal de mujeres con amenaza de parto pretérmino mediante modelos de predicción: estudio clínico randomizado multicéntrico abierto (OPTIM-PTL). (OPTIM-PTL). Fundació de Recerca Clínic Barcelona-Institut d'Investigacions Biomèdiques August Pi i Sunyer. 2022. IP: Silvia - Irena Ferrero Martínez.
- Efficacy of low dose acetylsalicylic acid in preventing adverse maternal and perinatal outcomes in SARS-CoV-2 infected pregnant women. (SARA). Farmacia; Fundació Privada

ISGLOBAL; Laboratori HSJD; Serrano; Stöger. 2022. IP: Edurne Mazarico Gallego.

- Balón de Cook versus dinoprostona vaginal para la inducción del parto a término en la restricción del crecimiento intrauterino (estudio COLIGROW). (COLIGROW). Herraiz. 2023. IP: Edurne Mazarico Gallego.
- Desarrollo de un Algoritmo Diagnóstico de Amenaza de Parto Pretérmino que integra variables clínicas y la cuantificación de la consistencia cervical utilizando el dispositivo Fine Birth. (FB-CT1-P). Ultrasound-Innovation Medtech S.L. 2023. IP: Silvia - Irena Ferrero Martínez.

Cardiovascular Diseases and Child Development

Georgia Sarquella Brugada, MD, PhD & Joan Sánchez de Toledo, MD, PhD

20
PROJECTS
15 competitive projects
5 non competitive projects

7
CLINICAL TRIALS & OBSERVATIONAL STUDIES
6 commercial clinical trials
1 academic clinical trials

24
PUBLICATIONS

99
IMPACT FACTOR
4.1 average impact factor

33%
PUBLICATIONS IN 1ST QUARTILE

13%
PUBLICATIONS IN 1ST DECILE

50%
LEADERSHIP

MEMBERS

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Esther Aurenzanz Clemente, Jose Carlos Cruzalegui Gómez, Esther Carro Fernandez, Sergio César Diaz, Isaac Moll Adrian

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TECHNICIAN

Adrian Ortega Sanabria

SELECTED PUBLICATIONS

- García-García A, Pérez de Diego R, Flores C, Rinchai D, Solé-Violán J, Deyà-Martínez À, García-Solis B, Lorenzo-Salazar JM, Hernández-Brito E, Lanz AL, Moens L, Bucciol G, Almuqamam M, Domachowski JB, Colino E, Santos-Perez JL, Marco FM, Pignata C, Bousfiha A, Turvey SE, Bauer S, Haerynck

- F, Oejo-Vinyals JG, Lendinez F, Prader S, Naumann-Bartsch N, Pachlopnik Schmid J, Biggs CM, Hildebrand K, Dreesman A, Cárdenes MÁ, Ailal F, Benhsaien I, Giardino G, Molina-Fuentes A, Fortuny C, Madhavarapu S, Conway DH, Prando C, Schidowski L, Martínez de Saavedra Álvarez MT, Alfaro R, Rodríguez de Castro F; ESID Registry Working Party; COVID Human Genetic Effort; Meyts I, Hauck F, Puel A, Bastard P, Boisson B, Jouanguy E, Abel L, Cobat A, Zhang Q, Casanova JL, Alsina L, Rodríguez-Gallego C. Humans with inherited MyD88 and IRAK-4 deficiencies are predisposed to hypoxemic COVID-19 pneumonia. *J Exp Med*. 2023 May 1;220(5):e20220170. FI: 15.3 [D1]
- García-García A, Fortuny C, Fumadó V, Jordan I, Ruiz-López L, González-Navarro EA, Egri N, Esteve-Solé A, Luo Y, Vlaga A, Cabedo MM, Launes C, Soler A, Codina A, Juan M, Pascal M, Deyà-Martínez A, Alsina L. Acute and long-term immune responses to SARS-CoV-2 infection in unvaccinated children and young adults with inborn errors of immunity. *Front Immunol*. 2023 Jan 20;14:1084630. FI: 7.3 [Q1]
 - Cesar S, Campuzano O, Cruzalegui J, Fiol V, Moll I, Martínez-Barrios E, Zschaek I, Natera-de Benito D, Ortez C, Carrera L, Expósito J, Berruero R, Bautista-Rodríguez C, Dabaj I, Gómez García-de-la-Banda M, Quijano-Roy S, Brugada J, Nascimento A, Sarquella-Brugada G. Characterization of cardiac involvement in children with LMNA-related muscular dystrophy. *Front Cell Dev Biol*. 2023 Mar 10;11:1142937. FI: 5.5 [Q1]
 - Martínez-Barrios E, Grassi S, Brión M, Toro R, Cesar S, Cruzalegui J, Coll M, Alcalde M, Brugada R, Greco A, Ortega-Sánchez ML, Barberia E, Oliva A, Sarquella-Brugada G, Campuzano O. Molecular autopsy: Twenty years of post-mortem diagnosis in sudden cardiac death. *Front Med [Lausanne]*. 2023 Feb 10;10:1118585. FI: 3.9 [Q2]

SELECTED PROJECTS

- Creación de un programa personalizado de actividad física dirigida para deportistas de competición diagnosticados de un síndrome hereditario asociado a

- muerte súbita (PAC-SAMS). [74742]. Consejo Superior de Deportes; Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD; Martínez. 2023-2023. IP:Georgia Sarquella Brugada.
- A Prospective and Retrospective Registry and Biomarker Study to Evaluate the Natural History of Pediatric Patients with Cardiomyopathy due to MYBPC3 Mutations. [TN-201-0003]. ICON Clinical Research, Ltd; Pharm Research Associates (U.K) Limited; Tenaya Therapeutics. 2023. IP:Georgia Sarquella Brugada.
 - AI-CARDiology Advanced Monitoring cloud-BASED platform. [TED2021-132879B-I00]. Ministerio De Ciencia E Innovacion. 2022-2024. IP:Joan Sánchez de Toledo, Bart Bijnens
 - Válvula aórtica bicúspide y rigidez arterial: nuevos marcadores ecográficos predictores de disfunción vascular y arteriopatía. Sociedad Española de Cardiología Pediátrica y Cardiopatías Congénitas. 2022-2023. IP:Laia Brunet Garcia.

SELECTED CLINICAL STUDIES

- Multi-Center, Multi-National, Open-Label, Efficacy and Safety Study of Etripamil Nasal Spray in Pediatric Patients with Paroxysmal Supraventricular Tachycardia. [MSP-2017-1265(NODE-202)]. Farmacia; Laboratori HSJD; Marszalek; Milestone Pharmaceuticals inc.; Unitat de Recerca (URC) FSJD. 2023. IP:Georgia Sarquella Brugada.
- A Phase 2/3 Randomized, Placebo-Controlled, Double-blind, Clinical Study to Evaluate the Efficacy, Safety, and Pharmacokinetics of Vericiguat in Pediatric Participants with Heart Failure due to Systemic Left Ventricular Systolic Dysfunction (VALOR). Estudio clínico de fase 2/3, aleatorizado, doble ciego y controlado con placebo para evaluar la eficacia, la seguridad y la farmacocinética del Vericiguat en pacientes pediátricos con insuficiencia cardíaca debido a disfunción sistólica sistémica del ventrículo izquierdo (VALOR). [MK-1242-036]. Bhambi; Diagnóstico para la imagen (DXI); Farmacia; Laboratori HSJD; Merck, Sharp & Dohme de España. S.A; Sermes Planificación, S.L; Servicio Otorrinolaringología; Simonte;

Unitat de Recerca (URC) FSJD. 2023. IP:Sergio César Diaz.

- Estudio multicéntrico para evaluar la seguridad y tolerabilidad a largo plazo de un tratamiento en abierto con sacubitrilo/valsartán en pacientes pediátricos con insuficiencia cardíaca debida a disfunción sistólica de ventrículo izquierdo sistémico, que han completado el estudio CLCZ696B2319. [CLCZ696B2319E1 - Ext.]. Farmacia; Laboratori HSJD; Novartis Farmacéutica, S.A; Pozas; Unitat de Recerca (URC) FSJD. 2020. IP:Joan Sánchez de Toledo.
- Estudio multicéntrico, abierto, para evaluar la seguridad, tolerabilidad, farmacocinética y farmacodinámica de LCZ696, seguido de un estudio de 52 semanas, aleatorizado, doble ciego, paralelo controlado con fármaco activo para evaluar la eficacia y seguridad de LCZ696 comparado con enalapril en pacientes pediátricos con edades desde 1 mes hasta < 18 años, con insuficiencia cardíaca debida a disfunción sistólica del ventrículo izquierdo sistémico. [CLCZ696B2319]. Farmacia; Laboratori HSJD; Novartis Farmacéutica, S.A; Unitat de Recerca (URC) FSJD. 2019. IP:Joan Sánchez de Toledo.

Immune and Respiratory Dysfunction in Critically Ill Children

Francisco José Cambra Lasasa, MD, PhD



5
PROJECTS
5 competitive projects
0 non competitive projects



4
CLINICAL TRIALS &
OBSERVATIONAL STUDIES
4 commercial clinical trials
0 academic clinical trials



28
PUBLICATIONS



171
IMPACT FACTOR
6.1 average impact factor



50%
PUBLICATIONS IN 1ST QUARTILE



11%
PUBLICATIONS IN 1ST DECILE



39%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Anna Solé Ribalta, Carmina Guitart Pardellans, Mónica Balaguer Gargallo, Elisabeth Esteban Torne, Iolanda Jordán García, Martí Pons Òdena

ASSISTANT RESEARCHER

Vanessa Andriño Estrecha, Nuria Millán García del Real, Elena Fresán Ruiz, Sergio Benito Fernández, Lluïsa Hernández Platero, Carme Alejandre Galobardes, Patricia Corniero Alonso, Aida Felipe Villalobos, Susana Segura Matute, David Vila Pérez

COLLABORATOR RESEARCHER

Montserrat Esquerda Areste

TBD

Sergi Huerta Calpe, Monica Girona Alarcon, Ricardo Suárez Ramírez

In the scientific production of the Group during the year 2023, we

highlight the publication of 39 articles related to the Group's research lines in high impact journals. Four are highlighted in the corresponding section.

We highlight the active participation in the drafting of the Second International Guidelines for the Diagnosis and Management of Pediatric Acute Respiratory Distress Syndrome (PALICC-2).

During the year we have participated in four clinical trials, three of them in the field of infectious diseases, one on antibiotic treatment, A Phase 1, open-label, non-comparative, multicenter clinical trial to evaluate the safety, tolerability, and pharmacokinetics of ceftolozane/tazobactam (MK-7625A) in pediatric participants with nosocomial pneumonia and two that analyze antiviral drugs.

A Phase I/IIa Open-label Study in Infants with Respiratory Syncytial Virus Lower Respiratory Tract Infection, Followed by a Double-blind, Placebo-controlled Part, to Evaluate the Safety, Tolerability, Pharmacokinetics and Antiviral Effect of RV521 and another that evaluates the pharmacokinetics, safety and tolerability of treatment with IV zanamivir in neonates and infants younger than 6 months with confirmed complicated influenza infection.

The fourth trial is a study on the efficacy and safety of LJPC-501 in pediatric patients aged 2 to 17 years with catecholamine-resistant hypotension associated with distributive shock.

Continuation of projects already initiated ARDS in CHILDREN and ECMO initiation strategies impact on NEURO-DEVELOPMENT – ASCEND.

Use of electrical impedance tomography in the ventilation of patients with severe acute bronchiolitis.

And the implication of nasopharyngeal bacterial microbiota in the development of ventilator-associated pneumonia in the pediatric intensive care unit is being analyzed. MiNoReP study.

As a new project in the field of Artificial Intelligence research, we highlight the international project led by our unit with the participation of several European pediatric hospitals

entitled Development, validation and pilot testing of models based on machine learning for prediction and early detection of sepsis in pediatric emergency and intensive care units. PESERS 2.0 Project.

SELECTED PUBLICATIONS

- Lee D, Le Pen J, Yatim A, Dong B, Aquino Y, Ogishi M, Pescarmona R, Talouarn E, Rinchai D, Zhang P, Perret M, Liu Z, Jordan I, Elmas Bozdemir S, Bayhan GI, Beaufilets C, Bizien L, Bisiaux A, Lei W, Hasan M, Chen J, Gaughan C, Asthana A, Libri V, Luna JM, Jaffré F, Hoffmann HH, Michailidis E, Moreews M, Seeleuthner Y, Bilguvar K, Mane S, Flores C, Zhang Y, Arias AA, Bailey R, Schlüter A, Milisavljevic B, Bigio B, Le Voyer T, Materna M, Gervais A, Moncada-Velez M, Pala F, Lazarov T, Levy R, Neehus AL, Rosain J, Peel J, Chan YH, Morin MP, Pino-Ramirez RM, Belkaya S, Lorenzo L, Anton J, Delafontaine S, Toubiana J, Bajolle F, Fumadó V, DeDiego ML, Fidouh N, Rozenberg F, Pérez-Tur J, Chen S, Evans T, Geissmann F, Lebon P, Weiss SR, Bonnet D, Duval X; CoV-Contact Cohort§; COVID Human Genetic Effort¶; Pan-Hammarström Q, Planas AM, Meys I, Haerynck F, Pujot A, Sancho-Shimizu V, Dalgard CL, Bustamante J, Puel A, Boisson-Dupuis S, Boisson B, Maniatis T, Zhang Q, Bastard P, Notarangelo L, Béziat V, Perez de Diego R, Rodriguez-Gallego C, Su HC, Lifton RP, Jouanguy E, Cobat A, Alsina L, Keles S, Haddad E, Abel L, Belot A, Quintana-Murci L, Rice CM, Silverman RH, Zhang SY, Casanova JL. Inborn errors of OAS-RNase L in SARS-CoV-2-related multisystem inflammatory syndrome in children. *Science*. 2023 Feb 10;379(6632):eabo3627. FI:56.9 (D1)
- Lorenzo D, Esquerda M, Bofarull M, Cusi V, Roig H, Bertran J, Carrera J, Torralba F, Cambra FJ, Vila M, Garriga M, Palau F. The reuse of genetic information in research and informed consent. *Eur J Hum Genet*. 2023 Dec;31(12):1393-1397. FI: 5.2 (Q1)
- Emeriaud G, Pons-Òdena M, Bhalla AK, Shein SL, Killien EY, Modesto I Alapont V, Rowan C, Baudin F, Lin JC, Grégoire G, Napolitano N, Mayordomo-Colunga J, Diaz F, Cruces P, Medina A, Smith L, Khemani RG; Pediatric Acute

Respiratory Distress Syndrome Incidence and Epidemiology (PARDIE) Investigators and Pediatric Acute Lung Injury and Sepsis Investigators (PALISI) Network. Noninvasive Ventilation for Pediatric Acute Respiratory Distress Syndrome: Experience From the 2016/2017 Pediatric Acute Respiratory Distress Syndrome Incidence and Epidemiology Prospective Cohort Study. *Pediatr Crit Care Med.* 2023 Sep 1;24(9):715-726. FI:4.1 (Q1)

- Solé-Ribalta A, Balaguer M, Bobillo-Pérez S, Girona-Alarcón M, Guitart C, Esteban E, Jordan-García I. Quantification of a qualitative sepsis code: laying the foundations for the automation revolution. *Eur J Pediatr.* 2023 May;182(5):2169-2172. FI:3.6 (Q1)

SELECTED PROJECTS

- Implicación de la microbiota bacteriana de la nasofaringe en el desarrollo de neumonía asociada a ventilación mecánica en la unidad de cuidados intensivos pediátricos. Estudio MiNoReP. (PI19/00106). Instituto de Salud Carlos III (ISCIII). 2020-2024. IP:Iolanda Jordán García.
- ARDS in CHILDREN and ECMO initiation strategies impact on NEURO-DEVELOPMENT - ASCEND. (SUBK00015894). National Institute of Health (EEUU). 2020-2024. IP:Susana Segura Matute.
- Use of electrical impedance tomography in the ventilation of patients with severe acute bronchiolitis. European Society of Paediatric and Neonatal Intensive Care. 2022-2024. IP:Martí Pons Òdena.
- "Desarrollo, validación y prueba piloto de modelos basados en aprendizaje automático para predicción y detección temprana de sepsis en Urgencias e intensivos pediátricos. Proyecto PESERS 2.0". (PFIS0373). Instituto de Salud Carlos III (ISCIII). 2024-2026. IP:Iolanda Jordán García.

SELECTED CLINICAL STUDIES

- Estudio aleatorizado con control activo para comparar la eficacia y la seguridad de isoflurano inhalado administrado por el AnaConDa-S (dispositivo de conservación

de anestésico) el midazolam intravenoso para la sedación en pacientes pediátricos con ventilación mecánica de entre 3 y 17 años (menos de 18 años). A Randomised Active-controlled Study to Compare Efficacy and Safety of Inhaled Isoflurane Delivered by the AnaConDa-S (Anaesthetic Conserving Device) to Intravenous Midazolam for Sedation in Mechanically Ventilated Paediatric Patients 3 to 17 (Less than 18) Years Old. (SED002). Farmacia; Laboratori HSJD; LINK Medical Research; Sedana Medical AB; Unitat de Recerca (URC) FSJD. 2021. IP:Monica Girona Alarcon.

- A Phase I/IIa Open-label Study in Infants with REspiratory Syncytial VIRus Lower Respiratory Tract Infection, Followed by a Double-blind, Placebo-controlled Part, to Evaluate the Safety, Tolerability, Pharmacokinetics and Antiviral Effect of RV521. (REVC003). Chavez; Farmacia; Laboratori HSJD; ReViral Ltd; Syneos Health Clinical Spain S.L.U; Unitat de Recerca (URC) FSJD. 2021. IP:Iolanda Jordán García.



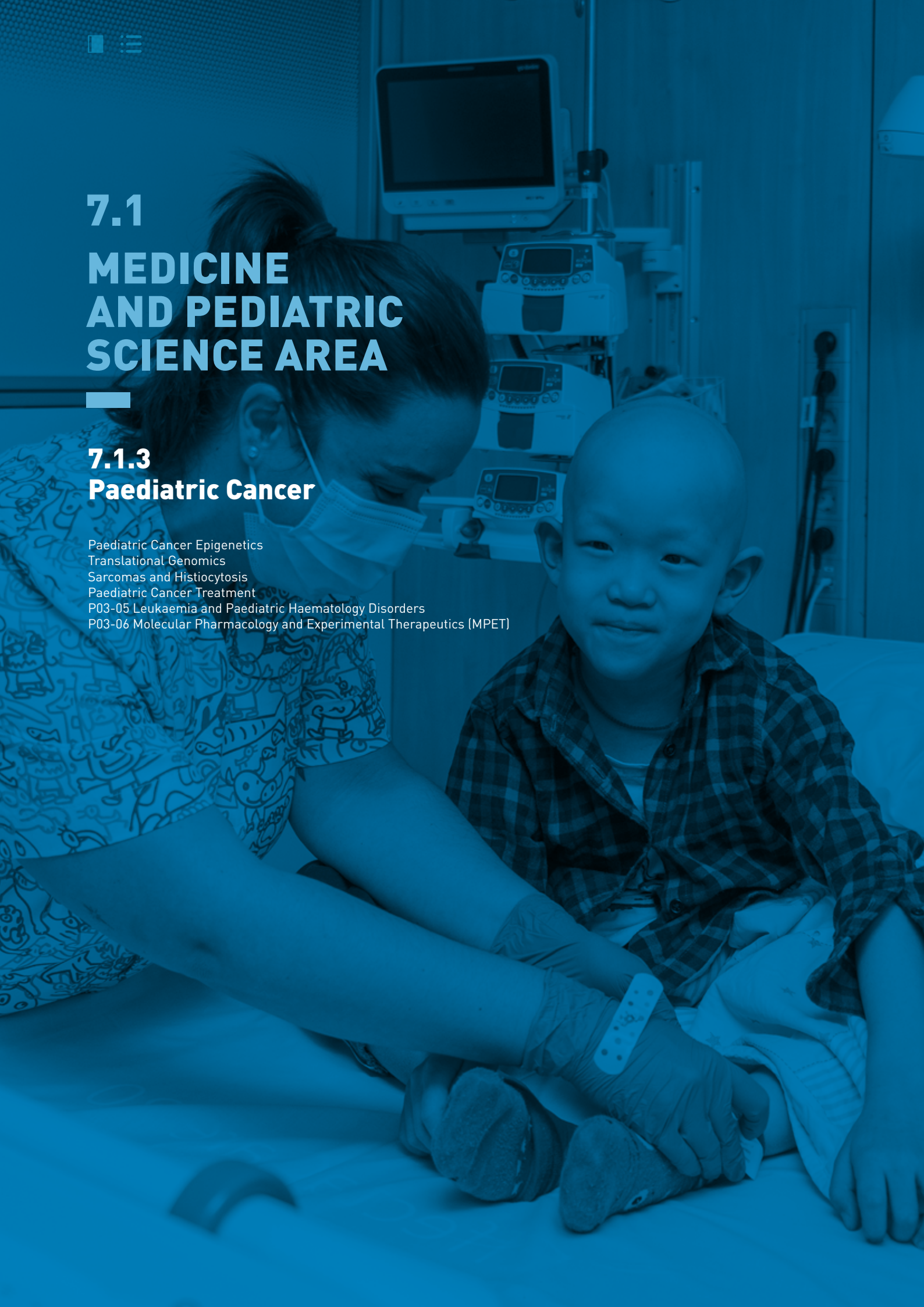
7.1

MEDICINE AND PEDIATRIC SCIENCE AREA

7.1.3

Paediatric Cancer

Paediatric Cancer Epigenetics
Translational Genomics
Sarcomas and Histiocytosis
Paediatric Cancer Treatment
P03-05 Leukaemia and Paediatric Haematology Disorders
P03-06 Molecular Pharmacology and Experimental Therapeutics (MPET)



Paediatric Cancer Epigenetics

Alexandra Avgustinova, PhD

6
PROJECTS
5 competitive projects
1 non competitive projects

0
CLINICAL TRIALS &
OBSERVATIONAL STUDIES

1
PUBLICATIONS

21
IMPACT FACTOR
21.3 average impact factor

100%
PUBLICATIONS IN 1ST QUARTILE

100%
PUBLICATIONS IN 1ST DECILE

0%
LEADERSHIP

MEMBERS

POSTDOC ASSOCIATED RESEARCHERS

Carlos Javier Rodriguez Hernandez

PHD STUDENTS

Valentin-Cosmin Pletea, Àlex Cebrià Xart

SELECTED PUBLICATIONS

- Llorente A, Blasco MT, Espuny I, Guiu M, Ballaré C, Blanco E, Caballé A, Bellmunt A, Salvador F, Morales A, Nuñez M, Loren G, Imbastari F, Fidalgo M, Figueras-Puig C, Gibler P, Graupera M, Monteiro F, Riera A, Holen I, Avgustinova A, Di Croce L, Gomis RR. MAF amplification licenses ERα through epigenetic remodelling to drive breast cancer metastasis. Nat Cell Biol. 2023 Dec;25(12):1833-1847. FI: 21.3 (D1)

SELECTED PROJECTS

- ONCO-COMP_Oncogenic competence during development – When, Where and Why?. (101076506). European Commission; Fundació Privada per a la Recerca

i la Docència Sant Joan de Déu - FSJD; Rodriguez. 2023-2028.

