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ANNUAL SCIENTIFIC
REPORT **2015**



Sant Joan de Déu
Institut de Recerca

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Foreword

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Introduction

The year 2015 was a time of change for the biomedical and social research of the Sant Joan de Déu Foundation and its largest hospitals, the Children's Hospital of Esplugues and the Health Park of Sant Boi de Llobregat. On February was signed the agreement for the establishment of the Sant Joan de Déu Research Institute (IRSJD) between Sant Joan de Déu Children's Hospital, the University of Barcelona, with the participation of the Institute of Biomedicine (IBUB) and the Institute of Neurosciences (UBneuro), and the Polytechnic University of Catalonia with the Biomedical Engineering Research Centre (CREB). Later has joined the Sant Joan de Déu Health Park with its area of Mental Health. The constitution and conformation of the IRSJD represents two major opportunities for our research activities and knowledge generation in human health and disease. First, it gives form and structure to the investigation of the clinical and basic groups that are doing research in the frame managed by the Foundation and the two Sant Joan de Déu campuses. Second, the Institute is an opportunity for interaction, collaboration and openness with our scientific and technological environment of two Catalan universities.

The IRSJD mission is to advance research in the understanding health and disease of humans, transferring this knowledge to clinical medicine to improve the quality of life of sick people. Before this mission, we focus our efforts and goals into the first decades of life and its impact on the full life cycle of the person. We do science in the field of pediatric medicine and human development, which is concretized and structured in 6 medical areas —pediatric neurosciences, developmental cancer, pediatric diseases start in early stages of life, infectious and immune diseases fetal/pediatric and environment, mental health— and transversal four areas involving fundamental biomedicine, neuropsychology and cognitive neuroscience, biomedical engineering, and epidemiology and health services.

The most immediate consequence of the creation of the Sant Joan de Déu Research Institute has been improved knowledge of the projects carried out in each of research centers and promoting new joint projects. This should represent an increase of scientific and translational biomedical research and our ability to impact clinical practice to improve diagnosis and treatment of our patients.

Over the course of 2015, 251 research projects have been carried out. Of this total, 168 were competitive projects, 139 of them being funded by Spanish entities and 29 by international entities. Likewise, 89 clinical trials and 43 observational studies were conducted. In 2015, our researchers have published 302 articles with a total impact factor of 1,138 points.

This has been what has been done, but science is curiosity, questions and search for answers, and as scientists in the field of health must continue this effort to provide solutions and hope to children and people with illnesses affecting their life cycle and their biographical experience.

Francesc Palau

Director Sant Joan de Déu Research Institute

Governing and advisory bodies

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Dr. Rafael Artuch Iriberrí
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Dr. Carles Escena Micó (IN-UB)
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Ms. Júlia Ribot Ballabriga

Director of Institut de Recerca Sant Joan de Déu
Director of Research Foundation Sant Joan de Déu
Scientific deputy director of Institut de Recerca Sant Joan de Déu
University of Barcelona
University of Barcelona
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Professor of Psychiatry, University of Cantabria (UC)
General Director of BIOCAT

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Vertical areas

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AREA

Paedriatic neuroscience

Groups
Projects
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PKU and other intermediary metabolism defects

The fact that our hospital is a reference centre for phenylketonuria in Catalonia has allowed us to carry out an in-depth study of the pathophysiological mechanisms involved in this disorder and to try out new treatments aimed to improve its metabolic control and neurological prognosis. On the basis of these studies, we have reached the conclusion that some neuropsychological and neurological changes are associated, on the one hand, with metabolic control, and on the other, with antioxidant deficiencies. We have begun new treatments (with tetrahydrobiopterin) on our

phenylketonuria patients, which have provided a better metabolic control and the possibility of freeing our patients from the special diet they were following. The follow-up of this treatment will be an important subject of study in the next three years. In connection with other intermediary metabolism defects, our work on metabolism disorders of homocysteine and other amino acids, which is primarily focused on characterising new disorders and new phenotypic presentations, may be noted.

Research team

Coordinator Jaume Campistol
Researchers Rafael Artuch, Rosa Gassió, Maria Julieta González, Sílvia Meavilla,
Carmen Fons, Camila Garcia Volpe, Rose Colome,
Aida Ormazabal, Cristina Sierra

Neurometabolic disorders with expression in the CNS

We have been studying neurotransmitter related disorders deficiencies since 2003 while serving various hospital centres in Spain, Portugal, Greece, Argentina, Turkey and India, among other countries. Basically, we study dopamine and serotonin defects and quite recently we have also turned our attention to GABA. From the clinical standpoint, our patients present severe encephalopathies which are detected by specifically studying these neurotransmitters in cerebrospinal fluid (CSF). Our main focus is diagnosis within a very clear therapeutic approach since many of these disorders have a treatment that can improve or normalise the patient's clinical condition. We have received several FIS grants to study this area, being the most recent addressed to the description of brain connectivity and brain mapping of these disorders. We are co-

founders of the I-NTD project, (<http://www.intd-online.org/>), the international working group for the study of neurotransmitter defects, which has started a world patients' registry and different research projects.

Moreover, some aspects of translational research are now also an important part of our research with the inclusion of studies of CSF proteomics and of cell lines of tyrosine hydroxylase (a limiting enzyme in dopamine synthesis) deficiency. We have been able to add these new procedures to our research thanks to the establishment in the last recent years of the laboratory of synaptic metabolism and the project "connecting the growing brain" who aims to study patterns of synaptic communication in neuropaediatric disorders (<http://www.connectingthegrowingbrain.com/>).

Research team

Coordinator Àngels García-Cazorla
Researchers Alba Tristán, Anna López, Inés Medina, Elisenda Cortès-Saladelafont, Mercedes Casado, Alfonso Oyarzábal

Mitochondrial energy metabolism

In this research group we have analysed the clinical and biochemical aspects of mitochondrial encephalomyopathies in childhood caused by changes in oxidative phosphorylation, studying as well the effects of treatment with various therapies. This line of research has been possible thanks to stable collaborations with reference centres within the framework of

the Centre for Biomedical Network Research on Rare Diseases (CIBERER) network, and thanks to the performance of research projects financed by FIS. We have carried out a research line for the diagnosis of primary and secondary deficiencies of coenzyme Q. This group has been possible thanks to the three FIS projects that have been consecutively obtained for research on this subject.

Research team

Coordinator Rafael Artuch
Researchers Carmen Fons, Raquel Montero, Mar O'Callaghan, Federico Ramos, Delia Yubero

Applied research in neuromuscular disorders

Our research group are currently focused on two main areas: (1) Muscular dystrophies, including Duchenne muscular dystrophy (DMD) and congenital muscular dystrophies (CMD), with a special focus on the role of the extracellular matrix and collagen VI. (2) Mitochondrial diseases including mitochondrial DNA depletion syndromes affecting muscle. In both cases, by means of "omics" techniques, bioinformatics and cell models, the gene expression footprint of patients' muscle

has been determined in comparison to healthy muscle. This has allowed the identification of new pathophysiological mechanisms, diagnostic markers and therapeutic targets for these disorders. In collaboration with the University of Barcelona and CIBERDEM, we have described a new function for collagen VI in the regulation of glucose metabolism in muscle cells and adipocytes. This finding forms the basis of a pending patent.

Research team

Coordinator Cecilia Jiménez-Mallebrera
Researchers Andrés Nascimento, Cristina Jou, Carlos Ortez, Mónica Llano-Diez, Ma Angels Rodríguez García, Sergi García, Lidia Alvarez

Movement disorders of genetic and neurometabolic origin

The Research Group on Paediatric Neurosciences forms part of the Integral Unit for the Study of Movement Disorders Caused by Rare Diseases. This unit, which is directed jointly by Hospital Sant Joan de Déu and Hospital Clínic de Barcelona, has been designated as a Reference Integral Unit by the Spanish National Health System. This group is focused on research on rare diseases of genetic and neurometabolic origin which occur with movement disorders in childhood. Such movement disorders can occur in isolated form or in association with other neurodevelopmental problems, and they are a major cause of physical disability in children. The research on this group of disorders is addressed to clinical characterisation, identification of new biomarkers, and molecular studies to identify new genetic defects.

Two consecutive FIS projects have been carried out in the last 5 years with their focus on the study of defects of the genes responsible for the transport, synthesis and processing of vitamins. Thiamine and cerebral folate deficiencies affect the developing central nervous system and may produce clinical conditions of acute and/or progressive encephalopathy, mitochondrial energy deficiency and cerebral myelination disorders. Their identification is a priority since the early administration of high doses of these vitamins may bring about an immediate recovery from symptoms and revert brain lesions. Thanks to these projects, we have established blood and cerebrospinal fluid biomarkers for their identification. Indeed, thanks to these findings, our hospital has become a reference centre in this field and we have taken part in the identification of new phenotypes associated with genetic defects.

Research team

Coordinator Belén Perez-Dueñas
Researchers Jordi Muchart, Darió Ortigoza, Pilar Poo, Merche Serrano

Neurogenetics and molecular medicine

The group of "Neurogenetics and Molecular Medicine" (NeuroGene) investigates the molecular basis of neurodevelopmental diseases and how the central and peripheral nervous systems work. The experimental approach includes genetic and genomic analyses, the cell biology and the study of the pathophysiology using animal models. NeuroGene is a new research group created in 2015 by the confluence of Dr. F. Palau, CSIC research professor coming from the Institute of Biomedicine of Valencia, Dr. J. Hoenicka, NHS investigator coming from the 12 de Octubre Hospital Research Institute in Madrid, and Dr. J. Armstrong and Dr. M. Serrano, originally located at the Sant Joan de Déu Childrens Hospital. NeuroGene's scientific work has the vocation of clinical translation within the Pediatric Institute for Rare Diseases (IPER) and transfers the knowledge generated to the National Health System. Researchers have tight relationship and projects with the Genetic and Molecular Medicine Department of Sant Joan de Déu Childrens Hospital.

The group's strategy is based on three fundamental aspects of the research activity: (1) Making science focused on the area of neurological and mental illnesses in paediatrics and adult medicine. (2) Develop scientific lines that combine established knowledge with novel and risky hypothesis under frontier science. (3) Develop a translational research model and biomedical innovation in the field of rare diseases based on the research about proposals or clinical hypotheses, and implement diagnostic and therapeutic services as instruments of the clinical translational and the interactions with biomedical industry. Research lines: Genetics and pathophysiology of neuropsychiatric disorders (Parkinson's disease, addictive disorders) [Dr. J. Hoenicka]; Genetics and pathophysiology of neuromuscular diseases (Charcot-Marie-Tooth neuropathy disease, Friedreich's ataxia) [Dr. F. Palau]; Genetics and pathophysiology of Rett Syndrome [Dr. J. Armstrong]; and Natural history and genomics of neurodevelopmental diseases (congenital disorders of glycosylation, Menkes disease, intellectual disability) [Dr. M. Serrano, Dr. F. Palau and Dr. J. Hoenicka].

Research team

Coordinator	Dr. Janet Hoenicka
Principal Investigators	Francesc Palau, Janet Hoenicka, Judith Armstrong, Mercedes Serrano
Researchers	Laura Domínguez, Lara Cantarero, Xotchilt Castro, Loreto Martorell, María del Mar Pérez Iribarne, Isabel Plensa, Antonio Martínez Monseny, Nuria Brandi, Laura Blasco, Azahara Civera, Sílvia Vidal
Technicians	Dolores López, Ana López, Lidia Álvarez, Paola Pacheco, Jordi Genovès, Montserrat Naudó, Daniel Castillo, Patricia Peral, Teresa Zabala, Lluïsa Ramírez