IP:Alexandra Avgustinova.

- Ayudas para contratos predoctorales para la formación de doctores 2021. (PRE2021-098532). Ministerio de Economía Y Competitividad (MINECO). 2022-2026. IP:Alexandra Avgustinova.
- GLEBioassay_Nano-Monitoring of Cancer Immunotherapy Efficiency: The Lateral Electrophoretic Bioassay platform. (AC21_2/00045). Instituto de Salud Carlos III (ISCIII). 2022-2024. IP:Carlos Javier Rodriguez Hernandez.
- El epigenoma malvado de los tumores rabdoides - definir para tratar. (PID2020-118241RA-I00). Ministerio de Economía Y Competitividad (MINECO). 2021-2023. IP:Alexandra Avgustinova.

Translational Genomics

Cinzia Emilia Lavarino, PhD

9
PROJECTS
3 competitive projects
6 non competitive projects

5
CLINICAL TRIALS &
OBSERVATIONAL STUDIES
4 commercial clinical trials
1 academic clinical trials

9
PUBLICATIONS

55
IMPACT FACTOR
6.1 average impact factor

56%
PUBLICATIONS IN 1ST QUARTILE

22%
PUBLICATIONS IN 1ST DECILE

11%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

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ASSISTANT RESEARCHER

Alba Parra Checa

TECHNICIAN

Cristina Darbra Magrú, Sonia Paco Mercader

SELECTED PUBLICATIONS

- Gómez-González S, Llano J, Garcia M, Garrido-Garcia A, Suñol M, Lemos I, Perez-Jaume S, Salvador N, Gene-Olaciregui N, Galán RA, Santa-María V, Perez-Somarrriba M, Castañeda A, Hinojosa J, Winter U, Moreira FB, Lubieniecki F, Vazquez V, Mora J, Cruz O, La Madrid AM, Perera A, Lavarino C. EpiGe: A machine-learning strategy for rapid classification of medulloblastoma using PCR-based methylgenotyping. iScience. 2023 Aug 12;26(9):107598. FI: 5.8(Q1)
- Fanlo L, Gómez-González S, Rozalén

- C, Pérez-Núñez I, Sangrador I, Tomás-Daza L, Gautier EL, Usieto S, Rebollo E, Vila-Ubach M, Carcaboso AM, Javierre BM, Celià-Terrassa T, Lavarino C, Martí E, Le Dréau G. Neural crest-related NXP1/α-NRXN signaling opposes neuroblastoma malignancy by inhibiting organotropic metastasis. *Oncogene*. 2023 Jul;42(28):2218-2233. FI: 8 (D1)
- Carton C, Evans DG, Blanco I, Friedrich RE, Ferner RE, Farschtschi S, Salvador H, Azizi AA, Mautner V, Röhl C, Peltonen S, Stivaros S, Legius E, Oostenbrink R; ERN GENTURIS NF1 Tumour Management Guideline Group. ERN GENTURIS tumour surveillance guidelines for individuals with neurofibromatosis type 1. *EClinicalMedicine*. 2023 Jan 13;56:101818. FI 15.1 (D1)
 - Esperanza-Cebollada E, Gómez-González S, Perez-Jaume S, Vega-García N, Vicente-Garcés C, Richarte-Franqués M, Rives S, Català A, Torrebadell M, Camós M. A miRNA signature related to stemness identifies high-risk patients in paediatric acute myeloid leukaemia. *Br J Haematol*. 2023 Jul;202(1):96-110. FI: 6.5 (Q1)

SELECTED PROJECTS

- Objeto [Proyectos Competitivos Grupo] SEHOP-PENCIL study - Personalised mEdicine for Cancer in children in Spain. (PMP21/00075). Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD; Instituto de Salud Carlos III (ISCIII); Lavarino. 2022-2025. IP:Cinzia Emilia Lavarino.
- Desarrollo de clasificadores epigenéticos para el Cáncer Pediátrico (ECPC). (PI20/00519). Instituto de Salud Carlos III (ISCIII). 2021-2025. IP:Cinzia Emilia Lavarino.
- Platform of Epigenetic Classifiers for Childhood CAncer (PECA). (201921-30). Fundació La Marató de TV3. 2020-2023. IP:Cinzia Emilia Lavarino.

SELECTED CLINICAL STUDIES

- Observational multicentre RWE study to characterize neurofibromatosis type 1 (NFI) diagnosed patients with

plexiform neurofibromas (PN) in Spain. "MULtyMAtin Study". (IQVIA-2889413). Alexion Pharma Spain, S.L. 2023. IP: Héctor Salvador Hernández.

- Estudio posautorización de seguridad de pacientes pediátricos que inician el tratamiento con selumetinib: Estudio de cohortes prospectivo de varios países. Post-Authorisation Safety Study of Paediatric Patients Initiating Selumetinib: A Multiple-Country Prospective Cohort Study. (D1346R00004). Astrazeneca Farmaceutica Spain, S.A.; Labcorp Drug Development Inc. 2022. IP: Héctor Salvador Hernández.
- Ensayo de Fase I/II, Abierto y de un solo brazo para Evaluar la Farmacocinética, Seguridad/ Tolerabilidad y Eficacia de la Formulación Granulada de Selumetinib en niños de ≥ 1 a < 7 Años con Neurofibromas Plexiformes (NP) Sintomáticos e Inoperables relacionados con Neurofibromatosis Tipo 1 (NF1) (SPRINKLE). (D1346C00004). Astrazeneca Farmaceutica Spain, S.A.; Diagnóstico para la imagen (DXI); Farmacia; Hospital de día PCCB; Hospitalización HSJD; Laboratori HSJD; Medina; Servicio de Cardiología; Servicio Oftalmología; Unitat de Recerca (URC) FSJD. 2021. IP: Héctor Salvador Hernández.
- Ensayo fase I, de un solo brazo, secuencial, para evaluar el efecto de los alimentos sobre la seguridad y farmacocinética de selumetinib después de dosis múltiples en niños adolescentes con neurofibromas plexiformes (NP) asociados a neurofibromatosis tipo 1 (NF1). A Phase I, Single-Arm, Sequential Study to Evaluate the Effect of Food on the Gastrointestinal Toxicity and Pharmacokinetics of Selumetinib after Multiple Doses in Adolescent Children with Neurofibromatosis Type 1 (NF1) Related Plexiform Neurofibromas (PN). (D1346C00015). Astrazeneca Farmaceutica Spain, S.A.; Diagnóstico para la imagen (DXI); Farmacia; Hospitalización HSJD; Laboratori HSJD; Servicio de Cardiología; Servicio Oftalmología; Unitat de Recerca (URC) FSJD; Vázquez. 2021. IP: Héctor Salvador Hernández.

Sarcomas and Histiocytosis

Jaume Mora Graupera, MD, PhD

25 PROJECTS
12 competitive projects
13 non competitive projects

22 CLINICAL TRIALS & OBSERVATIONAL STUDIES
13 commercial clinical trials
9 academic clinical trials

15 PUBLICATIONS

86 IMPACT FACTOR
5.7 average impact factor

47% PUBLICATIONS IN 1ST QUARTILE

13% PUBLICATIONS IN 1ST DECILE

33% LEADERSHIP

MEMBERS

STAFF RESEARCHER

Maite Gorostegui Obanos, Alicia Castañeda Heredia, Lucas Krauel Giménez-Salinas, Moira Garraus Oneca, Veronica Celis Passini

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Xenia Fiego Masriera, Carla Rios Astorch, Jara Martin Serrano, Irene Cuervas Oliveras, Alejandro Manzanares Quintela, Estela Prada Varela, Sara Pérez Jaume

TECHNICIAN

Óscar Muñoz Aznar, Noelia Salvador Marcos

COLLABORATOR RESEARCHER

Maria Inmaculada Hernandez Muñoz

2023 has been the first running up year of the Pediatric Cancer Center

Barcelona (PCCB), a unique center in Europe where science and clinical assistance meets for pediatric cancer patients. 2023 has been the 20th anniversary of the developmental tumor lab foundation, initiated in May 2003. As such, we celebrated the anniversary with the opening of our next generation laboratories including La Caixa funded new Experimental Research Solid Tumors Laboratory and the new Molecular diagnostic Laboratory. The new laboratories host over 50 people in 5 groups. Another highlight of the year has been the formalization of the institutional relationship with the IRB Barcelona with Alexandra Avgustinova, one of the Pls of the PCCB becoming dual affiliated between IRB and PCCB. The formal agreement between IRB and PCCB will facilitate further interactions between investigators, access to facilities both sides and exchange of personnel and students. 2023 also has been the year where we incorporated into the PCCB Research structure an expert in grant and scientific planning in order to strengthen the international expansion of PCCB groups activities.

SELECTED PUBLICATIONS

- Muñoz JP, Larrosa C, Chamorro S, Perez-Jaume S, Simao M, Sanchez-Sierra N, Varo A, Gorostegui M, Castañeda A, Garraus M, Lopez-Miralles S, Mora J. Early Salvage Chemo-Immunotherapy with Irinotecan, Temozolomide and Naxitamab Plus GM-CSF (HITS) for Patients with Primary Refractory High-Risk Neuroblastoma Provide the Best Chance for Long-Term Outcomes. *Cancers (Basel)*. 2023 Oct 3;15(19):4837. FI: 5.2 (Q2)
- Larrosa C, Mora J, Cheung NK. Global Impact of Monoclonal Antibodies (mAbs) in Children: A Focus on Anti-GD2. *Cancers (Basel)*. 2023 Jul 22;15(14):3729. FI:5.2 (Q2)
- Reinal I, Ontoria-Oviedo I, Selva M, Casini M, Peiró-Molina E, Fambuena-Santos C, Climent AM, Balaguer J, Cañete A, Mora J, Raya Á, Sepúlveda P. Modeling Cardiotoxicity in Pediatric Oncology Patients Using Patient-Specific iPSC-Derived Cardiomyocytes Reveals Downregulation of Cardioprotective microRNAs. *Antioxidants (Basel)*. 2023 Jul 3;12(7):1378. FI:7 (D1)

- Varo A, Castañeda A, Chamorro S, Muñoz JP, Gorostegui M, Celma MS, Lopez S, Simao M, Perez-Jaume S, Mora J. Novel infusion strategy reduces severe adverse events caused by the anti-GD2 monoclonal antibody naxitamab. *Front Oncol*. 2023 May 5;13:1164949. FI:4.7 (Q2)

SELECTED PROJECTS

- Unveiling the Ewing sarcoma cell of origin: a multistep transformation process that involves prenatal genetic and postnatal epigenetic mechanisms. (201928-30). *Fundació La Marató de TV3*. 2020-2023. IP:Jaume Mora Graupera.
- Influencia del contexto epigenético en la edad como factor pronóstico en Ewing Sarcoma. (PI20/00179). *Instituto de Salud Carlos III (ISCIII)*. 2021-2023. IP:Jaume Mora Graupera.
- Realidad virtual, aumentada e impresión 3D para la planificación quirúrgica de cirugías complejas en oncología pediátrica y creación de modelos de simulación quirúrgica para formación. *Fundació Institut de Bioingeniería de Catalunya (IBEC)*. 2023-2024. IP:Lucas Krauel Giménez-Salinas.
- Drosophila models for the study of pediatric cancer. (PFNR0153). *Científicos Diversos*. 2021 – 3000. Lucas Krauel Giménez-Salinas.

SELECTED CLINICAL STUDIES

- Ensayo de Fase pivotal 2 del anticuerpo Naxitamab (hu3F8) y factor estimulante de colonias de granulocitos y macrófagos (GM-CSF) en pacientes con Neuroblastoma de alto riesgo con enfermedad refractaria primaria o con respuesta incompleta al tratamiento de rescate en el hueso y / o médula ósea. (Y-MABS 201). *Anatomía patológica; Diagnóstico para la imagen (DXI); Farmacia; Hospital de día PCCB; Laboratori HSJD; Sanchez; Sermes Planificació, S.L; Servicio de Cardiología; SIMM Molecular S.L.; Unitat de Recerca (URC) FSJD; Y-mAbs Therapeutics A/S*. 2018. IP:Jaume Mora Graupera.
- Paediatric Hepatic International Tumour Trial. (RG_15-114 (PHITT)). *Anatomía patológica; Diagnóstico para la imagen (DXI); Farmacia; Laboratori HSJD;*

Servicio de Cardiología; Servicio Otorrinolaringología; Unitat de Recerca (URC) FSJD; University Of Birmingham. 2018. IP:Maira Garraus Oneca.

- Estudio de fase 1, multicéntrico, abierto, con intensificación de dosis y expansión de cohorte con Niraparib y Dostarlimab en pacientes pediátricos con tumores sólidos recurrentes o refractarios. A PHASE 1, MULTICENTRE, OPEN-LABEL, DOSE-ESCALATION AND COHORT EXPANSION STUDY OF NIRAPARIB AND DOSTARLIMAB IN PAEDIATRIC PATIENTS WITH RECURRENT OR REFRACTORY SOLID TUMOURS. (213406). *Anatomía patológica; Criado; Diagnóstico para la imagen (DXI); Farmacia; GlaxoSmithKline Research and Development Ltd.; Hospital de día PCCB; Laboratori HSJD; Rúmenova; Unitat de Recerca (URC) FSJD*. 2021. IP:Jaume Mora Graupera.
- A Phase II Single Center Study Of Naxitamab, Granulocyte-Macrophage Colony Stimulating Factor (GM-CSF) and Ifosfamide/ Carboplatin/Etoposide (NICE) For Patients With Relapsed/ Refractory, Soft tissue or Anti-GD2 Immunotherapy Refractory Neuroblastoma. NICE. (HSJD-HR-NB.2-2019). *Anatomía patológica; Científicos Diversos; Diagnóstico para la imagen (DXI); Farmacia; Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD; Hospital de día PCCB; Hospitalización HSJD; Laboratori HSJD; Santamaría; Servicio de Cardiología; SIMM Molecular S.L.; Unitat de Recerca (URC) FSJD*. 2020. IP:Jaume Mora Graupera.

Paediatric Cancer Treatment

Angel Montero Carcaboso, PhD



19
PROJECTS
11 competitive projects
8 non competitive projects



3
CLINICAL TRIALS &
OBSERVATIONAL STUDIES
1 commercial clinical trials
2 academic clinical trials



21
PUBLICATIONS



172
IMPACT FACTOR
8.2 average impact factor



57%
PUBLICATIONS IN 1ST QUARTILE



29%
PUBLICATIONS IN 1ST DECILE



24%
LEADERSHIP

MEMBERS

POSTDOC ASSOCIATED RESEARCHERS

Maria del Rosario Aschero

PHD STUDENTS

Nuria Martinez Velasco, Clàudia Resa Parés, Maria Cuadrado Vilanova, Helena Castillo Ecija

ASSISTANT RESEARCHER

Víctor Burgueño Sandoval, Leire Balaguer Lluna, Mercè Baulenas Farrés

TECHNICIAN

Ana Jiménez Cabaco, Mònica Vilà Ubach

COLLABORATOR RESEARCHERS

Guillermo Chantada, Ofelia Cruz Martínez, Andrés Morales La Madrid

SELECTED PUBLICATIONS

- Pascual-Pasto G, Resa-Pares C, Castillo-Ecija H, Aschero R, Baulenas-Farres M, Vila-Ubach M, Burgueño V, Balaguer-Lluna L, Cuadrado-Vilanova M, Olaciregui NG, Martinez-Velasco N, Perez-Jaume S, de Alava E, Tirado OM,

Lavarino C, Mora J, Carcaboso AM. Low Bcl-2 is a robust biomarker of sensitivity to nab-paclitaxel in Ewing sarcoma. *Biochem Pharmacol.* 2023 Feb;208:115408. FI 5.8 (Q1)

- Cuadrado-Vilanova M, Burgueño V, Balaguer-Lluna L, Aschero R, Castillo-Ecija H, Liu J, Perez-Jaume S, Pascual-Pasto G, Olaciregui NG, Gomez-Gonzalez S, Correa G, Suñol M, Schaiquevich P, Radvanyi F, Lavarino C, Mora J, Catala-Mora J, Chantada GL, Carcaboso AM. Follow-up of intraocular retinoblastoma through the quantitative analysis of conserved nuclear DNA sequences in aqueous humor from patients. *J Pathol Clin Res.* 2023 Jan;9(1):32-43. FI: 4.1 (Q2)
- Garin JPL, Sampor C, Grigorovski N, Ceciliano A, Fandiño A, Ossandon D, Perez V, Caixeta R, Neto JCA, Alarcón S, Silva S, Teixeira L, Antoneli CG, Mattosinho C, Leal-Leal C, Castela G, Requejo F, Catala J, Macedo C, Chantada GL. How we approach conservative treatment of retinoblastoma in South America in the era of local ocular treatments: A consensus of the Grupo America Latina de Oncología Pediátrica (GALOP). *Pediatr Blood Cancer.* 2023 Jul;70(7):e30352. FI: 3.2 (Q2)
- Aschero R, Castillo-Ecija H, Baulenas-Farres M, Resa-Pares C, Jimenez-Cabaco A, Rodriguez E, Monterrubio C, Perez-Jaume S, Suñol M, Chantada GL, Lavarino C, Mora J, Carcaboso AM. Prognostic value of xenograft engraftment in patients with metastatic high-risk neuroblastoma. *Pediatr Blood Cancer.* 2023 Jun;70(6):e30318. FI: 3.2 (Q2)

SELECTED PROJECTS

- Potenciació de la infecció i oncolísi de l'adenovirus VCN-01 amb topotecan. (SLT033/23/000112). Generalitat de Catalunya. 2023-2024. IP:Angel Montero Carcaboso.
- Cáncer infantil. Gliomatosis Cerebral. Ibercaja Gestión S.G.I.I.C. S.A. 2023-2024. IP:Angel Montero Carcaboso.
- A new Lu-DOTA-Zol drug for the treatment of osteosarcoma. ITM MEDICAL ISOTOPES GMBH. 2023-2024. IP:Angel Montero Carcaboso.

SELECTED CLINICAL STUDIES

- A Phase 3, Randomized,

- International Multicenter Trial of DAY101 Monotherapy Versus Standard of Care Chemotherapy in Patients with Pediatric Low-Grade Glioma Harboring an Activating RAF Alteration Requiring First-Line Systemic Therapy. (DAY101-002). Anatomía patológica; Day One Biopharmaceuticals; Diagnóstico para la imagen (DXI); Farmacia; Gómez; Laboratori HSJD; Pharmaceutical Research Associates España SAU; Servicio de Cardiología; Servicio Endocrinología; Servicio Neurología; Servicio Oftalmología; Unitat de Recerca (URC) FSJD; Vazquez. 2023. IP: Ofelia Cruz Martínez.
- SIOP Ependimoma II - Programa clínico internacional para el diagnóstico y tratamiento de niños, adolescentes y adultos jóvenes con ependimoma. (ET-13-002). Cetir Centre Diagnòstic Pedralbes S.A; Farmacia; Laboratori HSJD; Sociedad Española de Hematología y Oncología Pediátricas; Unitat de Recerca (URC) FSJD. 2017. IP: Ofelia Cruz Martínez.
- Phase Ib Clinical Trial on the Safety of Immunotherapy With Autologous Dendritic Cells Primed With Lysate Allogeneic Tumor Lines in Patients With Diffuse Intrinsic Pontine Glioma (DIPG). (FSJD-DIPG-DC). Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD. 2016 . IP: Andrés Morales La Madrid.
- Estudio prospectivo internacional en niños mayores de 3 a 5 años con meduloblastoma de riesgo estándar clínico de perfil biológico de bajo riesgo. (SIOP - PNET 5 MB - LR). Anatomía patológica; Diagnóstico para la imagen (DXI); Farmacia; Laboratori HSJD; Mouronte; Servicio de Cardiología; SERVICIO DE CIRUGIA; Servicio Otorrinolaringología; Sociedad Española de Hematología y Oncología Pediátricas; Unitat de Recerca (URC) FSJD. 2016 . IP:Ofelia Cruz Martínez, Vicente Santa-María López.

P03-05 Leukaemia and Paediatric Haematology Disorders

Mireia Camós Guijosa, MD, PhD



12

PROJECTS

7 competitive projects
5 non competitive projects



51

CLINICAL TRIALS &
OBSERVATIONAL STUDIES

34 commercial clinical trials
17 academic clinical trials



21

PUBLICATIONS



185

IMPACT FACTOR

8.8 average impact factor



52%

PUBLICATIONS IN 1ST QUARTILE



24%

PUBLICATIONS IN 1ST DECILE



29%

LEADERSHIP

MEMBERS

STAFF RESEARCHER

Izaskun Elorza Alvarez, Isabel Badell Serra, Julia Marsal Ricoma, Ruben Berrueco Moreno, Albert Català Temprano, Susana Rives Solà, Anna Ruiz Llobet, Montserrat Torredadell Burriel

POSTDOC ASSOCIATED RESEARCHERS

Nerea Vega García

PHD STUDENTS

Clara Vicente Garces, Susanna Gassiot Riu, Elena Esperanza Cebollada, Maria Trabazo del Castillo, Anna Alonso Saladrígues, Montserrat Meseguer Meda

ASSISTANT RESEARCHER

Maximiliano Ezequiel Distefano Muñoz

TECHNICIAN

Mercé Richarte Franqués, Alba Crespo Carrasco, Andrea Ruiz Sanjuan

Our research group achieved several major milestones in 2023. Among them is the designation by the European Medicines Agency of the academic CAR-T ARI-0001 trial for the Pediatric Investigation Plan (PIP). This is an essential step in gathering the necessary data before obtaining authorization for medicine for children. Also, the Hematology Laboratory achieved the ISO15189 accreditation for the immunoglobulin/T-cell receptor (IG/TR) screening for measurable residual disease monitoring and was accepted as a member of the EuroMRD group. These are compulsory requirements to join international clinical trials like the ALLTogether trial for first-line treatment in pediatric patients diagnosed with acute lymphoblastic leukemia (ALL).

In 2023, our team was awarded a number of competitive and private projects, including an international collaborative consortium to perform comprehensive genetic, phenotypic, and functional studies on GATA2 deficiency and develop clinical guidelines to follow those patients. During 2023, several projects were closed, like the study on the thrombin generation test to personalize the prevention of deep venous thrombosis in ALL patients. We kept working on the ongoing unicentric and collaborative projects on coagulation disorders, lymphoma, CAR-T cell and transplantation-related projects, bone marrow failure and myelodysplastic syndromes, immune cytopenias, and high-risk leukemia.

Several PhD research projects were developed in our group during this period, and Elena Esperanza had her doctoral thesis defense in December 2023.

Our team carried out numerous clinical trials in 2023. Among them, we highlight the start of a CART CD1a clinical trial in T-ALL patients and multiple clinical trials on lymphoma, leukemia, transplantation, and coagulation disorders.

Finally, our group has published more than 25 scientific articles in 2023, including papers on CART therapy, GATA2 deficiency, and post-transplant lymphoproliferative disorders accepted in top high-impact journals like the Journal of Clinical Oncology, Haematologica, or Blood.

SELECTED PUBLICATIONS

- Berrueco R, González-Forster E, Deya-Martinez A, Solsona M, García-García A, Calzada-Hernández J, Yiyi L, Vlagea A, Ruiz-Llobet A, Alsina L. Mycophenolate mofetil for autoimmune cytopenias in children: high rates of response in inborn errors of immunity. *Front Pediatr.* 2023 Oct 17;11:1174671. FI:2.6 (Q2)
- Esperanza-Cebollada E, Gómez-González S, Perez-Jaume S, Vega-García N, Vicente-Garcés C, Richarte-Franqués M, Rives S, Català A, Torredadell M, Camós M. A miRNA signature related to stemness identifies high-risk patients in paediatric acute myeloid leukaemia. *Br J Haematol.* 2023 Jul;202(1):96-110. FI:6.5 (Q1)
- Marin-Bejar O, Romero-Moya D, Rodriguez-Ubreva J, Distefano M, Lessi F, Aretini P, Liquori A, Castaño J, Kozyra E, Kotmayer L, Bueno C, Cervera J, Rodriguez-Gallego JC, Nomdedeu JF, Murillo-Sanjuán L, De Heredia CD, Pérez-Martinez A, López-Cadenas F, Martínez-Laperche C, Dorado-Herrero N, Marco FM, Prósper F, Menéndez P, Valcárcel D, Ballestar E, Bódör C, Bigas A, Catalá A, Wlodarski MW, Giorgetti A. Epigenome profiling reveals aberrant DNA methylation signature in GATA2 deficiency. *Haematologica.* 2023 Sep 1;108(9):2551-2557. FI:10.1 (Q1)
- Laetsch TW, Maude SL, Rives S, Hiramatsu H, Bittencourt H, Bader P, Baruchel A, Boyer M, De Moerloose B, Qayed M, Buechner J, Pulsipher MA, Myers GD, Stefanski HE, Martin PL, Nemecek E, Peters C, Yanik G, Khaw SL, Davis KL, Krueger J, Balduzzi A, Boissel N, Tiwari R, O'Donovan D, Grupp SA. Three-Year Update of Tisagenlecleucel in Pediatric and Young Adult Patients With Relapsed/Refractory Acute Lymphoblastic Leukemia in the ELIANA Trial. *J Clin Oncol.* 2023 Mar 20;41(9):1664-1669. FI:45.4 (D1)

SELECTED PROJECTS

- CLOSER: Childhood Leukemia: Overcoming distance between South America and Europe Regions. [825749]. European Commission. 2019-2024. IP:Mireia Camós Guijosa.
- New approaches for the identification and functional

characterization of prognostic genetic biomarkers in inherited childhood myelodysplastic/acute leukemia syndromes Acronym: GEMMA. [202001-30]. Fundació La Marató de TV3. 2021-2024. IP:Albert Català Temprano.

- Estudio multicéntrico fase 2 en pacientes pediátricos (0-18 años) con leucemia linfoblástica aguda B CD19+ en recaída o refractaria. (IC121/00033). Instituto de Salud Carlos III (ISCIII). 2022-2025. IP:Susana Rives Solà
- Diagnóstico integrado de la enfermedad de células falciformes (ECF) y otras anemias raras (AR) para la medicina personalizada (ESTUDIO INTEGRA). (PCP00369). Fundació Institut de Recerca Hospital Universitari Vall D'Hebron. 2023 - 2024. IP: Ruben Berruoco Moreno.

SELECTED CLINICAL STUDIES

- Seguridad y eficacia de la terapia hCD1a-CAR T (OC-1), en pacientes con leucemia/linfoma linfoblástico agudo de células T (T-ALL/LL) en recaída/refractario (R/R) (CARxALL). Safety and efficacy of hCD1a-CAR T (OC-1) therapy, in patients with relapsed/refractory (R/R) T-cell acute lymphoblastic leukemia/lymphoma (T-ALL/LL). (OC-01-21001). Anatomía patológica; Banc de Sang i Teixits; BioClever 2005, SLU; Diagnóstico para la imagen (DXI); Farmacia; Hospital de dia PCCB; Hospitalización HSJD; Laboratori HSJD; Onechain Immunotherapeutics S.L.; Servicio de Cardiología; SERVICIO DE CIRUGIA; SIMM Molecular S.L.; Unitat de Recerca (URC) FSJD. 2023. IP:Susana Rives Solà.
- Protocolo de estudio de tratamiento del consorcio ALLTogether para niños y adultos jóvenes (0-18 años) con diagnóstico reciente de leucemia linfoblástica (LLA). (ALLTogether1 - PILOT). Karolinska Institutet. 2023 - . IP: José Luis Dapena Diaz.
- Estudio piloto de la infusión de linfocitos T autólogos modificados genéticamente para expresar anti-CD19 en pacientes con leucemia o linfoma CD19+ resistente o refractario a tratamiento. (CART19-BE-01). Banc de Sang i Teixits; Diagnóstico para la imagen (DXI); Hospital Clínic Barcelona; Hospital de dia PCCB; Hospitalización HSJD; Laboratori HSJD; Servicio de Cardiología; SERVICIO DE CIRUGIA; Unitat de Recerca (URC) FSJD. 2017. IP:Susana Rives Solà.
- Estudio prospectivo español de pacientes hematológicos pre- y posttrasplante hematopoyético que ingresan en unidades de cuidados intensivos: análisis de la práctica clínica, resultados en salud, factores pronósticos y utilización de recursos sanitarios. (GET-PEN-2016-01). Grupo Español de Trasplante Hematopoyético y Terapia Celular. 2019. IP:Julia Marsal Ricoma.

P03-06 Molecular Pharmacology and Experimental Therapeutics (MPET)

Marçal Pastor-Anglada, PhD

0
CLINICAL TRIALS &
OBSERVATIONAL STUDIES

2
PUBLICATIONS

8
IMPACT FACTOR
4.1 average impact factor

50%
PUBLICATIONS IN 1ST QUARTILE

0%
PUBLICATIONS IN 1ST DECILE

0%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Sandra Perez-Torras

POSTDOC ASSOCIATED RESEARCHERS

Liska Caviedes Cárdenas, Jose Antonio Agüero Fernández, Aida Mata Ventosa, Nerea Urtasun Plans

PHD STUDENTS

Andrea Valentina Prieto Carruyo

SELECTED PUBLICATIONS

- Pena-Rodríguez E, García-Vega L, Lajarin Reinares M, Pastor-Anglada M, Pérez-Torras S, Fernandez-Campos F. Latanoprost-Loaded Nanotransfersomes Designed for Scalp Administration Enhance Keratinocytes Proliferation. Mol Pharm. 2023 May 1;20(5):2317-2325. IF: 4.9 (Q1).
- Galán V, Beléndez C, Echecopar C, Estival P, Sissini L, Olivas R, Bueno D, Molina B, Fuentes C, Regueiro A, Benítez I, Plaza M, Margarit A, Rifón J, Pascual A, Palomo P, Urtasun A, Fuster JL, Díaz de Heredia C, Fernández Navarro JM, González-

Vicent M, Ruz B, Pérez-Martínez A. Treosulfan-Based Conditioning Regimen In Pediatric Hematopoietic Stem Cell Transplantation: A Retrospective Analysis on Behalf of the Spanish Group for Hematopoietic Transplantation and Cellular Therapy (GETH-TC). *Transplant Cell Ther.* 2023 Nov;29(11):702.e1-702.e11. IF: 3.2 (Q2).

SELECTED PROJECTS

- Doctorat industrial en col.laboració amb empresa: Laboratori Reig Jofre. 2023DI00007. Agència de Gestió d'Ajuts Universitaris i de Recerca (AGAUR). Ajuts a universitats i a empreses per al desenvolupament d'un projecte de doctorat industrial, en qualsevol àmbit de coneixement i qualsevol sector empresarial. Doctorats industrials 2023. 2023-2026. IP:Sandra Perez Torras
- Proteínas transportadoras de nucleósidos (NTs) en la intersección de las vías biosintéticas de nucleótidos y la reprogramación metabólica. PID2021-124765OB-I00. Ministerio de Ciencia e Innovación (MICINN). Convocatoria 2021 de ayudas a «Proyectos de Generación de Conocimiento». Modalidad Proyectos de «Investigación Orientada». 2022-2025. IP:Marçal Pastor-Anglada.
- Descubrimiento y desarrollo temprano de nuevos activadores de CNTs para potenciar la inmunoterapia del cáncer. PLEC2022-009319. Ministerio de Ciencia e Innovación (MICINN). Convocatoria 2022 de ayudas a proyectos de investigación en líneas estratégicas. 2022-2025. IP:Marçal Pastor-Anglada.
- Farmacologia Molecular i Teràpies Experimentals (MPET). 2021 SGR 00677. Agència de Gestió d'Ajuts Universitaris i de Recerca (AGAUR). Ajuts per donar suport a l'activitat científica dels grups de recerca de Catalunya (SGR-Cat 2021). 2022-2025. IP:Marçal Pastor-Anglada.



7.1

MEDICINE AND PEDIATRIC SCIENCE AREA

7.1.4 Precision Medicine in Rare Diseases

Metabolomics and Biochemical Genetics
Neurogenetics and Molecular Medicine
Genomics for the Diagnosis of Rare Diseases
Human Molecular Genetics I
Human Molecular Genetics II
Diagnostic and Therapeutic Imaging



Metabolomics and Biochemical Genetics

Rafael Artuch Iriberry, MD, PhD



18
PROJECTS
9 competitive projects
9 non competitive projects



10
CLINICAL TRIALS &
OBSERVATIONAL STUDIES
10 commercial clinical trials
0 academic clinical trials



24
PUBLICATIONS



154
IMPACT FACTOR
6.4 average impact factor



75%
PUBLICATIONS IN 1ST QUARTILE



33%
PUBLICATIONS IN 1ST DECILE



38%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Roser Urreizti Frexedas, Jaume Campistol Plana, Mercedes Serrano Gimará, Camila García Volpe, Rosa Gassió Subirachs, Aida Ormazabal Herrero, Cristina Sierra March

POSTDOC ASSOCIATED RESEARCHERS

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PHD STUDENTS

Mónica Centeno Pla

ASSISTANT RESEARCHER

Helena Rodríguez González, Eric Kramer Rosado, Florencia Epifani, Gregorio Alexander Nolasco Tovar

TECHNICIAN

Lidia Celina Sánchez Silvestre, Adriana Balen Valeiro, Yahel Calzadilla Borrás, Aroa Fernández de Miguel, Juan Moreno García, Tania Pardal Chofle

Our group has increased its scientific activity in the last years, as its principal investigators continue with

active projects financed by public funds. Of note is the achievement of the IMPACT Únicas project, led by the group's PI, which aims to access the data of 5 pediatric hospitals nationwide that are experts in rare diseases, in a federated manner, in collaboration with the Barcelona supercomputing center.

The level of publication in this period is stable compared to previous years, with a strong translational component and collaboration with national and international groups.

At this momento, we have established the scientific committee of GenRare, a CIBERER platform that brings together different registries of rare diseases (Dr. Artuch is the president of this committee). Our hospital has been designated with several CSURs, and as a result, we are participating in MetabERN, the European reference network for metabolic diseases.

Regarding metabolomic studies, we continue to collaborate with different CIBERER and non-CIBERER groups in the phenotyping of cellular and animal models. This has generated growing scientific activity, and the idea we have for the future is to consolidate our metabolomics platform to provide biochemical phenotyping services to CIBER and external research groups, according to the scientific interest of the proposal.

Our most relevant contribution to the future is to remain a reference group for the study of rare diseases in the pediatric field. The axes of our future activity are aligned with the lines of research that we currently have, since our intention is to improve our research capabilities in these lines rather than addressing new groups of diseases.

SELECTED PUBLICATIONS

- Epifani F, Pujol Serra SM, Llorens M, Balcells S, Nolasco G, Bolasell M, Aguilera-Albesa S, Cancho Candela R, Cuevas Cervera JL, García Sánchez V, Garcia O, Miranda-Herrero MC, Moreno-Lozano PJ, Robles B, Roldán Aparicio S, Velázquez Fragua R, Serrano M. Untangling adaptive functioning of PMM2-CDG across age and its impact on parental stress: a cross-sectional study. *Sci Rep.* 2023 Dec

20;13(1):22783. FI:4.6 (Q2)

- Mariño Z, Molera-Busoms C, Badenas C, Quintero-Bernabeu J, Torra M, Forns X, Artuch R. Benefits of using exchangeable copper and the ratio of exchangeable copper in a real-world cohort of patients with Wilson disease. *J Inherit Metab Dis.* 2023 Sep;46(5):982-991. FI:4.2 (Q1)
- Paredes-Fuentes AJ, Oliva C, Urreizti R, Yubero D, Artuch R. Laboratory testing for mitochondrial diseases: biomarkers for diagnosis and follow-up. *Crit Rev Clin Lab Sci.* 2023 Jun;60(4):270-289. FI:10 (D1)
- Castilla-Vallmanya L, Centeno-Pla M, Serrano M, Franco-Valls H, Martínez-Cabrera R, Prat-Planas A, Rojano E, Ranea JAG, Seoane P, Oliva C, Paredes-Fuentes AJ, Marfany G, Artuch R, Grinberg D, Rabionet R, Balcells S, Urreizti R. Advancing in Schaaf-Yang syndrome pathophysiology: from bedside to subcellular analyses of truncated MAGEL2. *J Med Genet.* 2023 Apr;60(4):406-415. FI:4 (Q2)

SELECTED PROJECTS


- Contratos predoctorales de formación en investigación en salud_Beneficiari: Florencia Epifani. (FI22/00218). Instituto de Salud Carlos III (ISCIII). 2023-2026. IP:Mercedes Serrano Gimará.
- ÚNICAS. Implementation of a Pediatric Network for Personalised Medicine in rare pediatric diseases. A Pilot Project. (PMP22/00008). Artuch; Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD; Instituto de Salud Carlos III (ISCIII). 2023-2025. IP:Rafael Artuch Iriberry.
- Desarrollo de una plataforma de análisis metabolómico dirigido para el diagnóstico e investigación de enfermedades raras de base genética. (BR202102). Hospital Sant Joan de Déu - Esplugues HSJD. 2022-2025. IP:Rafael Artuch Iriberry.
- Stroke-like episodes en CDG: propuesta multicéntrica internacional para descubrir el verdadero fenotipo, mejorar el diagnóstico y tratamiento y entender los mecanismos subyacentes. (PI21/00068). Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD; Instituto de Salud Carlos III (ISCIII). 2022-2024. IP:Mercedes Serrano Gimará.