Projects

- "Connecting the growing brain": synaptic dysfunctions underlying neuropaediatric diseases. Fundació Privada BioRegió de Catalunya. November 2015. PI: Àngels García Cazorla.
- Difference in the cognitive and behavioural profile in children with FASD (Foetal Alcohol Spectrum Disorder) and ADHD in two age groups: phenotypic and neuroimaging correlation. Fundació Alicia Koplowitz. November 2015 - November 2017. PI: Anna Maria Sans Fitó.
- Observational study to assess the response of oral salbutamol in patients affected by type 2 spinal muscular atrophy. Clientes Diversos. May 2015 - May 2017. PI: Andrés Nascimento Osorio.
- CIBER - PIER: Instituto Pediátrico de Enfermedades Raras. Centro de Investigación Biomédica en Red (CIBER). May 2015 - December 2016. PI: Francesc Palau.
- Short stay José Castillejo. Ministerio de educación. April 2015 - July 2015. PI: Belén Pérez Dueñas.
- Myopathies associated with collagen VI deficiency: natural history, functional assessment and identification of new diagnostic and prognostic markers. Fundación Andrés Marcio, niños contra la Laminopatía. March 2015 - March 2018. PI: Cecilia Jiménez Mallebrera.
- Rio Hortega. Beneficiari: Elisenda Cortés. Instituto de Salud Carlos III (ISCIII). March 2015 - March 2017. PI: Àngels García Cazorla.
- Neurodegeneration with Brain Iron Accumulation: Clinical Assessment and Genetic Characterization by means of a Spanish Multi-Centre Research Network. Fundació La Marató de TV3. March 2015 - March 2018. PI: Belén Pérez Dueñas.
- Pantothenate Kinase Associated Neurodegeneration: Clinical Assessment and Genetic Characterization by means of a Multi-Centre Research Network. Retrophin, Inc. March 2015 - January 2017. PI: Belén Pérez Dueñas.
- Mitochondrial encephalomyopathies and secondary deficiencies of coenzyme Q10: Studies on paediatric patients and on cellular and murine models. Instituto de Salud Carlos III (ISCIII). January 2015 - December 2017. PI: Rafael Artuch Iriberrí.
- Severe neurological disorders in childhood. Banco Bilbao Vizcaya Argentaria, S.A. January 2015 - December 2015. PI: Rafael Artuch Iriberrí.
- Genetic studies on metabolic minority diseases. OSSJD - Obra Social Sant Joan de Déu. January 2015 - December 2016. PI: Rafael Artuch Iriberrí.
- Study of trace elements (Cu, Zn, Se and Mn) in cerebrospinal fluid of paediatric subjects and analysis of related biomarkers. Instituto de Salud Carlos III (ISCIII). January 2015 - December 2017. PI: Aida Ormazabal Herrero.
- Pre-doctoral health research training contract. Beneficiario: Alba Tristan. Instituto de Salud Carlos III (ISCIII). January 2015 - December 2018. PI: Àngels García Cazorla.
- Radiological and clinical quantitative assessment in PMM2-CDG patients through a national network of practitioners. Instituto de Salud Carlos III (ISCIII). January 2015 - December 2017. PI: Mercedes Serrano Gimarré.
- Duplication of MECP2: genetic and clinical study on Spanish patients. Asociación Miradas que hablan Duplicación Mecp2. January 2015 - January 2016. PI: Judith Silvia Armstrong Morón.
- Severe childhood neurological disorders. Fundación Banco Bilbao Vizcaya Argentaria. November 2014 - November 2015. PI: Rafael Artuch.
- Identification of populations showing high linear correlations between biochemical variables. SAF2013-50139-EXP. Ministerio de Ciencia e Innovación (MINECO). September 2014 - August 2015. PI: Rafael Artuch.
- Cellular model for the study and treatment of excitatory/inhibitory synaptic unbalance in Rett syndrome. Obra Social Sant Joan de Déu. September 2014 - August 2017. PI: Àngels García-Cazorla.
- Study of defects of thiamine transport and metabolism associated with recurrent childhood encephalopathies. FI grant: Beneficiari: Juan Dario Ortigoza. Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. May 2014 - April 2017. PI: Belén Pérez-Dueñas.
- Study of defects of thiamine transport and metabolism associated with recurrent childhood encephalopathies. Hospital Sant Joan de Déu Barcelona. March 2014 - March 2017. PI: Belén Pérez-Dueñas.
- Exome sequencing for discovery of candidate genes in Rett and Rett variants. Associació Catalana de la Síndrome de Rett - ACSR. January 2014 - . PI: Judith Armstrong Morón.

- Research Group on Paediatric Neurosciences and Metabolism. SGR 2014. 2014SGR1150. Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. January 2014 - December 2016. PI: Jaume Campistol.
- Role of collagen VI and biglycan in muscle homeostasis: regulation of muscle fibre size and metabolism. Implications for muscular dystrophies. PI13/00837. Instituto de Salud Carlos III. January 2014 - December 2016. PI: Cecilia Jiménez-Mallebrera.
- Research activity intensification programme in the Spanish National Health System 2013. INT13/00212. Instituto de Salud Carlos III. January 2014 - January 2015. PI: Rafael Artuch.
- Reina Sofia Foundation for Rett Syndrome. Fundación Reina Sofia. May 2013 -. PI: Judith Armstrong.
- Role of collagen VI in muscle homeostasis: regulation of muscle fibre size and of glucose and fatty acid metabolism. Hospital Sant Joan de Déu Barcelona. April 2013 - April 2016. PI: Cecilia Jiménez-Mallebrera.
- Inherited NeuRoMetabolic Diseases INFORMATION NETWORK. InNerMeD-I-Network. European Commission. April 2013 - September 2015. PI: Àngels García-Cazorla.
- Biomarkers of cerebral neurotransmission alteration in paediatric encephalopathies of genetic origin: studies on patients and on a cellular mode. Instituto de Salud Carlos III (ISCIII). January 2013 - December 2015. PI: Àngels García-Cazorla.
- Dissection of the mitochondrial pathophysiology of Charcot-Marie-Tooth disease. Ministerio de Economía y Competitividad (MINECO). January 2013 - December 2016. PI: Francesc Palau.
- Study of defects in the transport and metabolism of thiamine associated with recurrent encephalopathies in childhood. Instituto de Salud Carlos III (ISCIII). January 2013 - December 2015. PI: Belén Pérez-Dueñas.
- Study of biochemical markers of dopaminergic and serotonergic neurotransmission in relation to neurological and neuropsychological changes in phenylketonuria. Instituto de Salud Carlos III (ISCIII). January 2013 - December 2015. PI: Jaume Campistol.
- Study of RNA in CSF in Rett patients; new diagnostic approaches. Fondo Biorett. January 2013 - December 2015. PI: Judith Armstrong.
- Predoctoral PFIS grant - Phenotypic and molecular characterisation of coenzyme Q deficiency syndrome. Instituto de Salud Carlos III. September 2012 - September 2016. PI: Rafael Artuch.
- Neuropsychological rehabilitation in children with moderate or severe TBI. Randomised study; conference addressed to parents, to children via robotics and control group. Fundació La Marató de TV3. February 2012 - February 2015. PI: M^a Pilar Poo.
- Phenotypic and molecular characterisation of coenzyme Q deficiency syndrome. Instituto de Salud Carlos III (ISCIII). January 2012 - March 2015. PI: Rafael Artuch.
- Application of Global Gene Expression Analysis and Functional Genomics to the Study of the Physiopathology, Treatment and Diagnosis of Pediatric Myopathies. Instituto de Salud Carlos III (ISCIII). April 2010 - April 2016. PI: Cecilia Jiménez-Mallebrera.
- Glutaric aciduria type 1. Fundació El Somni dels Nens. May 2009 -. PI: Jaume Campistol.
- Clinical study of the natural history of Sanfilippo syndrome (MPS-III). Associació de les Mucopolisacàridosis i les síndromes Relacionades. March 2009 -. PI: Mercè Pineda.

Clinical trials

- Surveillance protocol of Cystadane® (Orphan Europe), in collaboration with the European Network, and register for homocystinurias and methylation deficiency (E-HOD). May 2015 -. PI: Silvia Maria Meavilla.
- Simulated procedure-controlled double-blind randomised phase 3 study to assess the clinical effectiveness and safety of ISIS 396443 administered via intrathecal route in paediatric patients with late-onset spinal muscular atrophy. March 2015 -. PI: Andrés Nascimento.
- Phase III extension trial of ataluren (PTC124) in terminator mutation dystrophinopathy patients. PTC Therapeutics, Inc September 2014 -. PI: Andrés Nascimento.
- Observational, Ambispective Natural History Study of Patients with Mucopolysaccharidosis Type IIIA (MPS IIIA) or San-filippo Syndrome Type A. July 2014 -. PI: Mercè Pineda.
- Phase III, multi-centre, double-blind, prospective, randomised, controlled, multiple-treatment study to evaluate the efficacy and safety of Dysport in the paediatric treatment of upper limb spasticity. May 2014 -. PI: Xenia Alonso.
- Prospective, cross-sectional, longitudinal study with additional retrospective review of clinical records to evaluate the clinical and biochemical characteristics and evolution of the disorder in mucopolysaccharidosis type IIIB patients. INC Research UK Limited. March 2014 -. PI: M^a del Mar OCallaghan.
- Phase IV, open, single-cohort study of long-term neurocognitive (NC) results in children aged 4-5 years with phenylketonuria treated with sapropterin dihydrochloride (Kuvan®) for 7 years. Merck KGaA. February 2014 -. PI: Jaume Campistol.
- Retrospective review of the clinical records of deceased patients with mucopolysaccharidosis type IIIB. January 2014 -. PI: M^a del Mar OCallaghan.
- Epidemiological study to evaluate the severity of RSV infection in patients with severe neurological disorders. January 2014 -. PI: Andrés Nascimento.
- Study of the natural history of molybdenum cofactor deficiency and isolated sulphite oxidase deficiency. October 2013 -. PI: María Julieta.
- Phase 3 study on the efficacy and safety of ataluren (PTC124) in patients with terminator mutation dystrophinopathy. PTC Therapeutics. July 2013 -. PI: Andrés Nascimento.
- International disease register with data on manifestations, interventions and results in patients with tuberous sclerosis complex (TOSC). Novartis Farmacèutica. November 2012 -. PI: Jaume Campistol.
- Multi-centre, open, long-term follow-up study to evaluate the safety and efficacy of brivaracetam as adjuvant treatment in paediatric patients with epilepsy. October 2012 -. PI: Francesc Xavier Sanmartí/ Alia Ramírez.
- Open study on terminator mutation dystrophinopathy patients previously treated with ataluren (PTC124®). Hospital Sant Joan de Déu Barcelona; PTC Therapeutics. October 2012 -. PI: Jaume Colomer/Andres Nascimento.
- Register of patients with Niemann-Pick disease, type C. Actelion Pharmaceuticals España. February 2011 -. PI: Mercè Pineda.
- European paediatric-maternal register of adults on Kuvan® (KAMPER). Merck. March 2010 -. PI: Jaume Campistol.

Publications

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- Campistol J. Neurological manifestations in metabolic diseases. *Arq Neuro-Psiquiat* 2015. 73: 59-63. FI: 0,843(Q4).
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Vertical areas

2

AREA

Paediatric
neuroscience

Groups

Projects

Clinical Trials

Publications

Supervised theses

Networks

Supervised theses

- Synaptic Dysfunction In Early Encephalopathies: From Genes to Function. Doctoral Candidate: Sofia Duarte. Director: Àngels Garcia-Cazorla. November 2015.

Networks

- Centre for Biomedical Network Research on Rare Diseases (CIBERER). Ministry of Economy and Competitiveness. Carlos III Health Institute.
- Spanish Charcot-Marie-Tooth Consortium (TREAT-CMT). Carlos III Health Institute (ISCIII) and International Rare Diseases Research Consortium (IRDiRC).
- European Treat-NMD Network.

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2015

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Sant Joan de Déu
Institut de Recerca



Vertical areas

3

AREA

Molecular and cellular biology
of developmental tumours

Groups
Projects
Clinical Trials
Publications
Networks

Vertical areas

3

AREA

Molecular and cellular biology of developmental tumours

Groups

Projects

Clinical Trials

Publications

Networks

Preclinical pharmacology and drug release in solid paediatrics tumours

Development of preclinical disease models (neuroblastoma, diffuse intrinsic pontine glioma/DIPG, rhabdomyosarcoma, Ewing's sarcoma, retinoblastoma) on the basis of biobank samples. Study of new treatments (pharmacological, biological and immunotherapy)

in preclinical models of paediatric solid tumours. Study of the penetration of chemotherapeutic agents in preclinical models of paediatric solid tumours. Development of new chemotherapy release systems in paediatric solid tumours.

Research team

Coordinator Ángel Montero-Carcaboso
Researchers Sonia Paco, Carles Monterrubió, Nagore Gené Olaciregui, Guillem Pascual Pasto, Guillermo Chantada, Ofelia Cruz, Andrés Morales, Andreu Parareda
Technician Mònica Vilà Ubach

Paediatric Leukaemia and Blood disorders

The primary goal is to deepen our understanding of the origin and molecular bases of paediatric haematological malignancies with a view to transferring new diagnostic, prognostic and therapeutic tools to clinical practice. We seek to conduct translational and

easily applicable research both on leukaemias and bone marrow failure syndromes, and on non-neoplastic blood changes, haemoglobin disorders and coagulation disorders.

Research team

Coordinator Mireia Camós
Researchers Roberta Malatesta, Nerea Vega García, Anna Alonso Saladrigues, Montse Mesegué Medà, María Trabazo del Castillo, Montse Torredadell Burriel, Anna Ruiz-Llobet, Rubén Berrueco Moreno, Albert Català Temprano, Julia Marsal Ricomà, Isabel Badell Serra, Susana Rives Solà
Technician Camino Estella Aguado

Vertical areas

3

AREA

Molecular and cellular biology of developmental tumours

Groups

Projects

Clinical Trials

Publications

Networks

Neuroblastoma and Personalized Medicine

Our group has two primary goals: (1) to identify new therapeutic targets in neuroblastoma, and (2) to provide a personalised treatment to paediatric patients affected by relapsed/refractory solid tumours, on the basis of the knowledge of their genetic changes. Important aspects of our activity are the assessment of

the calcium-sensor receptor and parathyroid hormone-related protein (PTH-RP) as new therapeutic targets in neuroblastoma, and the analysis of combined therapies based on the knowledge of the interaction of these molecules with the various altered signalling pathways in neuroblastoma.