SELECTED CLINICAL STUDIES

- Estudio de fase IV, abierto y de una sola cohorte, de los resultados neurocognitivos (NC) a largo plazo en niños de 4 a 5 años con fenilcetonuria tratados con dihidrocloruro de sapropterina (Kuvan®) durante 7 años. (EMR 700773-002). Merck KGaA; Parexel International S.L. 2014. IP:Jaume Campistol Plana.
- A Phase IIA Multicenter, Open-Label, 12-Week Study to investigate the pharmacokinetics and safety and to provide proof of mechanism of alogabat in children and adolescents aged 5–17 years with Angelman Syndrome (AS) with deletion genotype. (BP41315). F. Hoffmann-La Roche Ltd.; Farmacia; Laboratori HSJD; Moyano; Servicio de Cardiología; Servicio Neurofisiología; Servicio Neuropsicología; Unitat de Recerca (URC) FSJD. 2023. IP:Mercedes Serrano Gimaré.
- A Phase 1/2 Open-label, Multiple-dose, Dose-escalating Clinical Trial of the Safety and Tolerability of GTX-102 in Pediatric Patients With Angelman Syndrome (AS). (GTX-102-001). Diagnóstico para la imagen (DXI); Escobar; Farmacia; Hospitalización HSJD; Laboratori HSJD; Medpace Clinical Research, LLC; Servicio de Cardiología; Servicio Neurofisiología; Servicio Neuropsicología; Ultragenyx Pharmaceutical, Inc.; Unitat de Recerca (URC) FSJD. 2023. IP:Mercedes Serrano Gimaré.
- A Phase 2, Randomized, Open-Label, 24-Week Study to Assess the Pharmacodynamics, Safety, Tolerability, and Pharmacokinetics of Multiple Doses of GLM101 Administered Intravenously to Adult and Adolescent Participants with PMM2-CDG. (GLM101-002). And Glycomine, Inc; Diagnóstico para la imagen (DXI); el Harchaoui; Farmacia; García; Hernandez; Hospital de dia HSJD; Hospital de dia URC; Jorgesen; Laboratori HSJD; Precision for medicine (HU) KFT; Servicio de Cardiología; Servicio Neurofisiología; Servicio Oftalmología; SIMM Molecular S.L.; Unitat de Recerca (URC) FSJD. 2022. IP:Mercedes Serrano Gimaré.


Neurogenetics and Molecular Medicine

Janet Hoenicka, PhD
& Francesc Palau Martínez, MD, PhD


 **17**
PROJECTS
11 competitive projects
6 non competitive projects

 **0**
CLINICAL TRIALS &
OBSERVATIONAL STUDIES

 **25**
PUBLICATIONS

 **125**
IMPACT FACTOR
5 average impact factor

 **48%**
PUBLICATIONS IN 1ST QUARTILE

 **8%**
PUBLICATIONS IN 1ST DECILE

 **28%**
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Joan Maynou Fernández, Mónica Roldan Molina, Guerau Fernandez Isern

POSTDOC ASSOCIATED RESEARCHERS

Mar Borregán Prats, Esther Cuatrecasas Capdevila, Cristina Hernando Davalillo, Laura Martí Sanchez, Jordi Pijuan Marquilles, Lara Cantarero Abad, Loreto Martorell Sampol

PHD STUDENTS

Berta Estévez Arias

ASSISTANT RESEARCHER

Marcos Frías Nestares, Natalia Caelles Gramunt, Natalia Lourdes Serrano, Claire Melody Nguyen, Gemma Tort Vázquez, Yaiza Díaz Osorio, Adrián Alcalá San Martín, Jodie Kane, Alba Pascual Rodríguez, Mercè Bolasell Girgas, Antonio F Martínez Monseny

TECHNICIAN

Daniel Castillo Mestres, Jordi Genovès Escarré

In 2023, the group focused on translational actions on precision diagnosis and therapy for rare neurogenetic and undiagnosed diseases.

Firstly, we have confirmed the tethering between mitochondria and other organelles, such as lysosomes and peroxisomes, through GDAP1. This is an important mechanism and molecular target of Charcot-Marie-Tooth neuropathy's pathophysiology, as Cantarero et al. published in *Biology Open* 2023. Secondly, in collaboration with the Departments of Genetic Medicine and Pediatric Neurology, we have discovered new genotype-phenotype relationships, pathogenic mechanisms, and new disease genes related to *ANXA11*, *AIFM1*, and *DTNA* genes. These findings have been published by Pijuan et al. *Mov Disord* 2023, Natera-De Benito et al. *Ann Clin Transl Neurol* 2023, and Nascimento et al. *Acta Neuropathol* 2023. Thirdly, we have successfully resolved 27 undiagnosed cases using both in silico and experimental biology approaches. Fourthly, we have contributed to identifying several genetic loci and variants in Parkinson's disease within the Global Parkinson's Genetic Program by Kim et al. *Nat Genet*, 2023. Finally, we have administered exceptional treatment with elesclomol-copper, a repurposed drug, in children with Menkes disease and have achieved good results.

The group received funding from various public agencies, including EU Horizon, the Spanish Research Agency, the Instituto de Salud Carlos III, the Generalitat de Catalunya (SGR Precision Medicine in Genetic and Rare Diseases), and private foundations such as Fundación Ramón Areces, Menkes International Association, and Fundación Amigos de Nono and Asociación Española Síndrome Coffin-Siris.

One relevant initiative is the Translational Diagnosis and Therapy Program (PDTT), an innovative intramural action to improve the etiological diagnosis and treatment of rare and undiagnosed diseases through functional genomics. This action has led to the coordination of working group 3 of IMPaCT-Genomics and an ACCI CIBERER project, with the participation of 13 CIBERER groups.

SELECTED PUBLICATIONS

- Pijuan J, Moreno DF, Yahya G, Moisa M, Ul Haq I, Krukiewicz K, Mosbah R, Metwally K, Cavalu S. Regulatory and pathogenic mechanisms in response to iron deficiency and excess in fungi. *Microb Biotechnol.* 2023 Nov;16(11):2053-2071. FI: 5.7 (Q1)
- Natera-de Benito D, Olival J, Garcia-Cabau C, Jou C, Roldan M, Codina A, Expósito-Escudero J, Battle C, Carrera-García L, Ortez C, Salvatella X, Palau F, Nascimento A, Hoenicka J. Common pathophysiology for ANXA11 disorders caused by aspartate 40 variants. *Ann Clin Transl Neurol.* 2023 Mar;10(3):408-425. FI:5.3 (Q1)
- Lorenzo D, Esquerda M, Bofarull M, Cusi V, Roig H, Bertran J, Carrera J, Torralba F, Cambra FJ, Vila M, Garriga M, Palau F. The reuse of genetic information in research and informed consent. *Eur J Hum Genet.* 2023 Dec;31(12):1393-1397. FI:5.2 (Q1)
- Roldán M, Nolasco GA, Armengol L, Frías M, Morell M, García-Aragonés M, Epifani F, Muchart J, Ramírez-Almaraz ML, Martorell L, Hernando-Davalillo C, Urreizti R, Serrano M. Advanced Optical Microscopy: Unveiling Functional Insights Regarding a Novel *PPP2R1A* Variant and Its Unreported Phenotype. *Int J Mol Sci.* 2023 Sep 5;24(18):13699. FI:5 (Q1)

SELECTED PROJECTS

- BETTER_Better rEal-world healTh-daTa distributEd analytics Research platform. (101136262). European Commission. 2023-2027. IP:Francesc Palau Martínez.
- BE-LIGHT: Improving BiomEdical diagnosis through LIGHT-based technologies and machine learning. (101119924). European Commission. 2023-2026. IP:Mónica Roldan Molina.
- Genes y Fenotipos de Síndromes Parkinsonianos: validación funcional de hallazgos genéticos - ParkGenPhen. (PI22/00680). Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD; Hoenicka; Instituto de Salud Carlos III (ISCIII). 2023-2025. IP:Janet Hoenicka.
- Dinámica de la membrana mitocondrial y sus sitios de contacto entre orgánulos en la fisiopatología celular y del nervio de la enfermedad neurogenética. (PID2020-114655RB-100). Ministerio de Economía Y Competitividad (MINECO). 2021-2024. IP:Francesc Palau Martínez, Janet Hoenicka.

Genomics for the Diagnosis of Rare Diseases

Judith Armstrong Morón, PhD

9 PROJECTS
6 competitive projects
3 non competitive projects

0 CLINICAL TRIALS & OBSERVATIONAL STUDIES

11 PUBLICATIONS

64 IMPACT FACTOR
5.8 average impact factor

82% PUBLICATIONS IN 1ST QUARTILE

18% PUBLICATIONS IN 1ST DECILE

27% LEADERSHIP

MEMBERS

STAFF RESEARCHER

Núria Brandi Tarrrau, Délia Yubero Siles

PHD STUDENTS

Ainhoa Pascual Alonso, Clara Xiol Viñas

TECHNICIAN

Carlota Ros Bonaventura, Paola Pacheco Fernández

During 2023, we highlight two major scientific milestones. On one hand, we have characterized the expression profile in fibroblasts of RTT-MECP2 and MECP2 Duplication patients, and have proposed a molecular footprint for these syndromes. Additionally, we have identified a shared profile among patients within the Rett spectrum, opening new lines of research to detect a potential biomarker for neurodevelopmental disorders. This finding has been published in *Human Genomics* (*Hum Genomics.* 2023 Sep 15;17(1):85.), and has facilitated the defense of two ongoing doctoral thesis on the study of MECP2 gene

expression in Rett/Rett-spectrum syndrome and MECP2 Duplication syndrome. On the other hand, the research line on Rett syndrome has been consolidated and awarded of competitive public and private projects (ISCIII-FIS PI23/00108 and ADEY Foundation), allowing us to continue with the line initiated in the identification of MECP2 expression biomarkers in fibroblasts of patients within the Rett spectrum.

Regarding genomic approach and improvement of rare diseases genetic diagnosis, we have advanced on the genetic variants network (funded by Fundació La Marató de TV3 202040-30, until 31/12/2024), developing internal databases and with Beacon working on each involved Hospital. The network is being verified, and being able to connect between Hospitals brings privacy issues that must be solved. All the advances on the network development and concerns are stated on a paper expected to publish by the end of this year.

SELECTED PUBLICATIONS

- Nascimento A, Bruels CC, Donkervoort S, Foley AR, Codina A, Milisenda JC, Estrella EA, Li C, Pijuan J, Draper I, Hu Y, Stafki SA, Pais LS, Ganesh VS, O'Donnell-Luria A, Syeda SB, Carrera-García L, Expósito-Escudero J, Yubero D, Martorell L, Pinal-Fernandez I, Lidov HGW, Mammen AL, Grau-Junyent JM, Ortez C, Palau F, Ghosh PS, Darras BT, Jou C, Kunkel LM, Hoenicka J, Bönnemann CG, Kang PB, Natera-de Benito D. Variants in *DTNA* cause a mild, dominantly inherited muscular dystrophy. *Acta Neuropathol.* 2023 Apr;145(4):479-496. FI:12.7 (D1)
- Petazzi P, Jorge-Torres OC, Gomez A, Scognamiglio I, Serra-Musach J, Merkel A, Grases D, Xiol C, O'Callaghan M, Armstrong J, Esteller M, Guil S. Global Impairment of Immediate-Early Genes Expression in Rett Syndrome Models and Patients Linked to Myelination Defects. *Int J Mol Sci.* 2023 Jan 11;24(2):1453. FI:5.6 (Q1)
- Paredes-Fuentes AJ, Oliva C, Urreizti R, Yubero D, Artuch R. Laboratory testing for mitochondrial diseases: biomarkers for diagnosis and follow-up. *Crit Rev Clin Lab Sci.* 2023 Jun;60(4):270-289. FI:10 (D1)

- Pascual-Alonso A, Xiol C, Smirnov D, Kopajtich R, Prokisch H, Armstrong J. Identification of molecular signatures and pathways involved in Rett syndrome using a multi-omics approach. *Hum Genomics*. 2023 Sep 15;17(1):85. FI:4.5 (Q1)

SELECTED PROJECTS

- Estudio de pacientes dentro del Espectro Rett mediante un enfoque multiómico integrativo: delucidar la huella molecular de la vía funcional. (PI20/00389). Instituto de Salud Carlos III (ISCIII). 2021-2024. IP: Judith Silvia Armstrong Morón
- Catalan Interhospital Network of Genetic Variants to Improve Genetic Diagnosis in Rare Diseases. (590/C/2020). Fundació La Marató de TV3. 2021-2024. IP: Dèlia Yubero Siles
- Estudio de validación de dianas terapéuticas en muestra cerebral y sangre periférica de pacientes con síndrome de Rett. FUNDACIÓ ADEY; Fundació Noelia. Niños Contra la Distrofia Muscular Congénita por Déficit de Colágeno VI. 2023-2025. IP: Judith Silvia Armstrong Morón.
- Identificación de biomarcadores y dianas terapéuticas a través del análisis integrado de multiómica de la huella molecular del síndrome de Rett y trastornos del neurodesarrollo. PFIS0381. Instituto de Salud Carlos III (ISCIII). 2024-2026. IP: Judith Silvia Armstrong Morón.

Human Molecular Genetics I

Susana Balcells Comas, PhD

0
CLINICAL TRIALS &
OBSERVATIONAL STUDIES

8
PUBLICATIONS

58
IMPACT FACTOR
7.2 average impact factor

63%
PUBLICATIONS IN 1ST QUARTILE

50%
PUBLICATIONS IN 1ST DECILE

38%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Bru Cormand Rifà

POSTDOC ASSOCIATED RESEARCHERS

Raquel Rabionet Janssen, Noelia Fernández Castillo, Roser Corominas

TECHNICIAN

Mónica Cozar Morillo

In 2023 we have extended our molecular pathology studies of Schaaf-Yang (SYS) and TRAF7 syndromes. We have confirmed that the truncated MAGEL2 proteins of SYS accumulate in the nucleus, which points to a gain of function and constitutes a useful cellular biomarker. Using previously obtained patient and control iPSCs, we have differentiated neurons, astrocytes, and brain organoids, and obtained proof of concept results on the omics methodologies to be used for a full characterization of these cells and organoids. For TRAF7 syndrome, to advance into therapeutic avenues, we are assaying nutraceutical compounds on TRAF7 patients' fibroblasts and assaying their effects with previously identified RNA biomarkers.

Taking over on exome results of patients with atypical femoral fractures (AFF), we have explored the

contribution to its genetic etiology of rare variants in genes involved in skeletal dysplasias.

In our research on genetics of stroke recovery, we have initiated in vitro analyses involving a blood brain barrier cell model to assay the rare variants identified in a novel gene with protective effects.

We have studied genetic risk factors underlying various complex psychiatric and neurological disorders. On one hand, we have used several animal models to study the contribution of specific genes to multiple psychiatric conditions (*RBFOX1* in zebrafish and *SNORC* in mouse) and the therapeutic effects of certain repurposed drugs (fluoxetine) have been tested. On the other hand, genome-wide studies have been performed to identify genetic risk variants in addiction and ADHD and to investigate pleiotropic effects with other conditions.

Finally, we made progress in our project studying the Williams-Beuren and 7q11.23 microduplication syndromes. We generated iPSC-derived brain organoids and will assess transcriptomic and morphological alterations. In addition, we obtained developing brain and aorta samples of a WBS patient, and we are studying methylation alterations and scRNAseq of isolated nuclei from brain.

SELECTED PUBLICATIONS

- Antón-Galindo E, Cabana-Domínguez J, Torrico B, Corominas R, Cormand B, Fernández-Castillo N. The pleiotropic contribution of genes in dopaminergic and serotonergic pathways to addiction and related behavioral traits. *Front Psychiatry*. 2023 Oct 23;14:1293663. FI:4.7 (Q2)
- Demontis D, Walters GB, Athanasiadis G, Walters R, Therrien K, Nielsen TT, Farajzadeh L, Voloudakis G, Bendt J, Zeng B, Zhang W, Grove J, Als TD, Duan J, Satterstrom FK, Bybjerg-Grauholm J, Bækved-Hansen M, Gudmundsson OO, Magnusson SH, Baldursson G, Davidsdottir K, Haraldsdottir GS, Agerbo E, Hoffman GE, Dalsgaard S, Martin J, Ribasés M, Boomsma DI, Soler Artigas M, Roth Mota N, Howrigan D, Medland SE, Zayats

T, Rajagopal VM; ADHD Working Group of the Psychiatric Genomics Consortium; iPSYCH-Broad Consortium; Nordentoft M, Mors O, Hougaard DM, Mortensen PB, Daly MJ, Faraone SV, Stefansson H, Roussos P, Franke B, Werge T, Neale BM, Stefansson K, Børghlum AD. Genome-wide analyses of ADHD identify 27 risk loci, refine the genetic architecture and implicate several cognitive domains. *Nat Genet.* 2023 Feb;55(2):198-208. FI:30.8 (D1)

- Cabana-Domínguez J, Antón-Galindo E, Fernández-Castillo N, Singgih EL, O'Leary A, Norton WH, Strekalova T, Schenck A, Reif A, Lesch KP, Slattery D, Cormand B. The translational genetics of ADHD and related phenotypes in model organisms. *Neurosci Biobehav Rev.* 2023 Jan;144:104949. FI:8.2 (D1)
- Castilla-Vallmanya L, Centeno-Pla M, Serrano M, Franco-Valls H, Martínez-Cabrera R, Prat-Planas A, Rojano E, Ranea JAG, Seoane P, Oliva C, Paredes-Fuentes AJ, Marfany G, Artuch R, Grinberg D, Rabionet R, Balcells S, Urreiziti R. Advancing in Schaaf-Yang syndrome pathophysiology: from bedside to subcellular analyses of truncated MAGEL2. *J Med Genet.* 2023 Apr;60(4):406-415. FI:4 (Q2)

SELECTED PROJECTS

- Estudi de la vulnerabilitat al trastorn per consum de cocaïna després del consum d'alcohol. 220/U/2022. Fundació La Marató de TV3. Ajuts econòmics a projectes de recerca de la Fundació Marató de TV3. 2023-2026. IP:Bru Cormand Rifa.
- Modelos 2D y 3D de cerebro y hueso y su aplicación a la investigación de la patofisiología de las enfermedades. PID2022-141461OB-I00. Ministerio de Ciencia e Innovación (MICINN). Convocatoria 2022 de ayudas a «Proyectos de Generación de Conocimiento». Modalidad Proyectos de «Investigación Orientada». 2023-2026. IP:Susanna Balcells Comas.
- Pleiotropía y comorbilidad en el trastorno por déficit de atención con hiperactividad. PID2021-127776OB-I00. Ministerio de Ciencia e Innovación (MICINN). Convocatoria 2021 de ayudas a «Proyectos de Generación de Conocimiento».

Modalidad Proyectos de «Investigación Orientada». 2022-2025. IP:Bru Cormand Rifa.

- Ajut per incentivar i consolidar la recerca d'excel·lència ja existent a les universitats públiques de Catalunya. Programa ICREA Academia 2021. Fundació Institució Catalana de Recerca i Estudis Avançats (ICREA). Convocatòria del Programa Icrea Acadèmia. 2022-2026. IP:Bru Cormand Rifa.

Human Molecular Genetics II

Gemma Marfany Nadal, PhD

0 CLINICAL TRIALS & OBSERVATIONAL STUDIES

3 PUBLICATIONS

21 IMPACT FACTOR
7 average impact factor

67% PUBLICATIONS IN 1ST QUARTILE

33% PUBLICATIONS IN 1ST DECILE

33% LEADERSHIP

MEMBERS

POSTDOC ASSOCIATED RESEARCHERS

Serena Mirra

Our main line of research focuses on the generation and characterization of animal and cell models to study inherited retinal dystrophy genes with the final aim of finding an effective precision therapy.

To study the molecular bases of hereditary blindness, we generated mouse models by gene-editing techniques (CRISPR-Cas9) on two genes that cause retinitis pigmentosa and S-cone syndrome: CERKL and NR2E3, respectively. Lately, we performed more in-depth molecular studies to elucidate why mutations in these genes cause hereditary blindness. For NR2E3, we have performed single cell transcriptomics to characterize the photoreceptor populations of the mutant strains compared to wild-type (collaboration with Dr. Vaquerizas at Imperial College London). Regarding CERKL, we have collaborated with other IBUB and IRSJD groups (Dr. Villaroya and Dr. Artuch) as well as several CIBERER groups (Dr. Cuezva and Dr. Pallardó) to show that CERKL depletion causes alterations in mitochondrial

morphology and dynamics. We also performed transcriptomic and metabolomic analyses in retinal cells, opening new perspectives for a potential treatment by regulation of mitochondrial metabolism. All this work has generated a comprehensive article published in the best journal in the REDOX field. We are currently exploring potential gene therapy treatments by subretinal injection of adeno-associated viruses (AAVs). Finally, we also investigate genes that may contribute to the formation and stability of the sensory cilium, so that they could be candidates for causing ciliopathies, one of the symptoms being retinal neurodegeneration, specifically EVC/EVC2, RPGR and USP48.

We are also currently developing new avenues of research, such as the generation of human retinal organoids from hiPSCs. The year 2023 has very intense in terms of publishing the work resulting from many years of national collaborations with other IRSJD and CIBERER groups, centered in the functional characterisation of genes causing rare diseases, such as ciliopathies, retinal dystrophies and neurodevelopmental disorders.