Research team

Coordinator	Carmen de Torres
Researchers	Silvia Mateo Lozano, Carlos J. Rodríguez Hernández, Marta García López
Technician	Noelia Salvador

Translational genomics

Integration of the knowledge of the genome and of medicine to improve therapeutic strategies. The aim is to use "omics" technologies to identify the underlying molecular mechanisms of

the biology and clinical-pathological behaviour of developmental tumours, as a possibility for the development of diagnostic, prognostic and treatment-response biomarkers.

Research team

Coordinator	Cinzia Lavarino
Researchers	Soledad Gómez, Laura García, Alicia Garrido, Hector Salvador
Technician	Isadora Lemos

Vertical areas

3

AREA

Molecular and cellular biology of developmental tumours

Groups

Projects
Clinical Trials
Publications
Networks

Sarcoma and histiocytosis

The main areas of research include: 1) the study of the origin of sarcoma and phenotypic and genotypic characterization of the various clinical subtypes; 2) the existence of precursor or tumour stem cells; 3) improved treatment of patients with sarcomas

through the study of pre-clinical pharmacology; 4) Development of therapies targeted at the IgF1r pathway; 5) the study of directed therapies and molecular pathways in histiocytosis.

Research team

Coordinator	Jaume Mora
Researchers	M ^a Inmaculada Hernández Muñoz, Sara Sanchez Molina, Elisabet Figuerola Bou, Sara Perez Jaume, Estela Prada Varela, Veronica Celis, Moira Garraus
Technician	M ^a Jesus Nagel Moyano

Projects

- Cure4RB. Ministerio de Economía Y Competitividad (MINECO). July 2015 - December 2018. PI: Angel Montero Carcaboso.
- CEBIOTEX. Cebiotex S.L. October 2015 - April 2016. PI: Angel Montero Carcaboso.
- Adverse genetic changes in acute paediatric leukaemia. Fundación Pelayo. October 2015 - September 2016. PI: Mireia Camós Guijosa.
- Pilot study of the infusion of gene-modified autologous T lymphocytes expressing anti-CD19 in patients with treatment-resistant or refractory CD19+ lymphoma or leukaemia. Instituto de Salud Carlos III (ISCIII). January 2015 - December 2017. PI: Susana Rives Solà.
- Assessment of calcium-sensor receptor and parathyroid hormone-related protein (PTHrP) as new therapeutic targets in neuroblastoma. Instituto de Salud Carlos III (ISCIII). January 2015 - December 2017. PI: Carmen De Torres Gómez-Pallete.
- Epigenetics in the pathogenesis of neuroblastoma: new molecular perspectives for development of therapeutic strategies. Instituto de Salud Carlos III (ISCIII). January 2015 - December 2017. PI: Cinzia Lavarino.
- Translational research program in diffuse intrinsic pontine gliomas. Hospital Sant Joan de Déu - Esplugues HSJD. April 2014 - April 2017. PI: Angel Montero-Carcaboso.
- Genetic and epigenetic study of the regulatory molecular pathways of haematopoietic progenitor cells in paediatric leukaemias with rearrangement of the MLL gene. Fundación CRIS de Investigación para Vencer el Cáncer. March 2014 - March 2017. PI: Mireia Camos.
- 2014SGR1398 Developmental tumours research group. Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. January 2014 - December 2016. PI: Jaume Mora.
- Translational research programme on diffuse intrinsic pontine. Contrato Miguel Servet I. Instituto de Salud Carlos III. January 2014 - January 2019. PI: Angel Montero-Carcaboso.
- Translational research program in diffuse intrinsic pontine. Proyecto asociado a contrato Miguel Servet I. Instituto de Salud Carlos III. January 2014 - December 2016. PI: Angel Montero-Carcaboso.
- Search, validation and clinical translation of new therapeutic targets based on the study of genomics and integrative proteomics in Ewing's sarcoma. Asociación Española Contra el Cáncer. October 2013 - September 2018. PI: Jaume Mora.
- Study of molecular pathways regulating haematopoietic progenitor cells in paediatric leukaemias with rearrangement of gene MLL. Hospital Sant Joan de Déu - Esplugues HSJD. March 2013 - February 2016. PI: Mireia Camos.
- Study of molecular pathways regulating haematopoietic progenitor cells in paediatric leukaemias with rearrangement of gene MLL. Instituto de Salud Carlos III. January 2013 - December 2015. PI: Mireia Camos.
- Molecular mechanisms involved in the development of paediatric leukaemias. Fundación Científica de la Asociación Española Contra el Cáncer (AECC); Fundación Instituto Mar de Investigaciones Médicas. September 2012 - August 2015. PI: Mireia Camos.
- CHD5, PAFAH1B1 and NME1: Markers for prognosis and prediction of therapeutic response in neuroblastoma. Chemoresistance mechanisms. Hospital Sant Joan de Déu - Esplugues HSJD. April 2012 - March 2015. PI: Cinzia Lavarino.
- CHD5, PAFAH1B1 and NME1: Markers for prognosis and prediction of therapeutic response in neuroblastoma. Chemoresistance mechanisms. Instituto de Salud Carlos III (ISCIII). January 2012 - December 2015. PI: Cinzia Lavarino.
- Limited distribution of drugs in tumoral microenvironment as the cause of chemoresistance in neuroblastoma. Fundación Banco Bilbao Vizcaya Argentaria. January 2012 - December 2015. PI: Angel Montero-Carcaboso.
- Limited distribution of drugs in tumoral microenvironment as the cause of chemoresistance in neuroblastoma. Call for proposals: National R&D&I Programme, Fundamental Research Projects Subprogramme 2011; Category A: Young Researcher. Ministerio de Ciencia e Innovación. January 2012 - December 2015. PI: Angel Montero-Carcaboso.
- Neuroblastoma NEN. Associació de familiars i amics de pacients amb neuroblastoma. January 2012 -. PI: Cinzia Lavarino.
- Research on Ewings sarcoma - Asociación Pablo Ugarte. Asociación Pablo Ugarte. May 2011 -. PI: Jaume Mora.
- Neuroblastoma Chemo. Chemotherapy of neuroblastoma. Marie Curie Fellowship PIRG08-GA-2010-276998 European Commission. March 2011 - March 2015. PI: Angel Montero-Carcaboso.

Clinical trials

- Multi-centre single-group phase II trial to determine the effectiveness and safety of CTL019 in paediatric patients with relapsed/refractory acute B-cell lymphoblastic leukaemia. December 2015 -. PI: Susana Rives.
- Long-term follow-up of patients treated with CAR T-cell therapy targeted at CD19 based on lentiviral vectors. December 2015 -. PI: Susana Rives.
- Placebo-controlled randomised phase III clinical trial to assess the effectiveness and safety of MK 0517/fosaprepitant and ondansetron versus ondansetron in the prevention of chemotherapy-induced nausea and vomiting (CINV) in paediatric patients receiving emetogenic chemotherapy. October 2015 -. PI: Jaume Mora.
- Open multi-centre early-phase study of the safety and pharmacokinetics of anti-pd-l1 (mpdl3280a) antibody in paediatric patients and young adults with previously treated solid tumours. October 2015 -. PI: Jaume Mora.
- Uncontrolled open multi-centre phase III study to assess the pharmacokinetics, safety and effectiveness of BAY 94-9027 for the prophylaxis and treatment of haemorrhages in previously treated children (age < 12 years) with severe haemophilia A. October 2015 -. PI: Ruben Berruenco.
- Open multi-centre phase 2 study to assess the pharmacokinetics, pharmacodynamics, safety and activity of azacitidine and to compare azacitidine with historical controls in paediatric subjects with new diagnosis of advanced myelodysplastic syndrome or juvenile myelomonocytic leukaemia before hematopoietic stem cell transplantation. September 2015 -. PI: Albert Català.
- Single-dose trial to assess the formulation of rivaroxaban in dry powder for oral suspension in children from age 6 months to 12 years with previous thrombosis. July 2015 -. PI: Ruben Berruenco.
- Phase I-II study of vinblastine in combination with nilotinib in children, adolescents and young adults with refractory or recurrent low-grade glioma. May 2015 -. PI: Ofelia Cruz.
- International Randomised Controlled Trial of Chemotherapy for the Treatment of Recurrent and Primary Refractory Ewing Sarcoma. April 2015 -. PI: Jaume Mora.
- Multi-centre international clinical programme for the diagnosis and treatment of children, adolescents and young adults with Ependymoma (SIOP EP-II). March 2015 -. PI: Ofelia Cruz.
- Uncontrolled open multi-centre phase III study to assess the safety of BAY 81-8973 in children with severe haemophilia A under prophylactic treatment. January 2015 -. PI: Ruben Berruenco.
- Open single-arm study to evaluate the long-term safety and efficacy of romiplostim in thrombocytopenic paediatric patients with immune thrombocytopenic purpura (ITP). AMGEN. October 2014 -. PI: Ruben Berruenco.
- Open randomised active comparator-controlled clinical trial to evaluate the safety, efficacy, and pharmacokinetic and pharmacodynamic properties of oral rivaroxaban during 30 days of treatment, in children with diverse manifestations of venous thrombosis. Bayer Pharma AG. September 2014 -. PI: Ruben Berruenco.
- Multi-centre randomised open active comparator-controlled clinical trial to evaluate the efficacy and safety of rivaroxaban at a dose adjusted to the age and weight of children with acute venous thromboembolism. Bayer Pharma AG. September 2014 -. PI: Ruben Berruenco.
- Multi-centre open single-arm two-part phase I/IIA study to determine the safety, tolerability and pharmacokinetics of oral dabrafenib in children between the ages of 12 months and 18 years with advanced solid tumours and positive BRAF V600 mutation. Glaxosmithkline. September 2014 -. PI: Jaume Mora.
- Uncontrolled single-centre phase II clinical trial of dinutuximab (CH 14.18) with the cytokines GM-SCF (granulocyte-macrophage colony-stimulating factor) and IL-2 in patients with high-risk neuroblastoma who are not candidates for other immunotherapy trials. Fundació Privada per a la Recerca i la Docència Sant Joan de Déu. May 2014 -. PI: Jaume Mora.
- Project for updating the study on congenital haemophilia in Spain (PUCHS). Retrospective non-interventionist multi-centre study of the prevalence of haemophilia in Spain. Novo Nordisk Pharma. May 2014 -. PI: Teresa Toll.
- Multi-centre open phase I increasing-dose study of a single group, with monitoring of efficacy, to evaluate vemurafenib

Molecular and cellular biology of developmental tumours

Groups
Projects
Clinical Trials
Publications
Networks



(ro5185426) in children and adolescents with surgically incurable and unresectable melanoma in stage iiiic or iv presenting BRAF V600 mutations. F. Hoffmann- La Roche Ltd; Hospital Sant Joan de Déu - Esplugues HSJD; Quintiles. February 2014 -. PI: Ofelia Cruz.

- Formation of a Spanish cohort of childhood cancer survivors: SPAIN-CCSS cohort. January 2014 -. PI: Héctor Salvador.
- Randomised open comparative combined phase 1/2 dose-finding study to evaluate the efficacy and safety of plerixafor together with standard regimes for mobilisation of haematopoietic stem cells in peripheral blood, and subsequent harvest by apheresis, versus only standard regimes for mobilisation in paediatric patients between the ages of 2 and 18 years with solid tumours meeting requirements for autologous transplantations. January 2014 -. PI: Susana Rives.
- Observational prospective study of the expression of ABCB1/P-glycoprotein as a factor for the biological stratification of non-metastatic osteosarcoma of the extremities. Grupo Español de Investigación en Sarcoma. January 2014 -. PI: Jaume Mora.
- Phase II study of ipilimumab in children and adolescents (from 12 to <18 years) with unresectable malignant melanoma in stage III or stage IV, previously treated or untreated. Bristol-Myers Squibb International Corporation (BMSIC); Hospital Sant Joan de Déu - Esplugues HSJD. November 2013 -. PI: Ofelia Cruz.
- Rare sarcomas register: A tool to help to evaluate the number of cases of each subtype and their therapeutic orientation by the Spanish Research Group on Sarcomas (GEIS). Grupo Español de Investigación en Sarcoma. November 2013 -. PI: Jaume Mora.
- Prospective multi-centre phase II trial with gemcitabine and rapamycin as second-line treatment of metastatic osteosarcoma. GEIS 29. January 2013 -. PI: Jaume Mora.
- Monoclonal chimeric recombinant CH 14.18 antibody. April 2013 -. PI: Jaume Mora.
- Phase I/II study of sunitinib in young patients with advanced digestive stroma tumour. Cetir Centre Mèdic; Hospital Sant Joan de Déu - Esplugues HSJD; Pfizer, S.L.U. April 2013 -. PI: Ofelia Cruz.
- Controlled randomised open multi-centre phase III study to evaluate the safety and efficacy of oral LDE225 versus temozolomide in recurrent medulloblastoma patients with activation of the Hh pathway. Hospital Sant Joan de Déu - Esplugues HSJD; Novartis Farmacèutica. April 2013 -. PI: Ofelia Cruz.
- Multi-centre adaptive randomised open phase IB study of the pharmacokinetics and pharmacodynamics of oseltamivir (Tamiflu®) in the treatment of influenza in immunocompromised patients between the ages of 0 and 18 years, with confirmed influenza infection. F. Hoffmann- La Roche; Hospital Sant Joan de Déu - Esplugues HSJD. January 2013 -. PI: Ruben Berrueto.
- Active comparator-controlled randomised partly masked phase IV study to evaluate the pharmacokinetics and pharmacodynamics, safety and tolerability of fosaprepitant in paediatric patients for the prevention of emetogenic chemotherapy-induced nausea and vomiting (CINV). Hospital Sant Joan de Déu - Esplugues HSJD; Merck, Sharp & Dohme de España. October 2012 -. PI: Jaume Mora.
- Intergroup study for children and adolescents with non-Hodgkin's B lymphoma or lymphoblastic B-cell leukaemia; evaluation of effectiveness and safety of rituximab in high-risk patients. Hospital Sant Joan de Déu - Esplugues HSJD; Sociedad Española de Hematología y Oncología Pediátricas. July 2012 -. PI: Veronica Celis.
- Randomised open comparative multi-centre phase II study of bevacizumab treatment in paediatric patients with recently diagnosed supratentorial high-grade glioma. F. Hoffmann-La Roche. April 2012 -. PI: Ofelia Cruz.
- Multi-centre paediatric register of disease in essential thrombocythaemia (ET). Shire Pharmaceuticals Iberica. May 2011 -. PI: Teresa Toll.
- Prospective multi-centre uncontrolled phase II clinical trial of intensive chemotherapy, surgery and radiotherapy in tumours of the Ewing's sarcoma family in children, adolescents and adults. Ministerio de Sanidad y Política Social. January 2010 - June 2015. PI: Jaume Mora.
- First intergroup international study of classic Hodgkin's lymphoma in children and adolescents. Euronet-PHL-C1 January 2009 -. PI: Ofelia Cruz.

Publications

- Barneda-Zahonero B, Collazo O, Azagra A, Fernández-Duran I, Serra-Musach J, Islam AB, Vega-García N, Roberta Malatesta, Camós M, Gómez A, Román-González L, Vidal A, López-Bigas N, Villanueva A, Esteller M, and Parra M. The transcriptional repressor HDAC7 promotes apoptosis and c-Myc downregulation in particular types of leukemia and lymphoma. *Cell Death Dis* 2015. FI: 5,014(Q2).
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- Cockle JV, Picton S, Levesley J, Ilett E, Carcaboso AM, Short S, Steel LP, Melcher A, Lawler SE, and Brüning-Richardson A. Cell migration in paediatric glioma: characterisation and potential therapeutic targeting. *Br J Cancer* 2015. 112: 693-703. FI: 4,836(Q1).
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- Garzón M, García-Fructuoso G, Suñol M, Mora J, and Cruz O. Low-grade gliomas in children: single institutional experience in 198 cases. *Childs Nerv Syst* 2015. 31: 1447-1459. FI: 1,114(Q3).
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- Menacho-Márquez M, Rodríguez-Hernandez CJ, Villaronga MA, Pérez-Valle J, Gadea J, Belandia B, and Murguía JR. eIF2 kinases mediate β -lapachone toxicity in yeast and human cancer cells. *Cell Cycle* 2015. 14: 630-640. FI: 4,565(Q2).
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Vertical areas

3

AREA

Molecular and cellular biology of developmental tumours

Groups
Projects
Clinical Trials
Publications
Networks

- Niemeyer CM, Loh ML, Cseh A, Cooper T, Dvorak CC, Chan R, Xicoy B, Germing U, Kojima S, Manabe A, Dworzak M, De Moerloose B, Starý J, Smith OP, Masetti R, Catala A, Bergstraesser E, Ussowicz M, Fabri O, Baruchel A, Cavé H, Zwaan M, Locatelli F, Hasle H, van den Heuvel-Eibrink MM, Flotho C, and Yoshimi A. Criteria for evaluating response and outcome in clinical trials for children with juvenile myelomonocytic leukemia. *Haematologica* 2015. 100: 17-22. FI: 5,814(Q1).
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Networks

- Innovative Therapies for Children with Cancer. European consortium.

ANNUAL SCIENTIFIC
REPORT
2015

SJD

Sant Joan de Déu
Institut de Recerca



Vertical areas

4

AREA

Paediatric Diseases
originating in early life

Groups
Projects
Clinical Trials
Publications
Supervised theses
Networks

Metabolic endocrinology

The Metabolic Endocrinology Group includes the research lines on ovarian hyperandrogenism, insulin resistance, obesity, metabolic syndrome and diabetes. Of special note within this scope is the research line on foetal growth restriction, with potential irreversible

consequences for children's development. The study of the subsequent stages, from intrauterine life through adolescence, will allow setting up preventative strategies to delay or avoid the development of the associated complications.

Research team

Coordinator Lourdes Ibáñez
Researchers Paula Casano, Marta Díaz, Giorgia Sebastiani, Jovita Quilez, Rita Malpique

Perinatal origin of adult diabetes: animal models

The Developmental Origins of Health and Disease hypothesis proposes that early-life environmental exposures increase the risk of adult chronic diseases such as obesity and type 2 diabetes. The specific molecular mechanisms that lead to such long-lasting effects remain poorly characterized. Our laboratory aims to understand the molecular mechanisms that might link early neonatal overfeeding/childhood obesity and later

risk of obesity and diabetes. To achieve this objective we combine work with animal models (mouse) and human clinical cohorts. A major emphasis in the laboratory is exploring the potential role of Epigenetic mechanisms, including DNA methylation and small non-coding RNAs, in mediating adult programming of disease risk and even transgenerational inheritance of diabetes risk in response to nutritional imbalances during development.

Research team

Coordinator Josep Jiménez-Chillaron
Researchers Rubén Díaz Naderi, Marta Ramon Krauel, Sílvia Ribó, Ivonne Palacios
Technician Judith Cebrià

Vertical areas

4

AREA

Paediatric
Diseases
originating
in early life

Groups

Projects
Clinical Trials
Publications
Supervised theses
Networks

Neonatal physiopathology

Genetic potential and foetal environment may have irreversible consequences for a person's development and they may be the origin of disorders in the adult. The study of factors that influence the various stages of life: periconceptual, intrauterine, perinatal and postnatal will allow advances to be made in our knowledge

of the conditioning factors which may interfere with correct development. Moreover, the follow-up of the nutritional and health-related interventions carried out on babies up to their adolescence and adulthood will permit medical teams to develop better treatments and preventive strategies.

Research team

Coordinator	Alfredo Garcia-Alix
Technician	Thais Agut, Marta Camprubi, Isabel Iglesias, Martin Iriondo, Ana Martin, Africa Pertierra, Ana Riverola

Projects

- Doctorat Industrial 2015. Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. December 2015 - December 2018. PI: Lourdes Ibañez Toda.
- Prematurity - CCD. Clientes Diversos. September 2015 - December 2020. PI: Martin Iriondo Sanz.
- Maternal diet, microbiome and infant feeding practice: roles in initializing the fetal gut microbiome and establishing the infant gut microbiome. Icahn School of Medicine at Mount Sinai. July 2015 - July 2018. PI: Lourdes Ibañez Toda.
- Intensificació 2014_Dra. Lourdes Ibañez. Instituto de Salud Carlos III (ISCIII). February 2015 - February 2016. PI: Lourdes Ibañez Toda.
- Merck Agreement: 2nd Update Conference on Small for Gestational Age. Merck S.L.U. January 2015 - December 2016. PI: Lourdes Ibañez Toda.
- Prevention and treatment of hyperandrogenism associated with hyperinsulinism in girls in childhood and adolescence: a treatment focused on adipose tissue. Ministerio de Economía Y Competitividad (MINECO). January 2015 - January 2016. PI: Lourdes Ibañez Toda.
- Childhood obesity and risk of metabolic syndrome in the adult: epigenetic mechanisms. Instituto de Salud Carlos III (ISCIII). January 2015 - December 2017. PI: José Carlos Jiménez Chillarón.
- We will improve the life of children with diabetes. Banco Bilbao Vizcaya Argentaria, S.A. January 2015 - December 2015. PI: Ruben Díaz Naderi.
- Modifying maternal nutrition during lactation to improve offspring life-course health outcomes. Ajinomoto Co., Inc. January 2015 - January 2017. PI: Carles Lerin Martinez.
- Intelligent monitoring of premature newborn patients hospitalised in intensive care units. Ministerio de Economía Y Competitividad (MINECO). January 2015 - January 2016. PI: Ana Riverola De Veciana.
- Improving the life of children with diabetes. Fundación Banco Bilbao Vizcaya Argentaria. November 2014 - November 2015. PI: Ruben Díaz.
- Evaluation of cardiovascular risk in the first two years of life in children born large or small for their gestational age. Fundación Salud 2000. May 2014 - April 2017. PI: Lourdes Ibañez.
- Programme for intensification of research activity in the Spanish National Health System 2013. INT13/00095. Instituto de Salud Carlos III. February 2014 - February 2015. PI: Lourdes Ibañez.
- Early biomarkers of the safety and efficacy of nutritional support in extremely preterm infants (EPI). PI13/01562. Instituto de Salud Carlos III. January 2014 - December 2016. PI: Isabel Iglesias.
- Effect of prenatal and perinatal changes on postnatal development. Disorders of foetal origin. Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. SGR 2014. 2014SGR512. January 2014 - December 2016. PI: Lourdes Ibañez.
- Research personnel training, candidate: Luis Miguel Río. Ministerio de Ciencia e Innovación (MINECO). 2013 - 2017. PI: Carles Lerin.
- IRBETAINE-CIG Molecular mechanisms underlying the development of insulin resistance: role of betaine supplementation. European Commission. July 2013 - August 2015. PI: Carles Lerin.
- Ramon y Cajal researcher, Dr. Carles Lerin. Ministerio de Ciencia e Innovación (MINECO). July 2013 - January 2016. PI: Carles Lerin.
- Metabolic and biochemical effects of betaine on insulin sensitivity and physical exercise. Ministerio de Ciencia e Innovación (MINECO). January 2013 - January 2017. PI: Carles Lerin.
- Fire Squadron Grant 2012 Newborns. Hospital Sant Joan de Déu Barcelona. May 2012 - April 2015. PI: Isabel Iglesias.
- Non-genomic inheritance of diabetes in a mouse model of neonatal overnutrition: potential role of epigenetic mechanisms. Instituto de Salud Carlos III. January 2012 - January 2017. PI: Josep Jiménez-Chillarón.
- Genetic determinants of prenatal and postnatal growth: association with early markers of cardiovascular and metabolic syndrome risk. Instituto de Salud Carlos III. January 2012 - December 2015. PI: Lourdes Ibañez.
- Metabolic and biochemical effects of betaine on insulin sensitivity and physical exercise. Ministerio de Ciencia e Innovación. January 2012 - December 2015. PI: Carles Lerin.
- Prognostic factors of brain injury in fetuses with early restricted intrauterine growth. Instituto de Salud Carlos III. January 2012 - December 2015. PI: Antonio Vela.

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Clinical trials

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- Placebo-controlled double-blind randomised phase 3 trial of beloranib on obese subjects with Prader-Willi syndrome to assess eating behaviour, total body weight and safety over 52 weeks. September 2015 - March 2016. PI: Marta Ramon.

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- Identification of circulating microRNAs in short children born small for gestational age: Relationship with pre- and postnatal growth and metabolic parameters. October 2014 -. PI: Abel López-Bermejo. Investigator: Lourdes Ibáñez.
- Randomised, open, active-controlled, multinational, dose-escalation trial investigating safety, tolerability, pharmacokinetics and pharmacodynamics of a single dose of long-acting growth hormone (NNC0195-0092) compared with daily dosing of Norditropin® SimpleXx® in children with growth hormone deficiency. Novo Nordisk. January 2014 -. PI: Lourdes Ibáñez.
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Supervised theses

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- Body composition and endocrine-metabolic profile in children born large for gestational age. Doctoral Candidate: Miriam Pérez Cruz. Director: Lourdes Ibáñez. March 2015.

Networks

- CIBERDEM: Centre for Biomedical Network Research on Diabetes and Associated Metabolic Disorders. PI: Lourdes Ibáñez.