SELECTED PUBLICATIONS

- Cuñat O, Del Hoyo-Buxo B, Vila-Badia R, Serra-Arumí C, Butjosa A, Del Cacho N, Colomer-Salvans A, Dolz M, Cuevas-Esteban J, Iglesias-González M, Usall J, Profep Group. Negative symptoms in drug-naive patients with a first-episode psychosis (FEP). *Asian J Psychiatr.* 2023 Mar;81:103448. FI:9.5 (D1)
- Díaz-Cutraró L, García-Mieres H, Dimaggio G, Lysaker P, Moritz S, Ochoa S. Metacognition in psychosis: What and how do we assess it? *Span J Psychiatry Ment Health.* 2023 Jul-Sep;16(3):206-207. FI:9.2 (D1)
- Butjosa A, Usall J, Vila-Badia R, Mezquida G, Cuesta MJ, Rodríguez-Toscano E, Amoretti S, Lobo A, González-Pinto A, Espliego A, Corripio I, Vieta E, Baeza I, Bergé D, Bernardo M; PEPs Group. Impact of traumatic life events on clinical variables of individuals with first-episode psychosis and healthy controls. *Int J Soc Psychiatry.* 2023 Feb;69(1):134-145. FI:7.5 (Q1)
- Vera-Montecinos A, Rodríguez-Mias R, Vila E, Villén J, Ramos

B. Analysis of networks in the dorsolateral prefrontal cortex in chronic schizophrenia: Relevance of altered immune response. *Front Pharmacol.* 2023 Mar 23;14:1003557. FI: 5.6 (Q1)

SELECTED PROJECTS

- Modelización de enfermedades hereditarias de retina en ratón y organoides para explorar estrategias de terapia genética. PID2022-1409570B-I00. Ministerio de Ciencia e Innovación (MICINN). Convocatoria 2022 de ayudas a «Proyectos de Generación de Conocimiento». Modalidad Proyectos de «Investigación Orientada». 2023-2026. IP:Gemma Marfany Nadal.
- La UB divulga 2023-2024. FCT-22-18085. FECYT - Fundación Española para la Ciencia y la Tecnología. Convocatòria d'ajuts per al foment de la cultura científica, tecnològica i de la innovació. 2023-2026. IP:Gemma Marfany Nadal.
- La ciència accessible: Cartipassos i Focus. 23S02589-001. Ajuntament de Barcelona. Subvencions per a dur a terme projectes, activitats i serveis de districte i de ciutat, per a lany 2023. 2023-2023. IP:Gemma Marfany Nadal.

Diagnostic and Therapeutic Imaging

Emilio Hinarejos, MD, PhD & Sandra Bernaus

6 PROJECTS
6 competitive projects
0 non competitive projects

0 CLINICAL TRIALS & OBSERVATIONAL STUDIES

21 PUBLICATIONS

95 IMPACT FACTOR
4.5 average impact factor

48% PUBLICATIONS IN 1ST QUARTILE

29% PUBLICATIONS IN 1ST DECILE

29% LEADERSHIP

MEMBERS

STAFF RESEARCHER

Ignasi Barber Martínez de la Torre, Marta Gómez Chiari, Mónica Rebollo Polo

ASSISTANT RESEARCHER

Martí Engli Rueda, Christian Mata Miquel

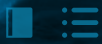
SELECTED PUBLICATIONS

- Valls-Estève A, Lustig-Gainza P, Adell-Gomez N, Tejo-Otero A, Engli-Rueda M, Julian-Alvarez E, Navarro-Sureda O, Fenollosa-Artés F, Rubio-Palau J, Krauel L, Munuera J. A state-of-the-art guide about the effects of sterilization processes on 3D-printed materials for surgical planning and medical applications: A comparative study. *Int J Bioprint.* 2023 May 17;9(5):756. FI: 8.4 (Q1)
- Inarejos Clemente EJ, Diaz Leyva J, Karakas SP, Duarte AM, Mas TR, Restrepo R. Radiologic and Clinical Features of Infantile Hemangioma: Potential Pitfalls and Differential Diagnosis. *Radiographics.* 2023

- Nov;43(11):e230064. FI: 5.5 (Q1)
- Inarejos Clemente EJ, Barber I, Navallas Irujo M, Ladera E, Sousa P, Salas B, Fernández CV, Rodríguez-Fanjul J, Navarro OM. US for Evaluation of Acute Abdominal Conditions in Neonates. *Radiographics*. 2023 Feb;43(2):e220110. FI: 5.5 (Q1)
 - Palau P, Solanes A, Madre M, Saez-Francas N, Sarró S, Moro N, Verdolini N, Sanchez M, Alonso-Lana S, Amann BL, Romaguera A, Martin-Subero M, Fortea L, Fuentes-Claramonte P, García-León MA, Munuera J, Canales-Rodríguez EJ, Fernández-Corcuera P, Brambilla P, Vieta E, Pomarol-Clotet E, Radua J. Improved estimation of the risk of manic relapse by combining clinical and brain scan data. *Span J Psychiatry Ment Health*. 2023 Oct-Dec;16(4):235-243. FI: 9.2 (D1)

SELECTED PROJECTS

- Artificial intelligence (AI) based - Cardiovascular Risk in ONcological pediatric patients assessment Tools. (TED2021-132025B-C44). Ministerio De Ciencia E Innovacion. 2022-2024. IP: Arnau Valls Esteve, Jose Luis Munuera del Cerro.
- Artificial intelligence (AI) based - Cardiovascular Risk in ONcological pediatric patients assessment Tools. (TED2021-132025B-C44). Ministerio De Ciencia E Innovacion. 2022-2024. IP: Arnau Valls Esteve, Jose Luis Munuera del Cerro.
- Valoración de la reabsorción ósea fisiológica del injerto alveolar óseo secundario mediante TAC de baja dosis en pacientes con fisura. *Sociedad Española de Reumatología*. 2021-2024. IP: Marta Gómez Chiari.
- Sistema de ayuda a la decisión clínica para la evaluación y predicción de enfermedad vascular cerebral pediátrica: Radiómica a partir de RM vascular e Inteligencia Artificial (Vasculómica). (PI20/00296). Instituto de Salud Carlos III (ISCIII). 2021-2023. IP: Jose Luis Munuera del Cerro.
- Plataforma ISCIII de apoyo a la I+D+I en Biomedicina y Ciencias de la Salud. Biomodelos-Impresión 3D. (PT20/00090). Instituto de Salud Carlos III (ISCIII). 2021-2024. IP: Arnau Valls Esteve, Jose Luis Munuera del Cerro.



7.1

MEDICINE AND PEDIATRIC SCIENCE AREA

7.1.5

Associated Clinical Research Groups

Paediatric Chronicity and Palliative Care
Orthopaedic Surgery and Traumatology
Environment Effects on Child/Adolescent Well-being
Hospital Dentistry and Periodontal Medicine
Paediatric Ophthalmology
Nursing Research

Paediatric Chronicity and Palliative Care

Sergi Navarro Vilarrubí, MD, PhD



4
PROJECTS
4 competitive projects
0 non competitive projects



0
CLINICAL TRIALS &
OBSERVATIONAL STUDIES



5
PUBLICATIONS



14
IMPACT FACTOR
2.7 average impact factor



40%
PUBLICATIONS IN 1ST QUARTILE



0%
PUBLICATIONS IN 1ST DECILE



60%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

XXX

PHD STUDENTS

XXX

COLLABORATOR RESEARCHER

XXX

In 2023 our team was formally constituted as a clinical research associated group, "Chronicity and paediatric palliative care", in the SJD research institute. The driving force of our group is to improve the quality of life of children and adolescents with serious and/or life-limiting illnesses by increasing knowledge in this field. This group is essentially interdisciplinary, involving professionals from the following areas: paediatricians, nurses, psychologists, spiritual advisors and data analysts.

While we are primarily a clinical group, dedicated to the care of children with complex chronic illnesses and palliative needs, in 2023 we achieved

an important milestone: we obtained a Horizon Europe grant with the project "PALLIAKID: Comprehensive Paediatric Palliative Care Approach" (Grant Agreement 101137169). It is a collaborative European project of 4.5 years of duration, with 17 partners of 10 European countries, coordinated by FSJD and funded with 7M euros. PALLIAKID is an interdisciplinary project aiming to evaluate the feasibility, effectiveness, and cost-effectiveness of novel interventions for children and young people with palliative and end-of-life care needs in different healthcare systems across Europe, focusing on those factors that affect the active engagement of patients and family caregivers.

A new research project for home monitoring of chronic complex patients, "Continuous monitoring of paediatric chronic patients through a Clinical Command Centre as a linking environment with Primary Care; C3AP" started in 2023 in close collaboration with primary care centres. We also continued our involvement in pilot 3 of Aiccelerate project, funded by Horizon Europe grant, where we collaboratively developed e-health solutions (social robot and App) for improving the care of children with severe neurologic conditions at home. Furthermore, Daniel Toro published an important paper in European Journal of Pediatrics analysing quality of life in paediatric palliative care through children's and parents' perspectives.

SELECTED PUBLICATIONS

- Toro-Pérez D, Limonero JT, Guillen M, Bolancé C, Vilarrubí SN, Camprodón-Rosanas E. Evaluating quality of life in pediatric palliative care: a cross-sectional analysis of children's and parents' perspectives. *Eur J Pediatr.* 2024 Mar;183(3):1305-1314. FI:3.6 (Q1)
- Toro-Pérez D, Camprodón-Rosanas E, Navarro Vilarrubí S, Bolancé C, Guillen M, Limonero JT. Assessing well-being in pediatric palliative care: A pilot study about views of children, parents and health professionals. *Palliat Support Care.* 2023 Mar 24;1-9. FI:2.2 (Q3)
- Popenhagen MP, Genovese P, Blishen M, Rajapakse D, Diem A, King A, Chan J, Pellicer Arasa E, Baird S, Ferreira da Rocha AC, Stitt

G, Badger K, Zmazek V, Ambreen F, Mackenzie C, Price H, Roberts T, Moore Z, Patton D, Murphy P, Mayre-Chilton K. Consensus-based guidelines for the provision of palliative and end-of-life care for people living with epidermolysis bullosa. *Orphanet J Rare Dis.* 2023 Sep 4;18(1):268. FI:3.7 (Q2)

- de la Vega R, Serrano-Ibáñez ER, Ruiz-Párraga GT, Palermo TM, Wicksell R, Fernández-Jiménez E, Oliva S, Roldán S, Monfort L, Peláez MJ, Leyva Carmona M, Ramírez-Maestre C, López-Martínez AE, Miró J, Esteve R. Protocol for a multi-phase, multi-center, real-world, hybrid effectiveness-implementation study of a digital intervention for pediatric chronic pain co-designed with patients (Digital SPA). *Digit Health.* 2023 Dec 19;9:20552076231219490. FI:3.9 (Q1)

SELECTED PROJECTS

- PALLIAKID_COMPREHANSIVE PAEDIATRIC PALLIATIVE CARE APPROACH. (101137169). European Commission. 2023-2028. IP:Sergi Navarro Vilarrubí.
- AICCELERATE_AI Accelerator – A Smart Hospital Care Pathway Engine. (101016902). European Commission. 2021-2024. IP:Arnau Valls Esteve, Sergi Navarro Vilarrubí, Joan Sánchez de Toledo.

Orthopaedic Surgery and Traumatology

César García Fontecha, MD, PhD



10
PROJECTS
10 competitive projects
0 non competitive projects



4
CLINICAL TRIALS &
OBSERVATIONAL STUDIES
3 commercial clinical trials
1 academic clinical trials



2
PUBLICATIONS



5
IMPACT FACTOR
2.6 average impact factor



50%
PUBLICATIONS IN 1ST QUARTILE



0%
PUBLICATIONS IN 1ST DECILE



100%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Antoni Compte Verdaguer, Laura Corominas Francés, Lydia De Sena De Cabo, Dimosthenis Kiimetoglou, David Moreno Romo, Laura Montserrat Pérez López, Melisa Stitzman Wengrowicz, Ferran Torner Rubies, Alejandro Peiró García, Maria Pilar Rovira Marti

PHD STUDENTS

Rocio Garcia Garcia

ASSISTANT RESEARCHER

Gerard Marturià Navarro

SELECTED PUBLICATIONS

- Moreno Mateo F, Bovonratwet P, Peiró García A. Early-onset scoliosis. *Curr Opin Pediatr.* 2024 Feb 1;36(1):105-111. FI: 3.6 (Q1)
- Parada-Avedaño I, Salvador H, García RG, Martorell-Sampol L, Fontecha CG, Torner-Rubies F, Perez-Lopez LM. Lateralized overgrowth as a guiding sign of abdominal neoplasms for pediatric

orthopedic surgeons. *Jt Dis Relat Surg.* 2023;34(1):3-8. FI: 1.6 (Q3)

- Parada-Avedaño I, Salvador H, García RG, Martorell-Sampol L, Fontecha CG, Torner-Rubies F, Perez-Lopez LM. Lateralized overgrowth as a guiding sign of abdominal neoplasms for pediatric orthopedic surgeons. *Jt Dis Relat Surg.* 2023;34(1):3-8. FI: 1.6 (Q3)
- Perez-Lopez LM, Perez-Abad M, Suarez Merchan MA, Cabrera Ortiz DA. Reverse Ishiguro Extension Block Technique as an Alternative for Irreducible Osseous Mallet Finger. *Tech Hand Up Extrem Surg.* 2024 Jun 1;28(2):62-66.

SELECTED PROJECTS

- Ensayo clínico aleatorizado comparativo del corsé diurno y nocturno. Sociedad para el estudio de las enfermedades del raquis (GEER). 2023-2027. IP:Alejandro Peiró García.
- SGR 2022-2024_Cirurgia Ortopèdia i Traumatologia Pediàtrica. [2021 SGR 00793]. Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. 2022-2025. IP:Laura Montserrat Pérez López.
- Comparison of the anatomical-functional state of the cerebral cortex before and after treatment of congenital pathology of the pediatric upper extremity. Federation of European Societies for Surgery of the Hand (FESSH). 2023-2024. IP:Laura Montserrat Pérez López.
- Computational model of guided growth in immature skeleton for custom-made correction of deformities. [PI20/00293]. Instituto de Salud Carlos III (ISCIII). 2021-2024. IP:César Galo García Fontecha.

SELECTED CLINICAL STUDIES

- A Phase 2, open-label, multicenter, 2-stage sequential cohort, dose escalation study to assess the safety, tolerability, pharmacokinetics, pharmacodynamics, and efficacy of subcutaneous SAR442501 in pediatric participants with Achondroplasia. [DRI16646]. Diagnóstico para la imagen (DXI); Farmacia; Laboratori HSJD; Pena; Sanofi-Aventis S.A; Servicio Oftalmología; SIMM Molecular S.L.; Unitat de Recerca (URC) FSJD. 2023.

IP:César Galo García Fontecha.

- ACHieve: estudio observacional, longitudinal y multicéntrico de niños/as con acondroplasia. (TCC-NHS-01). Ascendis Pharma Growth Disorders A/S; Southwood Research Ltd; Veristat, LLC. 2020. IP:César Galo García Fontecha.

Environment Effects on Child/Adolescent Well-being

Carles Luaces Cubells, MD, PhD

2
CLINICAL TRIALS &
OBSERVATIONAL STUDIES
1 commercial clinical trials
1 academic clinical trials

3
PUBLICATIONS

11
IMPACT FACTOR
3.6 average impact factor

33%
PUBLICATIONS IN 1ST QUARTILE

33%
PUBLICATIONS IN 1ST DECILE

67%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Gemma Claret Teruel, Ana Isabel Curcoy Barcenilla, Lidia Martínez Sánchez, Victoria Trenchs Sainz de la Maza

POSTDOC ASSOCIATED RESEARCHERS

Andrea Aldemira Liz, Vanessa Arias Constantí, Eva Gargallo Burriel, Cristina Parra Cotanda

PHD STUDENTS

David Muñoz Santanach, Sergi Navarro Vilarrubi, Marta Simó Nebot

SELECTED PUBLICATIONS

- Curcoy AI, Serra A, Morales M, Luaces C, Trenchs V. Retinal hemorrhages due to intracranial hypertension, is it possible to differentiate them from those associated with abuse? Child Abuse Negl. 2023 Oct;144:106387. FI: 4.8 [D1]
- Díaz-Miguel M, Martos Utande A, Trenchs Sainz de la Maza V. Impact of COVID-19 pandemic in oncological debut at Pediatric Emergency Department. Med Clin (Barc). 2023 Nov 24;161(10):450-451.

English, Spanish. FI: 3.9 (Q2)

- Arias Constanti V, Domingo Garau A, Rodríguez Marrodán B, Villalobos Pinto E, Rianza Gómez M, García Soto L, Hernández Borges Á, Madrid Rodríguez A. Do not do recommendations in different paediatric care settings. An Pediatr (Engl Ed). 2023 Apr;98(4):291-300.
- Romero RG, Riera LB, Marcos NS, Constanti VA, Maza VTS, Luaces C. New-onset type 1 diabetes mellitus in the Paediatric Emergency Department: impact of the COVID-19 pandemic. Ann Pediatr Endocrinol Metab. 2023 Sep;28(3):215-218.

SELECTED CLINICAL STUDIES

- RSV in Hospital Emergency Departments in Iberia (RHEDI) Study. (RHEDI). 2022. IP:Carles Luaces Cubells.
- Evaluación del uso de FebriDx en el Entorno Pediátrico y en el Manejo de Niños con Neumonía Adquirida en la Comunidad. (FebriDx). 2022. IP:Carles Luaces Cubells.

Hospital Dentistry and Periodontal Medicine

Alejandro Rivera Baró, MD, PhD

1
PROJECTS
1 competitive projects
0 non competitive projects

0
CLINICAL TRIALS &
OBSERVATIONAL STUDIES

2
PUBLICATIONS

5
IMPACT FACTOR
2.7 average impact factor

50%
PUBLICATIONS IN 1ST QUARTILE

0%
PUBLICATIONS IN 1ST DECILE

50%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Mireia Aznar Gómez, Lluís Brunet Llobet, Jaume Miranda Rius, Jordi Moncunill Mira, Maria Lluïsa Tobella Camps

During 2023, we have given continuity to the research line: "Periodontal disease in pregnant women associated with preterm delivery". After the reading of two Doctoral Theses, one international (KCMC-Moshi-Tanzania) and the other in our Hospital, in 2023 we have published an original article in Oral Diseases (Q1). *Miranda-Rius J, Brunet-Llobet L, Blanc V, Álvarez G, Moncunill-Mira J, Mashala EI, Kasebele Y, Masenga G, Nadal A, León R. Microbial profile of placentas from Tanzanian mothers with adverse pregnancy outcomes and periodontitis. Oral Dis. 2023 Mar;29(2):772-785.*

The objective was to investigate microbial profiles in placentas from

a population of East African mothers with and without adverse pregnancy outcomes and with regard to their periodontal status. We concluded that periodontal pathogens may involve the feto-placental unit, and the structure of the microbial community found in the placenta. Maternal periodontal status was associated with a clear differentiation in the microbiota of the placentas, and several oral bacteria present in this niche were associated with adverse pregnancy outcomes. Now, continuing this research line addressing the theme/topic, chronic oral infectious-inflammatory process with systemic repercussions, we also propose to analyze biomolecular products derived from periodontal disease in umbilical cord blood/placental tissue and correlate them with spontaneous preterm birth.

We have another project, related with periodontology, which studies prognostic factors of traumatized and transplanted teeth with CEIm authorization: PS-01-23.

Last September our group organized the 10th edition of Hospital Pediatric Dentistry course entitled: Autism spectrum disorder. Update on oral surgery protocols.

Finally, it should be noted that the digital repository of the University of Barcelona has published our original article with the Miranda & Brunet index (MBi). It has been assigned the following identifier: <http://hdl.handle.net/2445/209883>

SELECTED PUBLICATIONS

- Miranda-Rius J, Brunet-Llobet L, Blanc V, Álvarez G, Moncunill-Mira J, Mashala El, Kasebele Y, Masenga G, Nadal A, León R. Microbial profile of placentas from Tanzanian mothers with adverse pregnancy outcomes and periodontitis. *Oral Dis.* 2023 Mar;29(2):772-785. (D1).
- Perandones-González H, Rusiñol-Batlle L, Bosquez D, Brunet-Llobet L, Ivars M, Yubero D, Sarig O, Malki L, Peled A, Sprecher E, Baselga E. Woolly hair in tricho-dento-osseous syndrome. *Pediatr Dermatol.* 2023 Nov-Dec;40(6):1094-1096. (Q4)

SELECTED PROJECTS

- SGR 2022-2024_Hospital Dentistry & Periodontal Medicine Research

Group. (2021 SGR 00663). Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. 2022-2025. IP:Lluís Brunet Llobet.