Vertical areas

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Infectious diseases and systemic inflammatory response in pediatrics

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Vaccine-preventable diseases

The human species' first contact with many microorganisms takes place in childhood, when children have not been infected and have not developed the disease. Vaccination in early ages of life has proven to be an effective tool in the fight against infectious diseases. In order to evaluate the best vaccination strategies in a specific population, a good molecular characterization of the microorganisms circulating in the respective community is essential since microorganisms diversify genetically to adapt themselves as well as possible to a specific environment. The overall objective is to try to improve the diagnosis and characterization of pediatric infectious diseases through specific, sensitive, rapid, simple and inexpensive techniques.

Our research is divided into two lines of action: (1) epidemiologic surveillance of vaccine-preventable diseases, and (2) diagnostic innovation.

The general goal of the first line is the molecular epidemiologic surveillance of three pathologies with a significant rate of occurrence among children: pneumococcal disease, whooping

cough and meningococcal disease.

- Evaluation of conjugate vaccines in the prevention of pneumococcal disease.

- Host-pathogen interaction in the development of invasive diseases produced by pathobionts such as *S. pneumoniae*.

With respect to the infectious disease diagnostic line, the new isothermal molecular expansion techniques allow the development of molecular devices in point-of-care format without manipulation by the user. These techniques are valuable tools in the optimisation of processes in both the outpatient and inpatient settings, in public healthcare as well as in the care of individual patients. Additionally, photonics-based technology allows the development of high-precision automated optical devices, significantly improving the sensitivity of today's microscopes.

The general goal is to improve the diagnosis and characterisation of paediatric infectious diseases by means of sensitive, specific, rapid, simple and low-cost techniques.

Research team

Coordinator Carmen Muñoz-Almagro

Researchers Pedro Brotons de los Reyes, Ana Valero, Mariona Fernández de Sevilla, Juan José García-García, Susana Hernández-Bou, Cristian Launes, Cristina Esteva, María Andres, Desiree Henares, Miriam Triviño, Anny Camelo

Technician Carlos Cisneros, Amaresh Pérez Argüello, Jessica Saucedo

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Vertical transmitted infections and infections in vulnerable patients

The general goal of the research on vertical transmission infections and infections in vulnerable patients is to widen our knowledge of the natural history, transmission-associated factors, diagnosis,

prognosis and treatment of infections which are transmitted from mother to child (human immunodeficiency virus, hepatitis B and C virus, acute cytomegalovirus infection, toxoplasma).

Research team

Coordinator Claudia Fortuny

Tuberculosis

Research on the adherence and toxicity associated with first-line anti-tuberculosis drugs in the paediatric age, on the pharmacokinetic characteristics of these drugs on newborns and pre-school children, on the new tuberculosis diagnostic techniques in healthy children, on children from high tuberculosis endemic areas and on immunosuppressed children, and on the clinical-epidemiological

characteristics of tuberculosis in childhood in our area. Research on the epidemiological and clinical characteristics of infections by non-tuberculous mycobacteria in the pediatric age. Reference unit for the performance of clinical trials on new diagnostic methods and therapeutic strategies for tuberculosis in the paediatric age.

Research team

Coordinator Antoni Noguera-Julian
Researchers Joan Lluís Vinent Genestar, Eneritz Velasco Arnaitz, Maria Goretti López Ramos,
Aina Martínez Planas, Eva Maria Guix Comellas, Silvia Simó Nebot

Systemic inflammatory response in the paediatric age

The research is focused on the study of the immunologic mechanisms involved in primary and secondary immunodeficiencies, autoinflammatory diseases and paediatric systemic autoimmune diseases.

The various lines of work include the study of primary immunodeficiencies and especially those which are centred on innate immunity (Toll-IL-1R and IL-12/IFN-gamma pathways) and which explain part of the susceptibility to common infections in paediatrics; the study of paediatric common variable immunodeficiency; and the analysis of phenotypes and genotypes. Work is also carried out to improve the knowledge and management of secondary immunodeficiency in paediatric patients and in pregnant women (a research line on the effects on the newborn's immune system of treatment with anti-TNF alpha antibodies in expectant mothers with inflammatory bowel disease). Other lines of work involve the study of autoinflammatory syndromes such as mevalonate kinase deficiency or hyper-IgD syndrome, cryopyrin-associated autoinflammatory syndrome

(CAPS) or familial Mediterranean fever, and autoimmune diseases such as uveitis associated with juvenile idiopathic arthritis, juvenile dermatomyositis, and paediatric vasculitis disorders such as Kawasaki disease.

The goals of this group also include the identification of the genes causing disorders, the study of the involvement of epigenetics and the microbiome, and the recognition of pathogenic pathways, which will allow the development of new treatment proposals.

The groups approach is based on translational research, for which reason its lines of work are carried out at patients side, striving to improve their quality of life. To this end the group has the close cooperation of other practitioners of Hospital Sant Joan de Déu Barcelona and of other prestigious national and international institutions.

The Study Group on Immune Dysfunction Disorders in Paediatrics (GEMDIP) has been recognised as an emerging research group by AQU Catalunya (Agency for the Quality of the University System of Catalonia).

Research team

Coordinator Jordi Anton
Researchers Laia Alsina, Rosa Bou, Estibaliz Iglesias, Judith Sánchez, Joan Calzada,
Violeta Bittermann, Angela Deyà, Mónica Piquer, Anna Esteve-Solé

Projects

- A narrow repertoire of transcriptional modules responsive to pyogenic bacteria is impaired in patients carrying loss-of-function mutations in MYD88 or IRAK4. Colegio de Médicos de Córdoba. December 2015 - December 2016. PI: Laia Alsina Manrique de Lara.
- WINDBIOME_Aerial micro-biome diversity and their role in Kawasaki disease. Fundació Privada Daniel Bravo Andreu. December 2015 - December 2018. PI: Jordi Antón López.
- The Utility of the FilmArray Respiratory Panel Test for the Rational Use of Resources in the Pediatric Intensive Care Unit (PICU). BioFire Diagnostics, LLC. December 2015 - November 2016. PI: Carmen Muñoz Almagro.
- Luminex Evaluation Study. Luminex B.V. June 2015 - November 2015. PI: Carmen Muñoz Almagro.
- Setting up a sentinel system to assess the burden of whooping cough in EU/EEA. Service Agreement. European Centre for Disease Prevention and Control (ECDC). September 2015 - September 2016. PI: Carmen Muñoz Almagro.
- Assessment of somatic mosaicism in the NOD2 gene in seronegative juvenile idiopathic arthritis. Acadèmia de Ciències Mèdiques i de la Salut de Catalunya i de Balears. July 2015 - June 2017. PI: Estibaliz Iglesias Jimenez.
- Low-cost photonic reader for rapid detection of invasive bacterial diseases (REDSEA). Fundació Privada CELLEX. June 2015 - June 2016. PI: Carmen Muñoz Almagro.
- M-BAE_Training programme on infections of paediatric patients subjected to allogeneic transplantation of haematopoietic precursors. Instituto de Salud Carlos III (ISCIII). April 2015 - May 2015. PI: Antoni Noguera Julián.
- Contracts for intensification of the research activity in the National Health System (SNS) 2014. Beneficiario: Carmen Muñoz. Instituto de Salud Carlos III (ISCIII). April 2015 - April 2016. PI: Carmen Muñoz Almagro.
- Contrato Sara Borrell_Beneficiario: Irene Latorre. Instituto de Salud Carlos III (ISCIII). January 2015 - November 2015. PI: Carmen Muñoz Almagro.
- Phase IIA open clinical trial on absorption of an isoniazid suspension of 10 mg/ml for treatment of tuberculosis infection in children under the age of 6 years. Instituto de Salud Carlos III (ISCIII). January 2015 - December 2017. PI: Antoni Noguera Julián.
- Pharmacological treatment of tuberculosis in infants under the age of 6 months: validation of the single daily dose of isoniazid at 10mg/kg of weight. Fundación Mercedes Armengou Riera (MAR). January 2015 - December 2016. PI: Antoni Noguera Julián.
- Determination of haptoglobin levels in synovial fluid of the temporomandibular joint (TMJ) in patients with juvenile idiopathic arthritis. Correlation with exploratory findings and magnetic resonance imaging with contrast medium. Sociedad Española de Reumatología. January 2015 - December 2016. PI: Estibaliz Iglesias Jimenez.
- Be a hope donor. Banco Bilbao Vizcaya Argentaria, S.A. January 2015 - December 2015. PI: Jordi Antón López.
- Diagnosis of tuberculosis infection in paediatric patients under treatment with tumour necrosis factor alpha inhibitors: comparative study between tuberculin test and IGRA tests. Sociedad Española de Reumatología. January 2015 - December 2016. PI: Joan Calzada Hernández.
- Development of new genetic diagnostic techniques by means of high-performance sequencers, and their application to the diagnosis of osteogenesis imperfecta. Sociedad Española de Reumatología. January 2015 - December 2016. PI: Rosa Bou i Torrent.
- Evaluation of somatic mosaicism in the NOD2 gene in seronegative juvenile idiopathic arthritis. Sociedad Española de Reumatología Pediátrica. December 2014 - December 2016. PI: Estibaliz Iglesias.
- Utility of 256-channel multi-slice computed tomography to evaluate cardiac involvement in Kawasaki disease. Sociedad Española de Reumatología Pediátrica. December 2014 - December 2016. PI: Joan Calzada.
- Analysis of autoinflammatory mechanisms mediated by the NF-κB pathway and the inflammasome/caspase-1/IL-1 axis in juvenile idiopathic arthritis with negative antinuclear antibodies. Societat Catalana de Reumatologia. June 2014 - June 2016. PI: Estibaliz Iglesias.

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- Study of the primary and secondary defects of the IL12-IFN γ -TNF α pathway with susceptibility to intracellular microorganism infection. Hospital Sant Joan de Déu Barcelona. March 2014 - February 2017. PI: Laia Alsina.
- Analysis of autoinflammatory mechanisms mediated by the NF- κ B pathway and the inflammasome/caspase-1/IL-1 axis in juvenile idiopathic arthritis with negative antinuclear antibodies. 2nd Honourable mention. Beca Mutual Mèdica. January 2014 - December 2015. PI: Estibaliz Iglesias.
- Analysis of autoinflammatory mechanisms mediated by the NF- κ B pathway and the inflammasome/caspase-1/IL-1 axis in juvenile idiopathic arthritis with negative antinuclear antibodies. Sociedad Española de Reumatología. January 2014 - December 2015. PI: Estibaliz Iglesias.
- Defects in innate immunity and coinfection with respiratory virus: the perfect storm for developing invasive pneumococcal disease in children? Instituto de Salud Carlos III. PI13/01729. January 2014 - December 2016. PI: Carmen Muñoz-Almagro.
- Longitudinal study of mitochondrial DNA mutations in children exposed to HIV (infected and non-infected) and to antiretroviral drugs. Instituto de Salud Carlos III. PI13/01738. January 2014 - December 2016. PI: Clàudia Fortuny.
- Consolidated group on infectious diseases in the paediatric age. Consolidated research group on infectious diseases in the paediatric age. 2014SGR505. Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. January 2014 - December 2016. PI: Carmen Muñoz-Almagro.
- Identification of prognostic factors in muscle biopsy for the diagnosis of patients with juvenile dermatomyositis. Sociedad Española de Reumatología Pediátrica. January 2014 - December 2015. PI: Estibaliz Iglesias.
- Validation of the single daily dose of isoniazid at 10 mg/kg weight in newborns under the age of 3 months. Instituto de Salud Carlos III. PIC14/00228. January 2014 - December 2016. PI: Antoni Noguera.
- RESPOC. Fast, cost-effective, user-friendly point of care (POC) instrument for detection of respiratory pathogens, including *Streptococcus pneumoniae*. European Commission. December 2013 - May 2016. PI: Carmen Muñoz-Almagro.
- Molecular diagnosis of food allergy to shellfish in the paediatric population. Sociedad Española de Inmunología Clínica, Alergia y Asma Pediátrica. November 2013 - November 2015. PI: Olga Dominguez.
- Study of the primary and secondary defects of the IL-12/IFN- γ /TNF- α pathway with susceptibility to infection by intracellular micro-organisms. Instituto de Salud Carlos III (ISCIII). PI12/01990. January 2013 - December 2015. PI: Laia Alsina.
- Validation of the Minimally Invasive Autopsy (MIA) tool for cause of death investigation in developing countries. Bill & Melinda Gates Foundation. December 2012 - December 2015. PI: Carmen Muñoz-Almagro.
- SHARE-Single HUB and Access Point for Paediatric Rheumatology in Europe (SHARE). European Commission. September 2012 - August 2015. PI: Jordi Antón.
- Support technician at the Sequencing Unit. Instituto de Salud Carlos III (ISCIII). CA11/00447. Beneficiary: Cristina Esteva. April 2012 - April 2015. PI: Carmen Muñoz-Almagro.
- Use of dried-blood spot (DBS) samples for detection and molecular surveillance of *Streptococcus pneumoniae* in developing countries. Hospital Sant Joan de Déu - Esplugues HSJD. April 2012 - March 2015. PI: Carmen Muñoz-Almagro.
- Effectiveness of 13-valent pneumococcal conjugate vaccine in the prevention of invasive pneumococcal disease in children. A matched case-control study. Instituto de Salud Carlos III (ISCIII). PI11/02081. January 2012 - December 2015. PI: Juan José García-García.
- Predoctoral training grants in health research. Beneficiary: Eva del Amo. Instituto de Salud Carlos III (ISCIII). FI11/00720. September 2011 - September 2015. PI: Carmen Muñoz-Almagro.
- Reference laboratory for molecular epidemiologic monitoring of meningococcal disease, invasive pneumococcal disease and pertussis. Agència de Salut Pública de Catalunya. January 2011 - December 2015. PI: Carmen Muñoz-Almagro.
- Intensification for healthcare activity. Fundación Godia. PI: Carmen Muñoz-Almagro.

Clinical trials

- Strategy for maintenance of HIV suppression in children with elvitegravir + darunavir/ritonavir. December 2015 -. PI: Clàudia Fortuny.
- Placebo-controlled double-blind randomised multi-centre study to assess the effectiveness and safety of STG320 sublingual tablets of house dust mite allergen extracts in adults and adolescents with house dust mite allergic rhinitis. November 2015 -. PI: Maria Teresa Giner.
- Assessment of prevalence of unidentified mucopolysaccharidosis I, II, IVA and VI in patients with juvenile idiopathic arthritis with low inflammatory markers. November 2015 -. PI: Jordi Antón.
- Tolerance to the introduction of a formula based on highly hydrolysed whey proteins enriched with *Lactobacillus fermentum* (Damira-Pro1®) in children with allergy to cows milk protein. November 2015 -. PI: Ana M^a Plaza.
- Placebo-controlled double-blind study with parallel groups for 52 weeks to assess the effectiveness and safety of a fixed dose of 110 mg of reslizumab via subcutaneous route in patients with uncontrolled asthma and a high concentration of eosinophils in blood. October 2015 -. PI: Ana M^a Plaza.
- Open multi-centre phase Ib study to investigate the pharmacokinetics, pharmacodynamics and safety of tocilizumab after subcutaneous administration in patients with systemic juvenile idiopathic arthritis. September 2015 -. PI: Jordi Antón.
- PID-RSV study: epidemiological study on the rate of acute respiratory infections (ARI) caused by syncytial respiratory virus (SRV) requiring hospitalisation in paediatric populations with primary immunodeficiency (PID). September 2015 -. PI: Laia Alsina.
- National retrospective-prospective multi-centre study on the value of IGRAs in the diagnosis of adenitis caused by non-tuberculous mycobacteria, and on its various therapeutic approaches. July 2015 -. PI: Antoni Noguera.
- Study on the safety, tolerability and pharmacokinetics of two formulations of posaconazole (POS) via intravenous route (IV) and in powder for oral suspension, in immunosuppressed paediatric patients with neutropaenia. July 2015 -. PI: Clàudia Fortuny.
- Active-control multi-centre randomised single-blind study to assess the safety, tolerability, pharmacokinetics and effectiveness of ceftazidime and avibactam when provided with metronidazole versus meropenem in children from age 3 months to under 18 years with complicated intra-abdominal infections (cIAIs). July 2015 -. PI: Clàudia Fortuny Guash.
- Phase I study to assess the pharmacokinetics and safety of tocilizumab (TCZ) in patients under the age of 2 years with active systemic juvenile idiopathic arthritis (SJIA). June 2015 -. PI: Jordi Antón.
- Phase III study of tedizolid phosphate for 6 days from i.v. to oral in comparison to the comparator for 10 days in subjects aged 12 to < 18 years with complicated skin and skin structure infection (cSSSI). May 2015 -. PI: Juan José García.
- Phase IIa multi-centre trial to assess the effectiveness and safety of anakinra in patients with Kawasaki disease resistant to intravenous immunoglobulin. April 2015 -. PI: Jordi Antón.
- Double-blind randomised multi-centre study with parallel groups, controlled with vehicle to assess safety and effectiveness of calcitriol 3 mcg/g in ointment applied twice daily for 8 weeks in paediatric patients (aged 2 to 12 years) with mild to moderate plaque psoriasis. March 2015 - March 2016. PI: M^o Asunción Vicente Villa.
- Study to assess the financial impact of rotavirus (RV) at Catalan hospital centres. March 2015 - November 2015. PI: Juan José García.
- Open multi-centre study to assess the safety, tolerability, pharmacokinetics and effectiveness of ceftaroline in newborns and infants under 60 days of age with late-onset sepsis. March 2015 -. PI: Clàudia Fortuny.
- Crossover controlled randomised multi-centre trial to assess the hypoallergenicity of a new amino acid-based formula in children allergic to cows milk. March 2015 -. PI: Ana M^a Plaza Martín.
- Influence of immunogenicity of anti-TNF drugs on the effectiveness of treatment in juvenile idiopathic arthritis using serum calprotectin as activity marker of the disorder. Multi-centre trial. "ITACA Study". February 2015 -. PI: Jordi Antón.
- TBTC Study 36 - Platform for assessment of results of tuberculosis treatment. Observational study of treated individuals with pulmonary tuberculosis (TB). February 2015 -. PI: Antoni Noguera.

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- PCP00147 - Dysphagia area. Promoter: FRESENIUS. Fresenius Kabi España, S.A.U. January 2015 - April 2016. PI: Javier Martín De Carpi.
- Randomised phase 3 multi-centre study with parallel groups, masked from the researcher, to investigate the safety and effectiveness of fidaxomicin in oral suspension or tablets every 12 hours, and vancomycin in oral solution or capsules every 6 hours for 10 days, in paediatric subjects with diarrhoea associated with *Clostridium difficile*. January 2015 -. PI: Clàudia Fortuny.
- β -SPECIFIC 4 patients: Paediatric efficacy and safety study with canakinumab as first-line treatment. Open study on the efficacy and safety of dose reduction or dose interval lengthening of canakinumab (ACZ885) in patients with systemic juvenile idiopathic arthritis (SJIA). November 2014 -. PI: Rosa Bo/Jordi Antón.
- Prospective, multi-centre, single-arm study to evaluate the efficacy, safety and pharmacokinetics of denosumab in children with osteogenesis imperfecta. October 2014 -. PI: Rosa Bou.
- Retrospective study of food-allergic patients (milk and/or eggs and/or vegetables) treated with omalizumab as sole treatment or associated with oral immunotherapy based on a multi-centre register (OmaBase). August 2014 -. PI: Ana M^a Plaza.
- Long-term extension study to evaluate the safety and efficacy of tocilizumab administered subcutaneously to patients with polyarticular juvenile idiopathic arthritis and systemic juvenile idiopathic arthritis. June 2014 -. PI: Rosa Bou Torrent.
- Randomised, double-blind, placebo-controlled study of canakinumab in patients with hereditary periodic fevers (TRAPS, HIDS or FMFrc) with randomised withdrawal/frequency reduction of the dose and subsequent long-term open treatment phase. Novartis Farmacèutica. May 2014 -. PI: Jordi Antón.
- Tolerance to 2 highly hydrolysed childrens milk formulas based on rice proteins and casein in children with allergy to cows milk proteins. Biosearch. May 2014 -. PI: Ana M^a Plaza.
- Validation of the daily single dose of isoniazid at 10 mg/kg of weight in newborns aged under 3 months. April 2014 -. PI: Antoni Noguera.
- Spanish Register of Uveitis Associated with Juvenile Idiopathic Arthritis. Sociedad Española de Reumatología. April 2014 -. PI: Jordi Antón.
- CEntroS Project for Early Assistance for Respiratory Allergy (CESAR Project). Stallergenes Ibérica. March 2014 -. PI: Ana M.^a Plaza.
- Continued access to darunavir/ritonavir (DRV/rtv) for children aged 3 years or more and adolescents infected by HIV-1. March 2014 -. PI: Clàudia Fortuny.
- Phase 2/3, multi-centre, open, multi-cohort, two-part study to evaluate the pharmacokinetics, safety and antiviral activity of elvitegravir (EVG) administered with a base regime (BR) containing a protease inhibitor reinforced with ritonavir (IP/r) in paediatric subjects infected by HIV-1, previously treated with antiretrovirals. Gilead Sciences; Hospital Sant Joan de Déu Barcelona. February 2014 -. PI: Clàudia Fortuny.
- Double-blind randomised multi-centre comparative clinical trial to evaluate the effectiveness, safety and pharmacokinetics of daptomycin in comparison to the active reference treatment in paediatric patients with acute haematogenous osteomyelitis caused by gram-positive organisms. December 2013 -. PI: Juan José García-García.
- Phase III, randomised, double-blind, placebo-controlled study to determine the efficacy, safety and tolerability of lebrikizumab in adolescent patients with uncontrolled asthma treated with inhaled corticosteroids and a second control drug. F. Hoffmann- La Roche; Hospital Sant Joan de Déu Barcelona. November 2013 -. PI: Ana M^a Plaza.
- Multi-centre long-term follow-up study of HLH patients who have been treated with NI-0501, an anti-interferon gamma monoclonal antibody. Novimmune. November 2013 -. PI: Laia Alsina.
- Phase III, multi-centre, open clinical trial to evaluate the pharmacokinetics, efficacy and safety of abatacept administered subcutaneously (s.c.) in children and adolescents with polyarticular juvenile idiopathic arthritis and inadequate response (IR) to biological or non-biological disease-modifying antirheumatic drugs (DMARDs). Bristol-Myers Squibb International Corporation (BMSIC); Hospital Sant Joan de Déu - Esplugues HSJD. September 2013 -. PI: Jordi Antón.
- Multi-centre, randomised, placebo-controlled, double-blind study on parallel groups to evaluate the safety, efficacy and pharmacokinetics of belimumab, an anti-BLyS human monoclonal antibody, plus standard therapy, in paediatric patients with systemic lupus erythematosus (SLE). Glaxosmithkline. September 2013 -. PI: Jordi Antón.
- Phase Ib, multi-centre, open study to investigate the pharmacokinetics, pharmacodynamics and safety of tocilizumab after subcutaneous administration to patients with polyarticular juvenile idiopathic arthritis. Roche Farma. July 2013 -. PI: Jordi Antón.
- Phase 1/2, two-stage, open study on a single group to determine the safety, pharmacokinetics and efficacy of telaprevir in combination with peginterferon alfa-2b and ribavirin in

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paediatric subjects aged 3 to 17 years infected by genotype 1 hepatitis C virus. Hospital Sant Joan de Déu Barcelona; Vertex Pharmaceuticals Incorporated. June 2013 -. PI: Clàudia Fortuny.

- Comparative evaluation of the safety and efficacy of daptomycin versus standard treatment in paediatric patients aged 2 to 17 years with bacteraemia caused by *Staphylococcus aureus*. PharmaNet GmbH. May 2013 -. PI: Antoni Noguera.
- Multi-centre, single-arm, open pilot study to explore the safety, tolerability, pharmacokinetics and efficacy of multiple intravenous administrations of NI-0501, an anti-interferon gamma monoclonal antibody (anti-IFN), in paediatric patients with reactivated primary haemophagocytic lymphohistiocytosis. Novimmune. April 2013 -. PI: Laia Alsina.
- Phase III contact follow-up trial comparing the diagnostic functioning of C-Tb versus QuantiFERON®-TB Gold In-Tube, in combination with a randomised double-blind safety analysis in different parts of the body with C-Tb versus 2 TU of Tuberculin PPD RT23SSI (PPD). Statens Serum Institute. February 2013 -. PI: Antoni Noguera.
- Open extension study to evaluate the long-term safety of etanercept in children and adolescents with extended oligoarticular juvenile idiopathic arthritis, enthesitis-related arthritis or psoriatic arthritis, who previously took part in Protocol 0881A1-3338-WW (B1801014). Pfizer. January 2012 -. PI: Jordi Antón.
- Pharmacovigilance in patients with juvenile idiopathic arthritis (PHARMACHILD) under treatment with biological drugs and/or methotrexate. Register of the Pediatric Rheumatology International Trials Organization (PRINTO)/Pediatric Rheumatology European Society (PRES). January 2012 -. PI: Jordi Antón.
- International, multi-centre, prospective, single-arm, open study to evaluate the safety, efficacy and pharmacokinetics of atazanavir (ATV) powder potentiated with ritonavir (RTV) liquid with an optimised combination of nucleosides in children infected by HIV, untreated or previously treated with antiretrovirals, aged from 3 months to 8 years (Pediatric Atazanavir International Clinical Evaluation: the PRINCE II study). Bristol-Myers Squibb International Corporation (BMSIC). December 2011 -. PI: Clàudia Fortuny.