Paediatric Ophthalmology

Joan Prat Bartomeu, MD, PhD

1 PROJECTS
1 competitive projects
0 non competitive projects

5 CLINICAL TRIALS & OBSERVATIONAL STUDIES
4 commercial clinical trials
1 academic clinical trials

7 PUBLICATIONS

24 IMPACT FACTOR
3.4 average impact factor

29% PUBLICATIONS IN 1ST QUARTILE

14% PUBLICATIONS IN 1ST DECILE

29% LEADERSHIP

MEMBERS

STAFF RESEARCHER

Elisa Carreras Bertran, Ester Casas Gimeno, Jaume Català Mora, Jesús Díaz Cascajosa, Ana Isabel Díaz Cortés, Carlos Fresno Cañada, Carmen García de Vicuña Muñoz, Enrique Jimenez Gañan, Ana Llorca Cardeñosa, Marta Morales Ballús, Núria Planas Domènech, M^a Josep Planas Tapias, Enric Puigventós, Gemma Romeu Cerrillo, Alicia Serra Castanera, Dowon, Simone Suh, Begonya Yeste Mayoral

During 2023, our research group has had a certain continuity in relation to previous years, of special relevance are: 1) the assessment of the effect of intraocular gene therapy in patients with retinal dystrophies 2) the treatment of retinoblastoma using improved oncolytics viruses that can prevent ocular enucleation, 3) genetic studies of different rare ophthalmological diseases. A new line are the preliminary studies of the use of hyaluronate as a palpebral and

orbital tissue expander in children with developmental anomalies in these territories. Regarding innovation, the developments are varied. First, the assessment with a virtual reality device of how children with severe retinopathy can move through a room with obstacles. Second, the control of visual acuity by telematics through a smartphone application to have closer monitoring of children with amblyopia of different causes. Third, the diagnosis of the different types of retinopathy of prematurity through AI of photographs taken in the last decade of the children's eyes.

SELECTED PUBLICATIONS

- Curcoy AI, Serra A, Morales M, Luaces C, Trenchs V. Retinal hemorrhages due to intracranial hypertension, is it possible to differentiate them from those associated with abuse? *Child Abuse Negl.* 2023 Oct;144:106387. FI: 4.8 [D1]
- Rego-Lorca D, Català-Mora J, López-de-Eguileta A, Díaz-Cascajosa J. Spontaneous Regression of Choroidal Neovascularization in a Girl with Rubella Retinopathy. *Ocul Immunol Inflamm.* 2023 Dec;31(10):2069-2071. FI: 3.3 [Q2]
- Cuadrado-Vilanova M, Burgueño V, Balaguer-Lluna L, Aschero R, Castillo-Ecija H, Liu J, Perez-Jaume S, Pascual-Pasto G, Olaciregui NG, Gomez-Gonzalez S, Correa G, Suñol M, Schaiquevich P, Radvanyi F, Lavarino C, Mora J, Català-Mora J, Chantada GL, Carcaboso AM. Follow-up of intraocular retinoblastoma through the quantitative analysis of conserved nuclear DNA sequences in aqueous humor from patients. *J Pathol Clin Res.* 2023 Jan;9(1):32-43. FI:4.1 [Q2]
- Tomar AS, Finger PT, Gallie B, Kivelä T, Mallipatna A, Zhang C, Zhao J, Wilson M, Brennan R, Burges M, Kim J, Berry JL, Jubran R, Khetan V, Ganeshan S, Yarovoy A, Yarovaya V, Kotova E, Volodin D, Yousef Y, Nummi K, Ushakova TL, Yugay OV, Polyakov VG, Ramirez-Ortiz MA, Esparza-Aguilar E, Chantada GL, Schaiquevich P, Fandiño AC, Yam JC, Lau WW, Lam CP, Sharwood P, Moorthy S, Long QB, Essuman VA, Renner LA, Semenova E, Català-Mora

J, Correa Llano M, Carreras E; American Joint Committee on Cancer Ophthalmic Oncology Task Force; American Joint Committee on Cancer Ophthalmic Oncology Task. Retinoblastoma seeds: impact on American Joint Committee on Cancer clinical staging. *Br J Ophthalmol.* 2023 Jan;107(1):127-132. FI:4.1 [Q1]

SELECTED PROJECTS

- Desenvolupament d'un equip de realitat virtual per l'avaluació i seguiment de la visió funcional en pacients amb distròfies hereditàries de retina abans i després del tractament amb teràpies intraoculars avançades. (2021 LLAU 00019). Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. 2022-2023. IP:Jaume Català Mora.

SELECTED CLINICAL STUDIES

- A Phase 3, multicenter, randomized, evaluator-blinded clinical trial to assess the safety and efficacy of Clobetasol propionate ophthalmic nanoemulsion, 0.05% compared to Prednisolone acetate, 1% in the treatment of inflammation after cataract surgery in pediatric population 0 to 3 years of age (CLOSE-3). (CLOBOF3-17IA03). del Pino; Farmacia; Laboratorios Salvat, S.A.; Unitat de Recerca (URC) FSJD. 2022. IP:Marta Morales Ballús.
- A randomized, 2-arm, open-label, phase 3 study to evaluate the efficacy, safety, and tolerability of intravitreal aflibercept versus laser photocoagulation in patients with retinopathy of prematurity (ROP). (BAY 86-5321/20090). Bayer AG. 2019. IP:Alicia Serra Castanera.
- Ensayo clínico fase I, unicéntrico, abierto y de escalado de dosis, para evaluar la seguridad y la actividad del adenovirus oncolítico VCN-01 en pacientes con retinoblastoma refractario. (FSJD-RTB-2015). Anatomía patológica; Atienza; Diagnóstico para la imagen (DXI); Farmacia; Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD; Hospitalización HSJD; Laboratori HSJD; SERVICIO DE CIRUGIA; Servicio Neurofisiología; Servicio Oftalmología; Theriva Biologics SL; Unitat de Recerca (URC) FSJD. 2017. IP:Jaume Català Mora.

- Estudio postautorización, multicéntrico, multinacional, longitudinal y observacional para el registro de la seguridad en pacientes tratados con voretigén neparvovec. (CLTW888A12401/NOV-LUX-2019-01). Novartis Farmacéutica, S.A. 2020. IP:Jaume Català Mora.

Nursing Research

Montserrat Gutierrez



1

PROJECTS

1 competitive projects

0 non competitive projects



0

CLINICAL TRIALS &
OBSERVATIONAL STUDIES



0

PUBLICATIONS

MEMBERS

STAFF RESEARCHER

Maribel Ferrer Orona, Concepción Agustín Mata, Silvia Ciprés Roig, Encarna Gómez Gamboa, Jordi Mitjà, Gemma Pérez Acevedo

POSTDOC ASSOCIATED RESEARCHERS

M^a Dolors Navarro Rubio

PHD STUDENTS

Marta Fajardo Samper

ASSISTANT RESEARCHER

Pau Miquel Diego, Ikram Allaoui, Montserrat Gutierrez Juarez, Ares Sentenach Carbó

COLLABORATOR RESEARCHER

Carmen Jerez Molina

During 2023, it has been possible to participate in the analysis for the creation of the IRSJD Strategic Plan. After presentation of the same and Nursing SWOT, the appropriate strategy is configured for the development of a *Contingency Plan and monitoring in nursing research*, with a view to growing, adapting and impacting. It has been possible to capture a first round among relevant researchers, the new configuration of the nursing staff for the Grup Clinic Associats. Their credentials have been presented and the group's objectives have been reconsidered, respecting the established lines of research and always keeping in mind the current health needs of the population.

In relation to scientific production, our team has been awarded a competitive project. Dr Eva M. Luján leads the project: *Post-accompanying:*

communities of support for a sustainable transition of young people with mental health problems to adult life. This contributes significantly to enhancing the continuity of care and promoting the degree of functional autonomy necessary for the Mental Health patient.

Of the publications of scientific articles, the validation and cross-cultural adaptation to the Spanish version of the *Diabetes Management Questionnaire* (DMQ-Sp) stands out, which evaluates the degree of adherence to self-care in children with DM1. Another project that deals with obtaining improvements in the care of very low birth weight (VLBW) babies admitted to neonatal intensive care units (NICU), who suffer consequences of stress in actions such as weight recording. Maintaining adequate self-regulation during the weighing of these babies, such as weighing with diapers, demonstrates the impact of a less stressful intervention of the procedure.

Also notable is the start of international participation in the #ViernesCientíficosSJD, with the presence of Dra Maria Brenner, Professor of Children's Nursing UCD School of Nursing, Midwifery and Health Systems at University College Dublin.

SELECTED PROJECTS

- Post- acompanya'm: comunitats de suport per a una transició sostenible dels/les joves amb problemes de Salut Mental cap a la vida adulta. [DDS008/22/000240]. Generalitat de Catalunya. 2023-2025. IP:Eva Maria Luján Luján.
- Les narratives clíniques de la mort: una oportunitat per al desenvolupament ètic dels professionals?. [NARRA23]. Fundació Víctor Grifols i Lucas; Miquel. 2021-2024. IP:Pau Miquel Diego.



7.2 MENTAL HEALTH AREA

7.2.1 Mental Health and Society

Impact and Prevention of Mental Disorders
Health Technology Assessment in Primary Care and Mental Health
(PRISMA)
P01-15 Psychological Research in Fibromyalgia and Chronic Pain
(ÅGORA Group)

Impact and Prevention of Mental Disorders

Josep Maria Haro Abad, MD, PhD



35
 PROJECTS
 32 competitive projects
 3 non competitive projects



1
 CLINICAL TRIALS &
 OBSERVATIONAL STUDIES
 1 commercial clinical trials
 0 academic clinical trials



157
 PUBLICATIONS



1090
 IMPACT FACTOR
 6.9 average impact factor



61%
 PUBLICATIONS IN 1ST QUARTILE



29%
 PUBLICATIONS IN 1ST DECILE



40%
 LEADERSHIP

MEMBERS

STAFF RESEARCHER

Louis Etienne Charles Jacob, Mireia Féllez Nóbrega, Paula Cristobal Narvaez, Sara Siddi, Stefanos Tyrovolas

POSTDOC ASSOCIATED RESEARCHERS

Rodrigo Antunes Lima, Joan Domènech Abella, Beatriz Olaya Guzmán

PHD STUDENTS

Elena Condominas Casanovas, Carlota de Miquel Bleier, Anna Monistrol Mula

ASSISTANT RESEARCHER

Mireia Esplugues Tormo, Clara Szymanski Cortés, Helena Sainz Elias, Andrea Estefanía Miranda Mendizabal, Leticia Gonzalez Spinoglio, Emilio Tomas Lavilla, Fajar Matloob Butt, Rachid El Hafi Elmokhtari, Marta Franch Roca, Lydia Hanaa Faris, Aina Gabarrell Pascuet, Desirée Gutiérrez Marin, Iago Giné Vázquez, Ivet Bayes Marin, Blanca Mellor Marsa, Maria Victoria Moneta, Ziggi Santini, Elisabeth CW Vorstenbosch

MANAGEMENT

Silvena Pashova Velikova, Iñigo Chivite

Araiz, Judit Lopez Luque, Iris Mauricio Arribas, Carla Obradors Tarragó

During the year 2023, our team continued to advance in various lines of scientific research, with the aim of contributing to knowledge in the field of promoting emotional well-being and preventing mental disorders. Below are some of the highlighted studies and milestones achieved during this period:

Our team has focused on several competitive and private projects aimed at evaluating the impact of mental health problems and defining strategies to prevent them. These projects have strengthened our research in mental health and migration, mental health in the workplace, mental health in the elderly, and adolescent mental health.

Among the most notable achievements, we highlight the initiation of the development of the IMPROVA project on adolescent mental health and the MIO project in the migrant population.

During the year 2023, we continued our activity in the diagnosis of mental disorders, specifically a genomic test for the diagnosis of bipolar disorder.

Throughout the year 2023, our team has significantly contributed to the body of scientific knowledge through the publication of various articles in specialized journals. These include a study on mortality in people with mental disorders and the impact of COVID on mental health.

During this period, we have strengthened our collaborations with other research groups and academic entities, which has enriched our research and facilitated the achievement of important scientific milestones.

SELECTED PUBLICATIONS

- de Miquel C, Moneta MV, Weber S, Lorenz C, Olaya B, Haro JM. The Mediating Role of General and Cognitive Stress on the Effect of an App-Based Intervention on Productivity Measures in Workers: Randomized Controlled Trial. J Med Internet Res. 2023 Jul 3;25:e42317. FI:7.4 (D1)

- Domènech-Abella J, Gabarrell-Pascuet A, García-Mieres H, Mortier P, Felez-Nobrega M, Cristóbal-Narváez P, Vilagut G, Olaya B, Alonso J, Haro JM. Loneliness during the last phase of the COVID-19 pandemic in Spain: A longitudinal study of group-based trajectories, risk factors, and consequences in mental health. Psychiatry Res. 2023 Aug;326:115327. FI:11.3 (D1)
- Olaya B, Moneta MV, Plana-Ripoll O, Haro JM. Association between mental disorders and mortality: A register-based cohort study from the region of Catalonia. Psychiatry Res. 2023 Feb;320:115037. FI:11.3 (D1)
- Faris LH, Gabarrell-Pascuet A, Felez-Nobrega M, Cristóbal-Narváez P, Mortier P, Vilagut G, Olaya B, Alonso J, Haro JM, López-Carrilero R, Domènech-Abella J. The Association Between Substance Use Disorder and Depression During the COVID-19 Lockdown in Spain and the Moderating Role of Social Support: a Cross-Sectional Study. Int J Ment Health Addict. 2023;21(2):1157-1167. FI:8 (D1)

SELECTED PROJECTS

- EMPOWER_European platforM to PromOte Wellbeing and HHealth in the woRkplace. (848180). European Commission; Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD. 2020-2024. IP:Beatriz Olaya Guzmán.
- MIO: Migration Impact On health. (SR21-00779). Fundació Bancària "La Caixa". 2021-2024. IP:Josep Maria Haro Abad.
- IMPROVA - e-Intervention Enhancing Mental Health in Adolescents. (10108093). Antunes; European Commission; Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD. 2023-2027. IP:Josep Maria Haro Abad.
- "INTEGRATE-LMedC_Concept development for a research infrastructure to manage, integrate and sustain large medical cohort studies". 2023-2024. IP: Josep Maria Haro Abad.


SELECTED CLINICAL STUDIES


- Estudio de validación clínica para la prueba EDIT-B: una ayuda para el

diagnóstico diferencial del trastorno bipolar, basado en biomarcadores sanguíneos de edición de ARN. Clinical validation study for EDIT-B test: an aid for differential diagnosis of bipolar disorder, based on RNA editing blood biomarkers. EO202211. Alcediag. 2022 - . IP: Josep Maria Haro Abad.


Health Technology Assessment in Primary Care and Mental Health (PRISMA)


Maria Rubio Valera, PhD & Antoni Serrano Blanco, MD, PhD


 **26**
PROJECTS
25 competitive projects
1 non competitive projects

 **1**
CLINICAL TRIALS & OBSERVATIONAL STUDIES
1 commercial clinical trials
0 academic clinical trials

 **23**
PUBLICATIONS

 **160**
IMPACT FACTOR
6.9 average impact factor

 **65%**
PUBLICATIONS IN 1ST QUARTILE

 **26%**
PUBLICATIONS IN 1ST DECILE

 **26%**
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Claudia Palma Vázquez, Alexandra Lelia Dima, Víctor Chavarría Romero, Luisa Baladon Higuera, Vanessa Barneda, Maria Iglesias Gonzalez, Cristina López Ortiz, Enric Vicens Pons

POSTDOC ASSOCIATED RESEARCHERS

Mercè Soler Font, Maria Montserrat Gil Girbau, Carlos Peña Salazar, Ignacio Aznar Lou

PHD STUDENTS

Alba Sanchez Viñas, Cristina Carbonell Duacastella

TECHNICIAN

Marcos Antonio Catalan Vega

ASSISTANT RESEARCHER

Marina Fuente Moreno, Ana María Besoain Cornejo, Paula Arroyo Uriarte, Carmen Corral Partearroyo

COLLABORATOR RESEARCHER

Salvatore Fabrizio Contaldo, Maria del Carmen Gallardo González,

Silvia Ramírez Perdomo, Montserrat Iracheta Todó, Ana Fernández Sánchez, Maria Antonia March Pujol, Juan Manuel Mendive Arbeloa, Maria Teresa Peñarrubia Maria, Ramon Sabes Figuera, Luís Salvador Carulla

In 2023, the Catalan research authorities recognized our research group as a consolidated research group. We have continued providing evidence to decision makers in mental health, working in several projects, including: a) the support for the development of the National Mental Health Strategy and the prioritisation of their policy and planning strategies; b) the evaluation of an intervention to increase resilience in adolescents in schools to prevent mental disorders as part of the HENKA project; c) the design and development of the BEA project, “*Improvement of Mental Wellbeing of Adolescents Participating in Activities at the Community*”; and d) mapping health and social services in the region of Osona for the Barcelona Provincial Council.

Additionally, we have been granted a new European project (H2020), which aims to improve the management of patients with poor prognosis in ovarian cancer using new interventions and shared-decision making. The B-LONG project has been completed, detecting a high proportion of benzodiazepine chronic users in the community and performing a social theatre to increase awareness and disseminate results to the general population. Finally, the follow-up of the IMA-cRCT project, funded by an ERC Starting Grant, has been completed, recruiting over 4,000 patients. The process evaluation of the study has been completed, demonstrating that a shared-decision making intervention is feasible and acceptable in primary care and giving insight to translate the intervention.

SELECTED PUBLICATIONS

- Torres M, Serra-Sutton V, Soriano JB, Ferrer M, Trejo A, Benavides FG, Lumbreras B, Pérez-Gómez B, Pijoan JI, Monguet JM, Barbé F, Pollán M, Alonso J, Espallargues M, Serrano-Blanco A. Consensus on post COVID in the Spanish national health system: Results of the CIBERPOSTCOVID eDelphi study. J Infect Public Health. 2023 Nov;16(11):1784-1792. FI:6.7 (Q1)
- Parody-Rua E, Bustamante A,

Montaner J, Rubio-Valera M, Serrano D, Pérez-Sánchez S, Sánchez-Viñas A, Guevara-Cuellar C, Serrano-Blanco A. Modeling the potential efficiency of a blood biomarker-based tool to guide pre-hospital thrombolytic therapy in stroke patients. *Eur J Health Econ.* 2023 Jun;24(4):621-632. FI:4.4 (Q1)

- Kardas P, Aarnio E, Agh T, van Boven JFM, Dima AL, Ghiciuc CM, Kamberi F, Petrova GI, Nabergoj Makovec U, Trečičkienė I. New terminology of medication adherence enabling and supporting activities: ENABLE terminology. *Front Pharmacol.* 2023 Oct 13;14:1254291. FI:5.6 (Q1)
- Fuente-Moreno M, Garcia-Terol C, Domínguez-Salas S, Rubio-Valera M, Motrico E. Maternity care changes and postpartum mental health during the COVID-19 pandemic: a Spanish cross-sectional study. *J Reprod Infant Psychol.* 2023 Jan 29:1-16. FI:2.5 (Q2)

SELECTED PROJECTS

- IMA-cRCT_Effectiveness and cost-effectiveness of the Initial Medication Adherence intervention: cluster randomized controlled trial and economic model. (948973). European Commission. 2021-2025. IP: Maria Rubio Valera.
- Salvovar: a european multi-disciplinary clinical project meant to improve the management of patients with poor prognostic ovarian cancer after neoadjuvant chemotherapy: restoring hope, salvaging lives. (101104469). European Commission. 2023-2028. IP: Alexandra Lelia Dima.
- Evaluación de la adherencia a antipsicóticos a partir de datos de la práctica clínica real. (P119/00111). Instituto de Salud Carlos III (ISCIII). 2020-2024. IP: Antoni Serrano Blanco.
- Necessitats actuals i reptes de la Taula del Pacte Nacional de Salut Mental: Avaluació qualitativa. Fundació Universitària Balma de Vic. 2022-2023. IP: Antoni Serrano Blanco.

SELECTED CLINICAL STUDIES

- Ensayo clínico aleatorizado, multicéntrico para evaluar la seguridad y tolerabilidad de clozapina vs tratamiento de práctica clínica habitual en psicosis resistente a tratamiento en personas adolescentes y adultos jóvenes con discapacidad

intelectual. (CLOZ-AID). Fundación Pública Andaluza para la Gestión de la Investigación en Salud de Sevilla. 2022. IP: Carlos Peña Salazar.

P01-15 Psychological Research in Fibromyalgia and Chronic Pain (ÀGORA Group)

Juan Vicente Luciano Devis, PhD

10
PROJECTS
10 competitive projects
0 non competitive projects

1
CLINICAL TRIALS &
OBSERVATIONAL STUDIES
0 commercial clinical trials
1 academic clinical trials

26
PUBLICATIONS

151
IMPACT FACTOR
5.8 average impact factor

54%
PUBLICATIONS IN 1ST QUARTILE

19%
PUBLICATIONS IN 1ST DECILE

77%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Jesús Montero Marin, Juan Pablo Sanabria Mazo, Lluís Torruella Barraquer, Eugenia Cardenosa Valera, Antoni Rozadilla Sacanell

POSTDOC ASSOCIATED RESEARCHERS

Adrián Pérez Aranda

PHD STUDENTS

Ariadna Colomer Carbonell

ASSISTANT RESEARCHER

Jaime Navarrete Hidalgo

COLLABORATOR RESEARCHER

Albert Feliu Soler, Xavier Borràs

Hernández, Maria Teresa Navarro Gil

In 2023, the main novelty has been our incorporation into the Biomedical Research Center in Epidemiology and Public Health (CIBERESP; CB22/02/00052), dependent on the

Institute of Health Carlos III (ISCIII).

Next, it is important to highlight that Dr. Jesús Montero Marín, senior researcher Miguel Servet (ISCIII) at the PSSJD, obtained a M-AES mobility grant (ISCIII) and another mobility grant José Castillejo (Ministry of Education) to carry out two stays at the Department of Psychiatry of the University of Oxford. Likewise, the predoctoral researcher PFIS Juan Pablo Sanabria Mazo also obtained a M-AES mobility grant to carry out a 3-month research stay at the London School of Economics (LSE), which has subsequently allowed his doctoral thesis to have international mention. Finally, the predoctoral researcher FI Ariadna Colomer also completed a 3-month stay at the MD Anderson Cancer Center - University of Texas to obtain international mention in her doctoral thesis.

Throughout the year, the group has focused its efforts mainly on the data exploitation of the IMPACT clinical trial, focused on the effectiveness and cost-utility of two third-generation cognitive-behavioral therapies for patients with chronic low back pain and comorbid depression. In addition, we have continued with the data collection work of the INNOVA Project, a randomized clinical trial on the effectiveness and cost-utility compared to placebo of a new pharmacological therapy (low-dose naltrexone) for the management of pain in fibromyalgia. Finally, it is worth highlighting the continuation of our research line on central sensitization in collaboration with Dr. Randy Neblett, clinician and researcher at the Productive Rehabilitation Institute of Dallas for Ergonomics, which is resulting in numerous high-impact scientific publications.

SELECTED PUBLICATIONS

- Montero-Marin J, Hinze V, Mansfield K, Slaghekke Y, Blakemore SJ, Byford S, Dalgleish T, Greenberg MT, Viner RM, Ukoumunne OC, Ford T, Kuyken W; MYRIAD Team. Young People's Mental Health Changes, Risk, and Resilience During the COVID-19 Pandemic. *JAMA Netw Open.* 2023 Sep 5;6(9):e2335016. FI:13.8 (D1)
- Montero-Marin J, Hinze V, Crane C, Dalrymple N, Kempnich MEJ, Lord L, Slaghekke Y, Tudor K; MYRIAD

Team; Byford S, Dalgleish T, Ford TJ, Greenberg MT, Ukoumunne OC, Williams JMG, Kuyken W. Do Adolescents Like School-Based Mindfulness Training? Predictors of Mindfulness Practice and Responsiveness in the MYRIAD Trial. *J Am Acad Child Adolesc Psychiatry.* 2023 Nov;62(11):1256-1269. FI:13.3 (D1)

- Sanabria-Mazo JP, Colomer-Carbonell A, Borràs X, Castaño-Asins JR, McCracken LM, Montero-Marin J, Pérez-Aranda A, Edo S, Sanz A, Feliu-Soler A, Luciano JV. Efficacy of Videoconference Group Acceptance and Commitment Therapy (ACT) and Behavioral Activation Therapy for Depression (BATD) for Chronic Low Back Pain (CLBP) Plus Comorbid Depressive Symptoms: A Randomized Controlled Trial (IMPACT Study). *J Pain.* 2023 Aug;24(8):1522-1540. FI:4 (Q4)
- Sanabria-Mazo JP, Doval E, Bernadàs A, Angarita-Osorio N, Colomer-Carbonell A, Evans-Lacko S, Thornicroft G, Luciano JV, Rubio-Valera M. Over 40 years (1981-2023) assessing stigma with the Community Attitudes to Mental Illness (CAMI) scale: a systematic review of its psychometric properties. *Syst Rev.* 2023 Apr 14;12(1):66. FI:3.7 (Q4)

SELECTED PROJECTS

- SGR 2022-2024_Psychological Research in Fibromyalgia and Chronic Pain (AGORA). (2021 SGR 00721). Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. 2022-2025. IP:Juan Vicente Luciano Devis.
- Movilidad de personal investigador contratado en el marco de la AES (M-AES). (MV22/00022). Instituto de Salud Carlos III (ISCIII). 2023-2023. IP:Jesús Montero Marín.
- Coste-utilidad y efectos fisiológicos de una aplicación digital para pacientes con fibromialgia basada en la Terapia de Aceptación y Compromiso (Proyecto SMART-FM-Spain). (PI22/00829). Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD; Instituto de Salud Carlos III (ISCIII); Luciano. 2023-2025. IP:Juan Vicente Luciano Devis.
- CIBER Epidemiología y salud pública. (CB22/02/00052). Instituto

de Salud Carlos III (ISCIII). 2023-2024. IP:Juan Vicente Luciano Devis.

SELECTED CLINICAL STUDIES

- Ensayo clínico farmacológico controlado con placebo, aleatorizado, doble ciego y 12 meses de tratamiento para evaluar la efectividad, coste-utilidad y efectos neurobiológicos de la Naltrexona a dosis baja en pacientes con fibromialgia (Proyecto INNOVA). (FSJD-INNOVA-2020). Adknoma Health Research, S.L.; Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD. 2022. IP:Antoni Rozadilla Sacanell.



7.2

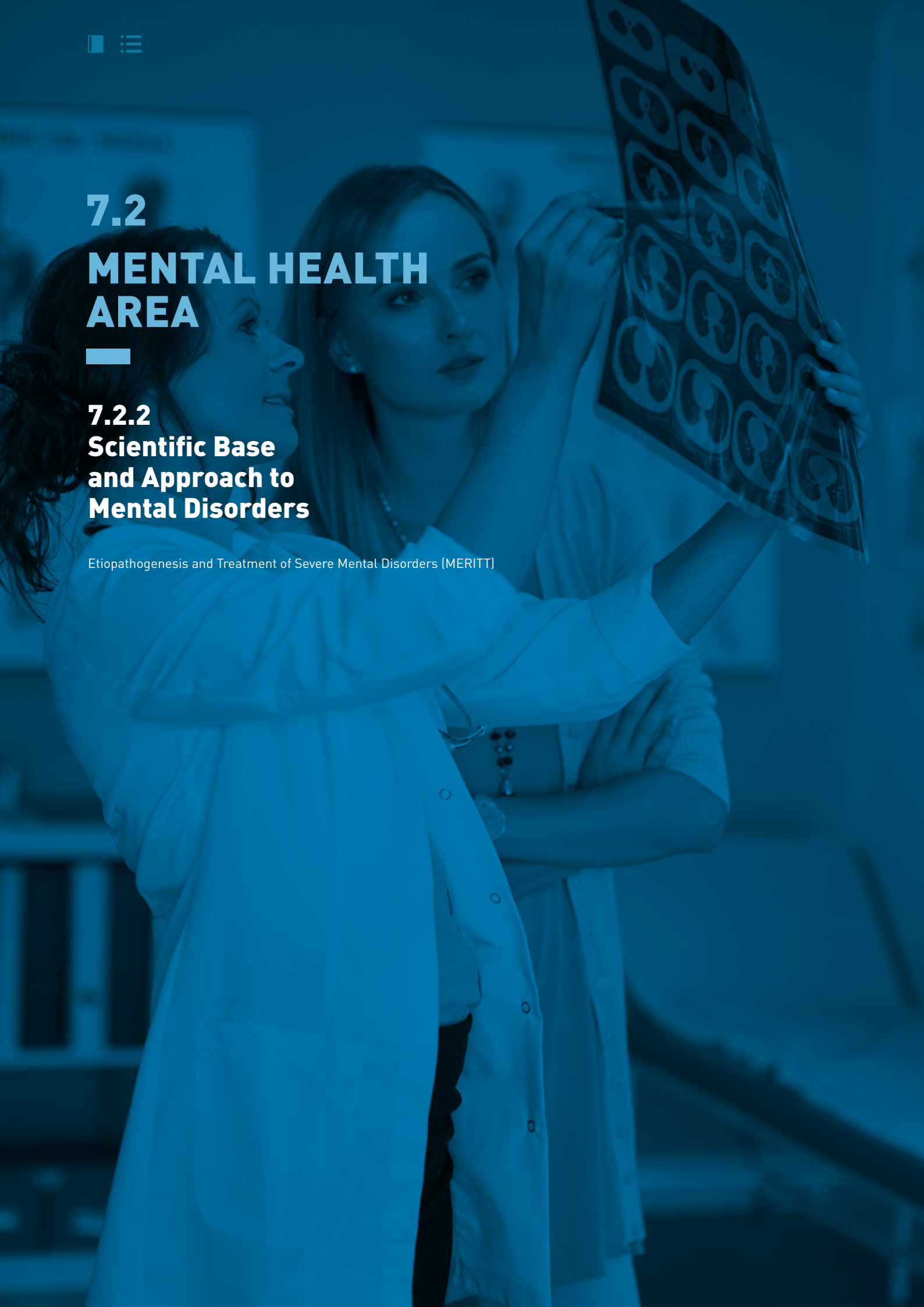
MENTAL HEALTH AREA



7.2.2


Scientific Base and Approach to Mental Disorders


Etiopathogenesis and Treatment of Severe Mental Disorders (MERITT)




Etiopathogenesis and Treatment of Severe Mental Disorders (MERITT)

Susana Ochoa Güerre, MD, PhD
& Judith Usall i Rodié, MD, PhD

 **29**
PROJECTS
27 competitive projects
2 non competitive projects

 **1**
CLINICAL TRIALS &
OBSERVATIONAL STUDIES
0 commercial clinical trials
1 academic clinical trials

 **30**
PUBLICATIONS

 **182**
IMPACT FACTOR
6.1 average impact factor

 **67%**
PUBLICATIONS IN 1ST QUARTILE

 **33%**
PUBLICATIONS IN 1ST DECILE

 **50%**
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Andrea Fontán Abeijón, Noemí del Prado Sánchez, José Manuel Fernández Rodríguez, Jaume Martínez Mas, Alvaro Muro Alvarez, Aitor-Eneko Olivé Albiztur, Jesús Pérez Pazos, María Ribas Siñol, Vicente Tort Herrando, María Luisa Barrigon Estevez, María José Escandell Morcillo, Belen Ramos JoseMaria, Antonio Manuel Alba Muñoz, Hilari Andres Mora, Belen Arranz Martí, Adela Berrozpe Blasco, Emma Casas Anguera, Marta Coromina Sadurní, Jorge Cuevas Esteban, Ruth Cunill Clotet, Gemma Escartin Martin, Gemma Escuder Romeva, Alexandrina Foix Sanjuan, Mar García Franco, Marcia Irazábal Giménez, Francisco Martínez Zambrano, Eva Miquel Fernandez, María Trinidad Peláez Martínez, Elena Rubio Abadal, Luis

San Molina, Sonia Vilamala Anton
POSTDOC ASSOCIATED RESEARCHERS
Victoria Espinosa Lorenzo, America Jennifer Vera Montecinos

PHD STUDENTS

Marina Verdaguer Rodriguez, Desiré Traver Edo, Luciana Beatriz Diaz Cutraro, Silvia Marcó Garcia, Elia Vila Andreu

ASSISTANT RESEARCHER

Francisco Vidal Domenech, Gemma Riquelme Alacid, Alicia Colomer Salvans, Jordi Galiano Landeira, Marta Domínguez del Campo, Clara Serra Arumí, Marta Ferrer Quintero, Anna Butjosa Molines, Gabriel Ledesma Iparraquirre, Regina Vila Badia

COLLABORATOR RESEARCHER

Joan Costa Escolà, Maria Elena Huerta Ramos, Raquel López Carrilero, Alfonso Monje Hernández

The MERITT group is a recognized consolidated group funded by AGAUR. In 2023, the MERITT group has been actively involved in 30 funded research projects, with a total of 7 new projects funded during the year. Among these new projects is a European project on Personalized Medicine (PERMEPSY; ERA-PERMED Call), led by our center, aimed at developing and assessing a Personalized Metacognitive intervention based on Machine Learning. Additionally, our group has been granted the projects "PLATEA: Improving care for adults with autism spectrum disorder" and "Mental health and drug dependence in women in penal execution" by the Generalitat de Catalunya. The MERITT group has also received funding from La Caixa Foundation for the study: "Protective and Risk factors On Suicide in Young people. A population-based study (PROSIA-Y)," which includes a new line of research focusing on the assessment of suicide. Furthermore, the project "Impact of traumatic experiences on the mental health of incarcerated women. Pilot study on the efficacy of EMDR therapy in this population" has been granted funding through the MagicLine call. Additionally, the MERITT group has obtained funds for hiring a predoctoral researcher to join our team.

In terms of scientific publications, our impact factor has increased compared to previous years, reaching a figure of 256,5 for 2023. We published a total of 37 indexed publications,

with 21 in Q1 and 10 in Q2. Many of these publications have been led by members of the group in collaboration with other national and international research centers.

SELECTED PUBLICATIONS

- Cuñat O, Del Hoyo-Buxo B, Vila-Badia R, Serra-Arumí C, Butjosa A, Del Cacho N, Colomer-Salvans A, Dolz M, Cuevas-Esteban J, Iglesias-González M, Usall J, Profep Group. Negative symptoms in drug-naive patients with a first-episode psychosis (FEP). *Asian J Psychiatr*. 2023 Mar;81:103448. FI: 9.5 (D1)
- Díaz-Cutraro L, García-Mieres H, Dimaggio G, Lysaker P, Moritz S, Ochoa S. Metacognition in psychosis: What and how do we assess it? *Span J Psychiatry Ment Health*. 2023 Jul-Sep;16(3):206-207. FI:9.2 (D1)
- Butjosa A, Usall J, Vila-Badia R, Mezquida G, Cuesta MJ, Rodríguez-Toscano E, Amoretti S, Lobo A, González-Pinto A, Espliego A, Corripio I, Vieta E, Baeza I, Bergé D, Bernardo M; PEPs Group. Impact of traumatic life events on clinical variables of individuals with first-episode psychosis and healthy controls. *Int J Soc Psychiatry*. 2023 Feb;69(1):134-145. FI:7.5 (Q1)
- Vera-Montecinos A, Rodríguez-Mias R, Vila È, Villén J, Ramos B. Analysis of networks in the dorsolateral prefrontal cortex in chronic schizophrenia: Relevance of altered immune response. *Front Pharmacol*. 2023 Mar 23;14:1003557. FI:5.6 (Q1)

SELECTED PROJECTS

- Protective and Risk factors On Suicide in Young people. A population-based study (PROSIA-Y). (SR23-00370). Fundació Bancària "La Caixa". 2023-2025. IP:Judith Usall i Rodié.
- Salut Mental i drogodependència de les dones a l'execució penal. Generalitat de Catalunya. 2023-2024. IP:Gemma Escuder Romeva.
- Towards a personalized medicine approach to psychological treatment for psychosis. (AC22/00010). Instituto de Salud Carlos III (ISCIII). 2023-2025. IP:Susana Ochoa Güerre.
- PLATEA. Millorant l'atenció a les persones adultes amb

trastorn de l'espectre autista.
(DDS008/22/000196). Generalitat de
Catalunya. 2022-2025. IP:Nuria Tous
Andreu.

SELECTED CLINICAL STUDIES

- Farmacogenética aplicada para predecir la respuesta al tratamiento del primer episodio psicótico (proyecto FARMAPRED-PEP). (FARMAPRED-PEP). 2023. IP:Judith Usall i Rodié.



7.3

TRANSVERSAL PROGRAM

7.3.1 Health Technologies and Innovation

Medical Technologies: Bioinformatics and Biomedical Signals
P04-02 Medical Technologies: Computer Graphics: Serious Games and
Gamification
Medical technologies: Biomaterials and Tissue Engineering
Medical Technologies: Biosignal Analysis for Rehabilitation and Therapy
Medical Technologies: Instrumentation and eHealth
Medical Technologies: Robotics and Vision
Medical Technologies: Biomechanics
Patients Engagement in Research
Innovation in Health Technologies



Medical Technologies: Bioinformatics and Biomedical Signals

Alexandre Perera Lluna, PhD

0
CLINICAL TRIALS & OBSERVATIONAL STUDIES

6
PUBLICATIONS

38
IMPACT FACTOR
6.3 average impact factor

83%
PUBLICATIONS IN 1ST QUARTILE

33%
PUBLICATIONS IN 1ST DECILE

17%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Jordi Fonollosa, Jon Garrido Aguirre, Jesús Escrivá Muñoz, Giovana Elizabeth Gavidia Bovadilla, Pedro Gomis Román, Alexander Vallmitjana Lees, Montserrat Vallverdú Ferrer

POSTDOC ASSOCIATED RESEARCHERS

Samir Kanaan Izquierdo

PHD STUDENTS

Pol Canal Noguer, Maria Barranco Altirriba, Angela Lopez del Rio, Xavier Marimon Serra, Sergi Picart Armada

ASSISTANT RESEARCHER

Pol Gil Figuerola, Joshua Llano Viles, Pol Sola de los Santos

TECHNICIAN

Joana Palés Huix, Enrico Manzini, Maria Maqueda

SELECTED PUBLICATIONS

- Marín D, Llano-Viles J, Haddi Z, Perera-Lluna A, Fonollosa J. Home monitoring for older singles: a gas sensor array system. *Sensors and Actuators b: Chemical*. 2023 Oct 15; 393:134036. IF: 8.4 [D1].
- Gómez-González S, Llano J, Garcia M, Garrido-Garcia A, Suñol M,

Lemos I, Perez-Jaume S, Salvador N, Gene-Olaciregui N, Galán RA, Santa-María V, Perez-Somarriba M, Castañeda A, Hinojosa J, Winter U, Moreira FB, Lubieniecki F, Vazquez V, Mora J, Cruz O, La Madrid AM, Perera A, Lavarino C. EpiGe: A machine-learning strategy for rapid classification of medulloblastoma using PCR-based methyl-genotyping. *iScience*. 2023 Aug 12;26(9):107598. IF: 5.8 [Q1].

- García-Hernández V, Arambilet D, Guillén Y, Lobo-Jarne T, Maqueda M, Gekas C, González J, Iglesias A, Vega-García N, Sentís I, Trincado JL, Márquez-López I, Heyn H, Camós M, Espinosa L, Bigas A. β -Catenin activity induces an RNA biosynthesis program promoting therapy resistance in T-cell acute lymphoblastic leukemia. *EMBO Mol Med*. 2023 Feb 8;15(2):e16554. IF: 11.1 [D1].
- Fayos De Arizón L, Viera ER, Pilco M, Perera A, De Maeztu G, Nicolau A, Furlano M, Torra R. Artificial intelligence: a new field of knowledge for nephrologists? *Clin Kidney J*. 2023 Jul 29;16(12):2314-2326. IF: 40.6 [Q1].

P04-02 Medical Technologies: Computer Graphics: Serious Games and Gamification

Raul Benítez Iglesias, PhD

0
CLINICAL TRIALS & OBSERVATIONAL STUDIES

6
PUBLICATIONS

33
IMPACT FACTOR
5.5 average impact factor

33%
PUBLICATIONS IN 1ST QUARTILE

17%
PUBLICATIONS IN 1ST DECILE

33%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Dolors Ayala Vallespi, Maria Amor Duch Guillen, Jose Luis Eguía Gómez, Mercè Ginjaume Egado, Robert Joan Arinyó, Núria Pla García, Josep Sempau Roma, Lluís Solano Albages, Daniela Tost Pardell, Marc Vigo Anglada

PHD STUDENTS

Núria Bonet Codina, Ariel von Barnekow

SELECTED PUBLICATIONS

- Solé-Martí X, Labay C, Raymond Y, Franch J, Benítez R, Ginebra MP, Canal C. Ceramic-hydrogel composite as carrier for cold-plasma reactive-species: Safety and osteogenic capacity in vivo. *Plasma Processes and Polymers*. 2023 Gen;20(1):2200155. IF: 3.5 [Q1].
- Latorre-Musoll A, Jornet N, Sempau J. On the beam hardening correction of the Transit-Guided Radiation Therapy attenuation model. *Phys*

Med. 2023 Aug;112:102660. IF: 3.4 (Q2).