- Short cycle therapy (SCT) (5 days on/2 days off) in young people infected by HIV-1 (PENTA 16). Fondazione Penta; Hospital Sant Joan de Déu Barcelona. December 2011 -. PI: Clàudia Fortuny.
- Evaluation of early treatment in children with juvenile idiopathic arthritis: retrospective analysis. September 2011 -. PI: Jordi Antón.
- Multi-centre, randomised, double-blind study with dual simulation to evaluate two doses of adalimumab versus methotrexate (MTX) in paediatric patients with chronic plaque psoriasis (Ps). Abbott Laboratories. March 2011 -. PI: M^a Asunción Vicente.
- Multi-centre, longitudinal post-commercialisation observational register of long-term efficacy and safety of Humira® (adalimumab) in children with moderately to severely active polyarticular or polyarticular-course juvenile idiopathic arthritis (JIA). (STRIVE STUDY). Abbott Laboratories. May 2010 -. PI: Jordi Antón.
- Open extension study with canakinumab (ACZ885) in patients with systemic juvenile idiopathic arthritis and active systemic manifestations. Novartis Farmacéutica. June 2009 -. PI: Jordi Antón.
- Open, multi-centre trial on multiple-dose administration pharmacokinetics and on safety and efficacy during 48 weeks of maraviroc in combination with an optimised base treatment designed for subjects aged 2 to 18 years infected by HIV-1 with CCR5 tropism who previously received treatment with antiretrovirals. Pfizer. April 2009 -. PI: Clàudia Fortuny.

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- Baquero-Artigao F, Mellado Peña MJ, Del Rosal Rabes T, Noguera Julián A, Goncé Mellgren A, de la Calle Fernández-Miranda M, Navarro Gómez ML; Working Group on gestational, congenital, and postnatal tuberculosis, Spanish Society of Pediatric Infectious Diseases (PFIC). Guía de la Sociedad Española de Infectología Pediátrica sobre tuberculosis en la embarazada y el recién nacido (I): epidemiología y diagnóstico. *Tuberculosis congénita. An Pediatr (Barc)* 2015. FI: 0,833(Q4).
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Vertical areas

5

AREA

Infectious diseases
and systemic
inflammatory
response
in pediatrics

Groups

Projects

Clinical Trials

Publications

Supervised theses

Networks

Supervised theses

- Kidney function in paediatric patients infected by HIV. Study of new biomarkers and their possible relation to inflammatory markers of the infection. Doctoral Candidate: Angela Deyà. Director: Anton Noguera and Clàudia Fortuny. October 2015.
- Neuropsychological profile at age 6-8 years in healthy children of mothers with HIV or HCV. infection. Doctoral Candidate: Roser Colomé. Director: Clàudia Fortuny. July 2015.

Networks

- Centre for Biomedical Network Research of Epidemiology and Public Health (CIBERESP).Ministry of Economy and Competitiveness. Carlos III Health Institute.

ANNUAL SCIENTIFIC
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2015

SJD

Sant Joan de Déu
Institut de Recerca



Vertical areas

6

Emerging groups and groups under training

Foetal/Paediatric Diseases and environment

- Groups
- Projects
- Clinical Trials
- Publications
- Supervised theses
- Networks

FOETAL/PAEDIATRIC DISEASES AND ENVIRONMENT

Paediatric Intensive Care Unit research projects group

We seek to base our research projects on the crossover concept of the critical patient. The most interesting research lines to date have been in the field of respiratory and infectious pathology.

- Treatment of acute respiratory failure by means of non-invasive ventilation (NIV). The use of this technique in various aetiological conditions. Predictive factors of success and failure.
- Analysis of nosocomial infection and application of corrective measures.

- Infection markers in the critical patient. Distinction of inflammatory versus infectious pathology.
- Analysis of stress markers in post-operative patients of craniostenosis and scoliosis.
- Analysis of stress in paediatrics resident physicians during their rotation in intensive care.
- Bioethical problems in intensive care.

Research team

Coordinator Francisco José Cambra
Researchers Carme Alejandre, Georgina Armero, Mónica Balaguer, Patricia Corniero, Eli Esteban, Aida Felipe, Iolanda Jordán, Lluïsa Hernández, Nuria Millán, Martí Pon, Susana Segura, Luciana Rodriguez, David Vila

Influence of the milieu on the well-being of children and adolescents

Group focused on research in the following spheres: Study of the epidemiologic and clinical factors involved in childhood accidents. Differential diagnosis of child abuse. Early detection and secondary prevention of social-healthcare problems in adolescence: drugs,

sexuality and violence. Impact of external agents (radiation, pollution, etc.) on child health. Improvement of paediatric patient care in the physician's consultation room: presence of parents, analgesia, differentiated care by age (children / adolescents...).

Research team

Coordinator Carles Luaces Cubells
Researchers Gemma Claret Teruel, Ana Isabel Curcoy Barcenilla, Marta Simó Nebot, Lidia Martínez Sánchez, Victoria Trenchs Sainz de la Maza, Eva Gargallo Burriel, Andrea Aldemira Liz, Sergi Navarro Vilarrubí

Foetal Environment and obstetric complications

We consider it necessary to make a deeper study of the condition of the foetus precisely to be able to evaluate it at any point in the course of gestation. We have methods allowing us to determine the foetus's well-being but when these methods indicate that it may be

in a risk situation, we lack precise diagnostic tests to quantify the foetal condition as would be required to make a precise diagnosis and to suitably adapt the obstetric procedure to be followed.

Research team

Coordinator Maria Dolores Gómez Roig
Researchers Laura Almeida, Isidora Andujar, Josep Maria Boguñá, Sergi Cabré, Carolina Esteve, Silvia Ferrero, Edda Marimón, Edurne Mazarico, Jaume Miñano, Elisabeth Palacios, Miriam Perez, Joan Sabriá, Maria José Tojo

Projects

- Assessment of prenatal exposure to abuse substances and environmental toxic substances in foetuses with intrauterine growth restriction and its influence on placental pathology. Hospital Sant Joan de Déu - Esplugues HSJD. October 2015 - September 2018. PI: Maria Dolores Gómez Roig.
- Combined Maternal-Foetal and Neonatal Medicine Group, Hospital Clínic de Barcelona and Hospital Sant Joan de Déu. Agaur - Agència de Gestió d'Ajuts Universitaris i de Recerca. SGR 2014. 2014SGR928. January 2014 - December 2016. PI: Maria Dolores Gómez-Roig.
- Maternal-Child Health and Development Network (SAMID). Instituto de Salud Carlos III. Enero 2013 - December 2016. PI: Maria Dolores Gómez-Roig.
- Evaluation of prenatal exposure to antidepressants and anxiolytics by their determination in alternative biological matrices. Instituto de Salud Carlos III. January 2012 - December 2015. PI: Maria Dolores Gómez-Roig.
- Low molecular weight heparin (LMWH) for prevention of complications derived from placental insufficiency in at-risk patients without thrombophilia: randomised multi-centre study. Ministerio de Sanidad y Política Social. January 2011 - December 2015. PI: Maria Dolores Gómez-Roig.

Clinical trials

- Open trial registering subjects aged 6 to 18 years with pain requiring treatment with extended-release opioid, to evaluate the safety and efficacy of tapentadol ER versus morphine ER, followed by an open extension. November 2014 -. PI: S Navarro Vilarrubi.
- Low molecular weight heparin (LMWH) for prevention of complications resulting from placental insufficiency in risk patients without thrombophilia: randomized multicenter study. January 2011 -. PI: Maria Dolores Gómez Roig.

Publications

- Angel Solà J, Sagué Bravo S, Parra Cotanda C, Trenchs Sainz de la Maza V, Luaces Cubells C. Has the presence of parents during invasive procedures in the emergency department increased in the last few years?. *An Pediatr (Barc)* 2015. 82: 6-11. FI: 0,833(Q4).
- Benito Fernández J, Luaces Cubells C, Gelabert Colomé G, Anso Borda I. Actualización del sistema de codificación diagnóstica de la Sociedad Española de Urgencias de Pediatría. *An Pediatr (Barc)* 2015. 82: 1-7. FI: 0,833(Q4).
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- Gómez-Roig MD, Mazarico E, Valladares E, Guirado L, Fernández-Arias M, Vela A. Aortic Intima-Media Thickness and Aortic Diameter in Small for Gestational Age and Growth Restricted Fetuses. *PLoS One* 2015. FI: 3,234(Q1).
- Gómez-Roig MD, Mazarico E, Sabria J, Parra J, Oton L, Vela A. Use of Placental Growth Factor and Uterine Artery Doppler Pulsatility Index in Pregnancies Involving Intrauterine Fetal Growth Restriction or Preeclampsia to Predict Perinatal Outcomes. *Gynecol Obstet Invest* 2015. 80: 99-105. FI: 1,696(Q3).
- Hernández S, Trenchs Sainz de la Maza V, Alarcón Gamarra M, Camacho Díaz JA, Gene A, Luaces Cubells C. Etiología y curso clínico de la infección del tracto urinario en los lactantes menores de 3 meses. *Enferm Infecc Microbiol Clin* 2015. 33: 516-520. FI: 2,172(Q3).
- Hernández S, Trenchs Sainz de la Maza V, Esquivel Ojeda JN, Gene A, Luaces Cubells C. Factores predictores de contaminación ante un hemocultivo con crecimiento bacteriano en Urgencias. *An Pediatr (Barc)* 2015. 82: 426-432. FI: 0,833(Q4).
- Hernández S, Trenchs V, Batlle A, Gene A, Luaces Cubells C. Occult bacteraemia is uncommon in febrile infants who appear well, and close clinical follow-up is more appropriate than blood tests. *Acta Paediatr* 2015. 104: 76-81. FI: 1,674(Q2).
- Martínez Monseny A, Martínez Sánchez L, Margarit Soler A, Trenchs Sainz de la Maza V, Luaces Cubells C. Poisonous plants: An ongoing problem. *An Pediatr (Barc)* 2015. 82: 347-353. FI: 0,833(Q4).
- Mazarico E, Gómez-Roig MD, Guirado L, Lorente N, Gonzalez-Bosquet E. Relationship between smoking, HPV infection, and risk of cervical cancer. *Eur. J. Gynaecol. Oncol.* 2015. 36: 677-680. FI: 0,611(Q4).
- Pérez-Cruz M, Cruz-Lemini M, Fernández MT, Parra JA, Bartrons J, Gómez-Roig MD, Crispi F, Gratacós E. Fetal cardiac function in late-onset intrauterine growth restriction vs small-for-gestational age, as defined by estimated fetal weight, cerebroplacental ratio and uterine artery Doppler. *Ultrasound Obstet Gynecol* 2015. 46: 465-471. FI: 3,853(Q1).
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- Vilà de Muga M, Serrano Llop A, Rifé Escudero E, Jabalera Contreras M, Luaces Cubells C. Impacto de un modelo estandarizado para la declaración y análisis de incidentes en la mejora de un servicio de Urgencias pediátrico. *An Pediatr (Barc)* 2015. 83: 248-256. FI: 0,833(Q4).

Supervised theses

- Improved quality of care for intoxicated paediatric patients by means of quality indicators. Doctoral Candidate: Lidia Martínez Sánchez. Director: Santiago Nogué and Carles Luaces. November 2015.
- Magnesium sulphate in continuous versus discontinuous regime in the framework of watchful waiting in severe pre-eclampsia: randomised clinical trial. Ministerio de Sanidad y Política Social. January 2012 - December 2015. PI: Joan Sabrià.
- Development of a prospective quality control method for quantitative sonographic markers included in the screening of aneuploidies. Doctoral Candidate: Joan Sabrià. Director: Dolores Gómez-Roig. July 2015.

Networks

- Maternal-Child Health and Development Network (SAMiD). Carlos III Health Institute.



Horizontal areas

7

Biomedical engineering
(CREB)

Groups
Projects
Publications
Supervised theses
Networks

Bioinformatics and data analysis platform

It is considered essential to propose a platform for the analysis of data in the broadest sense, including bioinformatics and biosignals. The goal is to establish a bioinformatics platform for the IR-SJD with

a twofold aim: to provide the set of research groups with support on massive data analysis and to carry out an active programme of research, providing added value to the platform.

Research team

Coordinator: Alexandre Perera i Lluna
Researchers: Sergi Picart, Mònica Rojas, Sergio Romero, Alexander Vallmitjana, Montserrat Vallverdú, Manel González

Medical imaging, 3D printing and serious games for diagnosis and rehabilitation

Support for the rapid prototyping of serious games for the performance of short-term experimental studies, the development of a web platform of speech rehabilitation, and support for the development of a series of serious games targeted at the

assessment of eating habits and to the acquisition of knowledge for patients with childhood diabetes. Provision of a service for reconstruction of 3D models based on medical imaging and rapid prototyping with 3D printers.