- Ashwath VA, Sikha OK, Benítez R. TS-CNN: A three-tier self-interpretable CNN for multi-region medical image classification. IEEE Access. 2023 Jul; 11:78402-78418. IF: 3.9 (Q2).
- Ni M, Li Y, Wei J, Song Z, Wang H, Yao J, Chen YX, Belke D, Estillore JP, Wang R, Vallmitjana A, Benitez R, Hove-Madsen L, Feng W, Chen J, Roston TM, Sanatani S, Lehman A, Chen SRW. Increased Ca2+ Transient Underlies RyR2-Related Left Ventricular Noncompaction. Circ Res. 2023 Jul 7;133(2):177-192. IF: 20.1 (D1).

Medical technologies: Biomaterials and Tissue Engineering

Cristina Canal Barnils, PhD

0
CLINICAL TRIALS & OBSERVATIONAL STUDIES

26
PUBLICATIONS

185
IMPACT FACTOR
7.1 average impact factor

89%
PUBLICATIONS IN 1ST QUARTILE

27%
PUBLICATIONS IN 1ST DECILE

81%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Andrea Malandrino, Albert Espona Noguera, José Manuel García Torres, Montserrat Español Pons, Francesc Xavier Gil Mur, Jordi Guillem Martí, Jose María Manero Planella, Carles Mas Moruno, Marta Pegueroles Neyra, Daniel Rodríguez Rius

POSTDOC ASSOCIATED RESEARCHERS

Irene Lodoso Torrecilla, Francesco Tampieri, Cédric Labay, Juan Tornin Cavielles

PHD STUDENTS

David Piñera Avellaneda, Laura del Mazo Barbara, Victor Chausse Calbet, Claudia García Mintegui, Xavier Solé Martí, Mar Bonany Mariñosa, Judit Buxadera Palomero, Miguel Mateu Sanz, Joanna Magdalena Konka, Lluís Oliver Cervelló, Yago Raymond Llorens, Elisa Rupérez de Gracia

TECHNICIAN

Meritxell Sánchez Hervás, Noelia Aparicio Bádenas, Meritxell Molmeneu Trias, Miquel Punset Fusté

The Biomaterials, Biomechanics, and Tissue Engineering Research Group brings together a multidisciplinary

team of researchers with complementary backgrounds, such as engineers, chemists, physicists, and biologists, ensuring the ability to address scientific challenges in the biomedical field from a global and integrated perspective. In 2022, the group once again received its designation as an SGR research group by the Generalitat de Catalunya.

During 2023, BBT members published a total of 38 articles in JCR-indexed journals and presented their work in more than 42 plenary presentations, invited talks, oral presentations, and posters at international conferences and workshops.

Throughout 2023, BBT executed 7 European projects (including an ERC Advanced Grant and an ERC Proof of Concept), 13 projects funded by the State Research Agency (AEI), 2 by ACCIÓ, 1 by the Barcelona City Council, and 1 by the Jané Mateu Foundation, and is part of two “R&D&I Networks of the Generalitat de Catalunya.” Additionally, the group has been intensely involved in the design, management, organization, and execution of R&D&I projects for national and international companies, and the implementation of specific studies and experimental analysis and/or characterization services, with a biomedical focus.

SELECTED PUBLICATIONS

- Corvi JO, McKittrick A, Fernández JM, Fuenteslópez CV, Gelpí JL, Ginebra MP, Capella-Gutierrez S, Hakimi O. DEBBIE: The Open Access Database of Experimental Scaffolds and Biomaterials Built Using an Automated Text Mining Pipeline. Adv Healthc Mater. 2023 Oct;12(25):e2300150. FI:10 (D1)
- Chausse V, Casanova-Batlle E, Canal C, Ginebra MP, Ciurana J, Pegueroles M. Solvent-cast direct-writing and electrospinning as a dual fabrication strategy for drug-eluting polymeric bioresorbable stents. Addit Manuf. 2023 Jun 5;71:103568. doi: 10.1016/j.addma.2023.103568. IF:11 (D1)
- Tornín J, Mateu-Sanz M, Rey V, Murillo D, Huergo C, Gallego B, Rodríguez A, Rodríguez R, Canal C. Cold plasma and inhibition of STAT3 selectively target tumorigenicity in osteosarcoma. Redox Biol. 2023 Jun;62:102685. FI:11.4 (D1)

- Živanić M, Espona-Noguera A, Lin A, Canal C. Current State of Cold Atmospheric Plasma and Cancer-Immunity Cycle: Therapeutic Relevance and Overcoming Clinical Limitations Using Hydrogels. *Adv Sci (Weinh)*. 2023 Mar;10(8):e2205803. FI:15.1 (D1)

Medical Technologies: Biosignal Analysis for Rehabilitation and Therapy

Miquel A. Mañanas Villanueva, PhD

0
CLINICAL TRIALS & OBSERVATIONAL STUDIES

3
PUBLICATIONS

12
IMPACT FACTOR
4.1 average impact factor

0%
PUBLICATIONS IN 1ST QUARTILE

0%
PUBLICATIONS IN 1ST DECILE

67%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Joan Francesc Alonso Lopez, Sergio Romero Lafuente

SELECTED PROJECTS

- Shirzadi M, Marateb HR, Rojas-Martínez M, Mansourian M, Botter A, Vieira Dos Anjos F, Martins Vieira T, Mañanas MA. A real-time and convex model for the estimation of muscle force from surface electromyographic signals in the upper and lower limbs. *Front Physiol*. 2023 Feb 27;14:1098225. IF: 4 (Q2)
- Sarmiento CA, Serna LY, Hernández AM, Mañanas MÁ. A Novel Strategy to Fit and Validate Physiological Models: A Case Study of a Cardiorespiratory Model for Simulation of Incremental Aerobic Exercise. *Diagnostics (Basel)*. 2023 Feb 27;13(5):908. IF:3.6 (Q2)
- Shirzadi M, Marateb HR, McGill KC, Muceli S, Mananas MA, Farina D. An Accurate and Real-Time Method for Resolving Superimposed Action Potentials in MultiUnit Recordings. *IEEE Trans Biomed Eng*. 2023 Jan;70(1):378-389. IF: 4.6 (Q2)

Medical Technologies: Instrumentation and eHealth

Xavier Rosell Ferrer, PhD

0
CLINICAL TRIALS & OBSERVATIONAL STUDIES

4
PUBLICATIONS

22
IMPACT FACTOR
5.5 average impact factor

50%
PUBLICATIONS IN 1ST QUARTILE

25%
PUBLICATIONS IN 1ST DECILE

50%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Ramon Bragós Badia, Mireya Fernández Chimenó, Miguel Angel García Gonzalez, Miguel J García Hernandez, Lexa Nescolarde Selva, Juan Ramos Castro, Pere Joan Riu Costa

SELECTED PUBLICATIONS

- Nescolarde L, Talluri A, Yanguas J, Lukaski H. Phase angle in localized bioimpedance measurements to assess and monitor muscle injury. *Rev Endocr Metab Disord*. 2023 Jun;24(3):415-428. IF:8.2. (D1).
- Nescolarde L, Orlandi C, Farina GL, Gori N, Lukaski H. Fluid-Dependent Single-Frequency Bioelectrical Impedance Fat Mass Estimates Compared to Digital Imaging and Dual X-ray Absorptiometry. *Nutrients*. 2023 Nov 1;15(21):4638. IF:5.9 (Q1).
- Estrella T, Alfonso C, Ramos-Castro J, Alsina A, Capdevila L. A Serious Game to Self-Regulate Heart Rate Variability as a Technique to Manage Arousal Level Through Cardiorespiratory Biofeedback: Development and Pilot Evaluation

Study. JMIR Serious Games. 2023 Aug 24;11:e46351. IF:4 (Q2).

- Roma-Dollase D, Gualani V, Gohlke M, Abich K, Morales J, Gonzalvez A, Martín V, Ramos-Castro J, Sanjuan J, Nofrarias M. Resistive-Based Micro-Kelvin Temperature Resolution for Ultra-Stable Space Experiments. Sensors (Basel). 2022 Dec 23;23(1):145. IF:3.9 (Q2).

Medical Technologies: Robotics and Vision

Alicia Casals Gelpi, PhD

0
CLINICAL TRIALS & OBSERVATIONAL STUDIES

5
PUBLICATIONS

19
IMPACT FACTOR
3.8 average impact factor

40%
PUBLICATIONS IN 1ST QUARTILE

0%
PUBLICATIONS IN 1ST DECILE

20%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Joan Aranda López, Josep Fernández Ruzafa, Manel Frigola Bourlon, Enric Xavier Martín Rull, Antonio Benito Martínez Velasco

POSTDOC ASSOCIATED RESEARCHERS

Albert Hernansanz Prats

PHD STUDENTS

Narcís Sayols Baixeras

TECHNICIAN

Oriol Escoté Lluís

COLLABORATOR RESEARCHER

Antonio Benedico Blanes, Laureano Tinoco, Carlos Morata

During 2023 we have collected the results of the ongoing projects on surgical and treatment robotics from which we have gained strength in advancing towards more cognitive surgical robotic systems through the progress in autonomous navigation or decision making, both in colonoscopy and laparoscopy; friendly interfaces with visual and haptic feedback as a manual and cognitive aid to the surgeon, sensor substitution (force to heat) towards safer heart ablation treatments, and assistive aids in

teleoperation for fetal surgery. These works have reinforced the cooperation with the international partners and gave place to new partners, in hospital de Sant Pau. No new spin-off has been created but our previous spin-off RobSurgical has finished the clinical trials that should lead to the certification of the Bitrack robot. In the area of assistive and rehabilitation robotics new contacts for getting new projects have been established and new proposals are under review to keep progressing in the field by developing new control strategies in gait rehabilitation assisted with crutches and exoskeletons, especially in gait characterization and to avoid falls in stroke patients. The progress in the intelligent control of a robotized wheelchair has opened challenges both, in the design of an interface adaptable to different disabilities, and in the control of a multirobot system combining mobility (chair) with actuation (robotic arm), dealing with technical issues that range from sensorics (from joysticks to EEG), perception, robot redundancy and ergonomics and safety.

SELECTED PUBLICATIONS

- Pore A, Li Z, Dall'Alba D, Hernansanz A, De Momi E, Menciassi A, Casals Gelpí A, Dankelman J, Fiorini P, Vander Poorten E. Autonomous Navigation for Robot-Assisted Intraluminal and Endovascular Procedures: A Systematic Review. IEEE Transactions on Robotics. 2023; 39(4): 2529-2548. doi: 10.1109/TRO.2023.3269384. IF: 7,8 (Q1)
- Finocchiaro M, Zabban C, Huan Y, Mazzotta AD, Schostek S, Casals Gelpí A, Hernansanz A, Menciassi A, Arezzo A, Ciuti G. Physical Simulator for Colonoscopy: A Modular Design Approach and Validation. IEEE Access. 2023; 11: 36945-36960. doi: 10.1109/ACCESS.2023.3266087. IF: 3,9 (Q2)

Medical Technologies: Biomechanics

Josep Maria Font Llagunes, PhD

0
CLINICAL TRIALS & OBSERVATIONAL STUDIES

7
PUBLICATIONS

25
IMPACT FACTOR
3.6 average impact factor

43%
PUBLICATIONS IN 1ST QUARTILE

29%
PUBLICATIONS IN 1ST DECILE

43%
LEADERSHIP

MEMBERS

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Oriol Alís González, Ana Barjau Condomines, Daniel Clos Costa, Rosa Pàmies Vilà

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Massimo Cenciarini, Albert Peiret Giménez, Joan Lobo Prat

PHD STUDENTS

Jesús de Miguel Fernández, Michael Ray Epke, Antonio Rodríguez Fernández, Míriam Febrer Nafria

In 2023, the BIOMECH group received funding for one new competitive research project financed by the CaixaImpulse Consolidate program (Fundació La Caixa). The project is about a new system for measuring brain signals in newborns. Apart from this project, the group has been working in other competitive projects like NIMBLE, which aims to do basic research related to neuromusculoskeletal modeling and simulation of motor function recovery after robot-assisted therapy in spinal cord injury (AEI Generation of Knowledge call). Another project is ABLE Daily Exo, which is related to the development of smart robotic exoskeletons to assist people with

disabilities in everyday life activities (Public-private partnership call together with the group spin-off ABLE Human Motion). Finally, this year the group also continued the collaboration with Sant Joan de Déu Barcelona Children's Hospital under the project ArmTracker, which is devoted to the development of a wearable upper limb device to monitor arm activity in children with Duchenne muscular dystrophy and spinal muscular atrophy (La Marató de TV3 call). Ten articles were published in JCR indexed journals in this period, 17 papers were presented in scientific conferences, and two doctoral theses were defended. Additionally, the group's PI chaired the IFToMM Technical Committee for Multibody Dynamics and finalized his term in December. The group has been also an active member of the innovation networks Xartec Salut and i4KIDS, led by CREB (Research Centre for Biomedical Engineering) and Sant Joan de Déu Barcelona Children's Hospital, respectively. Lastly, group members have won three scientific awards during 2023.

SELECTED PUBLICATIONS

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- Wakeling JM, Febrer-Nafria M, De Groot F. A review of the efforts to develop muscle and musculoskeletal models for biomechanics in the last 50 years. *J Biomech.* 2023 Jun;155:111657. FI:2.4 (Q3)
- de Miguel-Fernández J, Lobo-Prat J, Prinsen E, Font-Llagunes JM, Marchal-Crespo L. Control strategies used in lower limb exoskeletons for gait rehabilitation after brain injury: a systematic review and analysis of clinical effectiveness. *J Neuroeng Rehabil.* 2023 Feb 19;20(1):23. FI:5.1 (D1)
- de Miguel Fernandez J, Rey-Prieto M, Rio MS, Lopez-Matas H, Guirao-Cano L, Font-Llagunes JM, Lobo-Prat J. Adapted Assistance and Resistance Training With a Knee Exoskeleton After Stroke. *IEEE Trans Neural Syst Rehabil Eng.* 2023;31:3265-3274. FI:4.9 (D1)

Patients Engagement in Research

Joana Claverol Torres, MSc & Begonya Nafria Escalera, MSc

10
PROJECTS
9 competitive projects
1 non competitive projects

0
CLINICAL TRIALS & OBSERVATIONAL STUDIES

4
PUBLICATIONS

15
IMPACT FACTOR
3.8 average impact factor

50%
PUBLICATIONS IN 1ST QUARTILE

0%
PUBLICATIONS IN 1ST DECILE

0%
LEADERSHIP

MEMBERS

STAFF RESEARCHER

Estibaliz Urarte Rodríguez, Júlia Giner Gràcia, Elisa Mora de Checa, Victor Royo Calavera, Claudia Alsina Steinemann

During 2023, our research team was involved in six competitive projects, all of them based on the active participation of patients or caregivers in health research. Our contribution to these projects has allowed us to strengthen the capabilities of our institution at the European level, providing more evidence about the benefits of including patient and caregiver insights as key elements in the design and execution of various types of projects.

One of the most relevant projects was Conect4children, granted by the Innovative Health Initiative (IHI) in 2018, which will ensure its sustainability with the creation of a non-profit spin-off that will deliver

different services to the European research community. Patient and Public Involvement in pediatric clinical trials will be one of the core services of this spin-off, named c4c-S. In delivering these services, IRSJD will play a key role as the central contact point and will provide the infrastructure of the European Young Person's Advisory Groups to deliver the services required by academic institutions and pharmaceutical companies.

Another relevant project was EU-CAYAS.net, the European Childhood Cancer Survivors Platform, which allowed the creation of an infrastructure to connect and provide information to this target population at the European level. Our research team was involved in key outputs such as a position paper about the long-term follow-up of childhood cancer survivors, based on the identification of unmet needs from Spanish patients and in collaboration with other European countries.

Our final goal in contributing to these international projects is to share good practices and deliver the lessons learned to the scientific community. In this sense, we have been involved in eight articles, seven of which were published in 2023. Three out of the seven articles were produced in the context of the Conect4children project.

SELECTED PUBLICATIONS

Reidemeister S, Nafria Escalera B, Marín D, Balayla J, Klingmann I, Klingmann V. Young patients' involvement in a composite endpoint method development on acceptability for paediatric oral dosage forms. *Res Involv Engagem.* 2023 Nov 29;9(1):108.

- Dhaenens BAE, Mahler F, Batchelor H, Dicks P, Gaillard S, Nafria B, Kopp-Schneider A, Ribeiro MA, Schwab M, Sparber-Sauer M, Leubner J, de Wildt SN, Oostenbrink R. Optimizing expert and patient input in pediatric trial design: Lessons learned and recommendations from a collaboration between conect4children and European Patient-Centric Clinical Trial Platforms. *Clin Transl Sci.* 2023 Aug;16(8):1458-1468. FI:3.9 (Q3)
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- Cheng K, Mahler F, Lutsar I, Nafria Escalera B, Breitenstein S, Vassal G, Claverol J, Noel Palacio N, Portman R, Pope G, Bakker M, van der Geest T, Turner MA, de Wildt SN; c4c Expert Group Leads. Clinical, methodology, and patient/carer expert advice in pediatric drug development by conect4children. *Clin Transl Sci.* 2023 Mar;16(3):478-488. FI:3.9 (Q3)

SELECTED PROJECTS

- c4c_conect4children (Collaborative Network for European Clinical Trials For Children). (777389). European Commission. 2018-2025. IP:Joana Claverol Torres.
- EJP RD: European Joint Programme on Rare Diseases. (825575). European Commission. 2019-2024. IP:Joana Claverol Torres.
- Science4Pandemics: Cizitens engagement digital platform for collective intelligence in pandemics. (220365). EIT Health e.V; Fundació Privada per a la Recerca i la Docència Sant Joan de Déu - FSJD. 2022-2024. IP:Begoña Nafria Escalera.
- EU Network of Youth Cancer Survivors. (101056918). European Commission. 2022-2024. IP:Begoña Nafria Escalera.

Innovation in Health Technologies

Arnau Valls Esteve

20
PROJECTS
15 competitive projects
5 non competitive projects

0
CLINICAL TRIALS &
OBSERVATIONAL STUDIES

5
PUBLICATIONS

17
IMPACT FACTOR
3.4 average impact factor

40%
PUBLICATIONS IN 1ST QUARTILE

0%
PUBLICATIONS IN 1ST DECILE

80%
LEADERSHIP

MEMBERS

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ASSISTANT RESEARCHER

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SELECTED PUBLICATIONS

- Valls-Esteve A, Lustig-Gainza P, Adell-Gomez N, Tejo-Otero A, Engli-Rueda M, Julian-Alvarez E, Navarro-Sureda O, Fenollosa-Artés F, Rubio-Palau J, Krauel L, Munuera J. A state-of-the-art guide about the effects of sterilization processes on 3D-printed materials for surgical planning and medical applications: A comparative study. *Int J Bioprint.* 2023 May 17;9(5):756. FI: 8.4 (Q1)
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J, Barber-Martinez de la Torre I, Munuera J, Fondevila C, Krauel L. Patient-Specific 3D Printed Soft Models for Liver Surgical Planning and Hands-On Training. Gels. 2023 Apr 16;9(4):339. FI: 4.6 (Q1)

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SELECTED PROJECTS

- i4KIDS-EUROPE_Inclusive and Interconnected Ecosystem to Boost Paediatric Innovation in Europe-i4KIDS-EUROPE. (101096505). European Commission. 2023-2025. IP:Arnau Valls Esteve
- Hub d'Innovació Pediàtrica - i4KIDS. (2021 XARDI 00006). Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. 2023-2025. IP:Jaume Pérez Payarols,Arnau Valls Esteve
- PHEMS: Pediatric Hospitals as European drivers for multi-party computation and synthetic data generation capabilities across clinical specialties and data types. (101094195). European Commission. 2023-2026. IP:Arnau Valls Esteve
- Desarrollo de modelos biomecánicos avanzados para planificación y mejora de la practica quirúrgica para cirugía maxilofacial labiopalatina (BIOMOD). Fundació Institut de Bioingenieria de catalunya (IBEC). 2023-2024. IP:Josep Rubio Palau.



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