Research team

Coordinator: Daniela Tost Pardell
Researchers: Dolors Ayala, Núria Bonet, Jose Luis Eguía, Robert Joan, Núria Pla, Lluís Solano, Marc Vigo, Ariel Von Barnekow

Biomaterials and tissue engineering

Support for the development of controlled drug-release systems (antineoplastics and antibiotics), for the functionalisation of surfaces and specifically metal surfaces intended for implants,

and for the development of atmospheric plasma treatments which allow cancer cells to be selectively destroyed without harm to surrounding tissues.

Research team

Coordinator Maria Pau Ginebra Molins
Researchers Noelia Aparicio, Marc Avilés, Albert Barba, Judit Buxadera, Cristina Canal, Anna Díez, Montserrat Español, Roberta Fraioli, Francesc Xavier Gil, Jordi Guillem, Mireia Hoyos, Kanupriya Khurana, Cédric Labay, Jose Maria Manero, Carles Mas, Meritxell Molmeneu, Mònica Ortiz, Marta Pegueroles, Miquel Punset, Daniel Rodriguez, Elisa Rupérez, Joanna Maria Sadowska, Romain Hegues Marie Schieber, Sergi Torrent, Èlia Vidal, Zhitong Zhao

Processing and interpretation of biomedical signals

Analysis and interpretation of signals of cerebral origin obtained by means of non-invasive techniques such as electroencephalography (EEG) and magnetoencephalography (MEG). Analysis of signals of muscular origin as a tool for motor diagnosis and rehabilitation.

Modelling and simulation of the respiratory system in order to predict the ventilatory response to different respiratory stimuli and in various conditions which may be the result of pulmonary disorders.

Research team

Coordinator Miguel Angel Mañanas
Researchers Pere Caminals, Alexandre Perera, Joan Francesc Alonso, Raul Benítez, Jesús Escrivá, Giovana Elizabeth Gavidia, Pedro Gomis, Maria Maqueda, Xavier Marimon, Sergi Picart, Mónica Rojas, Sergio Romero, Alexander Vallmitjana, Montserrat Vallverdú, Manel González

Horizontal areas

7

AREA

Biomedical
engineering
(CREB)

Groups

Projects

Publications

Supervised theses

Patents

Robotics and Vision

Guidance on process automation, adaptation and robotisation of facilities, and on different types of facilities, development of vision systems for inspection and guiding of robots, development of image processing systems, design of mobile robots (intelligent

walkers, prams, ...), and study and design of technical aids for persons with physical and sensory disabilities, as well as guidance on and development of advanced control systems.

Research team

Coordinator Antonio Benito Martínez
Researchers Joan Aranda, Antonio Benedico, Josep Fernández, Manel Frigola,
Enric Xavier Martín, Carlos Morata, Laureano Tinoco

Medical radiophysics

Trials and calibration of photon radiation and beta radiation measuring systems, guidance on aspects relating to dosimetric applications of radiations, development of methodologies for the optimisation of the exposure of healthcare personnel when

they take part in radiology and interventionist cardiology tasks, estimation of radiation dose in computed tomography, radiology and paediatric cardiology.

Research team

Coordinator Merce Ginjaume
Researchers Maria Amor Duch, Youri Alexandrovich Koubychine, Xavier Ortega, Josep Sempau

Horizontal areas

7

AREA

Biomedical
engineering
(CREB)

Groups

Projects
Publications
Supervised theses
Patents

Biomechanics

Kinematic and dynamic measurements of human movement in the biomechanics laboratory, dynamic analysis of human movement, mechanical design of care and rehabilitation devices.

Research team

Coordinator Josep Maria Font
Researchers Ana Barjau, Daniel Clos, Rosa Pàmies, Gil Serrancol

Biomaterials

Surface, mechanical, porous, biological and structural characterisation of materials by means of various tools, including scanning electron microscopy, optical interferometry, chemical analyses, in-vitro tests, cell lines, etc.

Research team

Coordinator Francesc Xavier Gil
Researchers Maria Pau Ginebra, Noelia Aparicio, Marc Avilés, Albert Barba, Judit Buxadera, Cristina Canal, Anna Díez, Montserrat Español, Roberta Fraioli, Jordi Guillem, Mireia Hoyos, Kanupriya Khurana, Cédric Labay, Jose Maria Manero, Carles Mas, Meritxell Molmeneu, Mònica Ortiz, Marta Pegueroles, Miquel Punset, Daniel Rodríguez, Elisa Rupérez, Joanna Maria Sadowska, Romain Hegues Marie Schieber, Sergi Torrent, Èlia Vidal, Zhitong Zhao

Computer graphic

Guidance on computer graphics projects in the biomedical field; development of projects on modelling, reconstruction, display of medical data, and planning of computer-assisted procedures and surgery; design of interactive virtual environments.

Research team

Coordinator Pere Caminals
Researchers Alexandre Perera, Joan Francesc Alonso, Raul Benitez, Jesús Escrivá, Giovana Elizabeth Gavidia, Pedro Gomis, Miquel A. Mañanas, Maria Maqueda, Xavier Marimon, Sergi Picart, Mónica Rojas, Sergio Romero, Alexander Vallmitjana, Montserrat Vallverdú

Instrumentation and bioengineering

Design and characterisation of medical and biotechnological instrumentation, of ultrasonic transducers, and of control and monitoring equipment; redesign of equipment and facilities.

Guidance on the effect of non-ionising radiations and on compliance with EMC and electrical safety standards and directives.

Research team

Coordinator Xavier Rosell
Researchers Ramon Bragós, Mireya Fernández, Miguel J García, Miguel Angel Garcia, Tomás Garcia, Lexa Nescolarde, Juan Ramon, Pere Joan Riu

Projects

- Development and improvement of algorithms and techniques of computational drug design, using artificial intelligence techniques. AGAUR. July 2015 - July 2018. PI: Alexandre Perera.
- Impact of training of elite athletes on heart function and neural regulation. MINECO. January 2015 - December 2017. PI: Alexandre Perera.
- Advanced osteoinductive and antimicrobial coatings to engineer biomaterials for improving osteointegration in osteoporotic and diabetic pathologies. MICINN. January 2015 - December 2017. PI: Jose Maria Manero Planella.
- Design of methods for assessing neurological and neuromuscular deterioration processes associated with ageing. MINECO. January 2015 - December 2017. PI: Miguel Angel Mañanas.
- Cost effective self-management of urinary incontinence addressed to women. Commission of European Communities. January 2015 - June 2018. PI: Miguel Angel Mañanas.
- Strengthening of a technological platform for specialised training in the health area and the development of biomedical technology. Corporación Ruta N Medellín. September 2014 - November 2016. PI: Miguel Angel Mañanas.
- SeniorLudens. INDRA Sistemas S.A. April 2014 - September 2016. PI: Daniela Tost Pardell.
- Integrated system for continuous monitoring of patients in the home and intra-hospital environments and of mobility for new healthcare and market models. Corporación Ruta N Medellín. March 2014- March 2015. PI: Miguel Angel Mañanas.
- Bioinformatics and biomedical signals laboratory. AGAUR. January 2014 - December 2016. PI: Alexandre Perera.
- Serious Games on Heart Failure patients. Estimation of their benefits on the Spanish Health System. MINECO. January 2014 - October 2015. PI: Alexandre Perera and Pere Caminal.
- Medical radiophysics and dosimetry. AGAUR. January 2014 - December 2016. PI: Mercè Ginjaume Egido, Josep Sempau Roma.
- Electronic and biomedical instrumentation. AGAUR. January 2014 - December 2016. PI: Xavier Rosell.
- Pore4Bone: Biomimetic calcium phosphates: tailoring porosity from the nano- to the macroscale for osteoinduction, drug delivery and bone tissue engineering. MINECO. January 2013 - December 2016. PI: Maria Pau Ginebra.
- Treatment of osteonecrosis of the femoral head with advanced cell therapy and biomaterials on an ovine experimental model. La Fundació La Marató de TV3. January 2013 - December 2016. PI: Maria Pau Ginebra.
- Biomimetic calcium phosphates: modulating porosity from nano- to macro-scale. MINECO. January 2013 - December 2015. PI: Maria Pau Ginebra.
- Serious games for heart surgery training. MICINN. January 2012 - March 2015. PI: Daniela Tost Pardell.
- NOVEL BIOMIMETIC STRATEGY FOR BONE REGENERATION. Commission of European Communities. November 2011 - October 2015. PI: Maria Pau Ginebra.
- RUTA N grant, National Royalties Fund 2012-2014 financing: Development of technological tools for specialised training in the health area. Corporación Ruta N Medellín. December 2013 - December 2015. PI: Miguel Angel Mañanas.
- New biofunctionalised metallic biomaterials for orthopaedic applications. MINECO. January 2013 - December 2015. PI: Francesc Xavier Gil.
- Novel biofunctionalized metallic biomaterials for orthopaedic, dental and cardiovascular Applications. Ministerio de Ciencia e Innovación. January 2013 - December 2015. PI: Francesc Xavier Gil and Jose Maria Manero Planella.
- Cardiovascular Research Network. Instituto de Salud Carlos III. January 2013 - December 2016. PI: Xavier Rosell.
- Development of methodologies for estimation of the dose to the crystalline lens. Operational implications of the application of the new dose limit. Consejo de seguridad nuclear. November 2012 - November 2015. PI: Mercè Ginjaume Egido.
- DEVELOPMENT OF NEW BIOFUNCTIONALIZED MATERIALS FOR APPLICATION IN REGENERATIVE MEDICINE. Commission of European Communities. November 2012. October 2015. PI: Francesc Xavier Gil.

- ASSAM - ASSISTANTS FOR SAFE MOBILITY. MIN industria, energía y turismo. June 2012 - June 2015. PI: Antonio Benito Martínez Velasco.
- Integrated prevention and Detection sOlutioNs Tailored to the population and Risk Factors associated with FALLs. Commission of European Communities. April 2012 - March 2015. PI: Antonio Benito Martínez Velasco.
- Fall Detector for the Elder. Commission of European Communities. March 2012 - February 2015. PI: Antonio Benito Martínez Velasco.

- Study and development of learning techniques in robotic environments with shared control. MICINN. January 2012 - June 2015. PI: Antonio Benito Martínez Velasco.
- Multi-channel non-invasive sensing and analysis systems for clinical monitoring and rehabilitation. MICINN. January 2012 - December 2015. PI: Miguel Angel Mañanas.

Publications

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schizophrenia: An adaptive sLORETA P300 localization approach. *Schizophr Res.* 2015; 169: 318-325. IF: 4.453(Q1).

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Horizontal areas

7

AREA

Biomedical engineering (CREB)

Groups

Projects

Publications

Supervised theses

Patents

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Supervised theses

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- Effect of Doping Ions and Organic Molecules on the Precipitation and Biological Interactions of Nanostructured Calcium Phosphates. Universitat Politècnica de Catalunya. Doctorand: Zhitong Zhao. Director: Maria Pau Ginebra Molins and Montserrat Español Pons. 2015.
- Contributions to new challenges in radiotherapy. From small field dosimetry to stereotactic body radiation therapy. Universitat Politècnica de Catalunya. Doctorand: Josep Puxeu Vaqué. Director: Mercè Ginjaume Egido i Maria Amor Duch Guillen. 2015.

- Optimization and muscle synergy approaches for studying muscle redundancy during walking. Universitat Politècnica de Catalunya. Doctorand: Gil Serracolí Masferre. Director: Josep Maria Font Llagunes and Benjamin J. Fregly. 2015.
- Comparative study of cyclic fatigue, resistance to torque and microfiltration, at in vitro level, between two connections: internal conical vs. external hexagonal in Klockner implants. Universitat Politècnica de Catalunya. Doctorand: David Martin Gili. Director: Francesc Xavier Gil and Lluís Giner Tarrida. 2015.

Patents

- Portable device and method for measuring electromyographic signals of a user. Patent number (application date). EP15380033.9 (2015).
- Device for simulating natural orifice endoscopic surgery. Patent number (application date). P201531796 (2015).



Horizontal areas

8

Genetic, molecular biology and gene therapy (IBUB)

- Molecular biology and gene regulation of adipose tissue and its pathologies
- Molecular pharmacology and experimental therapies
- Human molecular genetics
- Pharmacological targets in inflammation and metabolic diseases
- Intestinal microbiota

Genetic, molecular biology and gene therapy (IBUB)

Molecular biology and gene regulation of adipose tissue and its pathologies

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Molecular biology and gene regulation of adipose tissue and its pathologies

Identification of the systemic and intracellular factors controlling energy metabolism and determination of how its changes cause metabolic disorders both in early stages of life and as a long-term effect. Establishment of the mechanisms and factors by which brown adipose tissue is of key importance in neonatal and childhood energy metabolism. Establishment of the endocrine

control mechanisms of accumulation and distribution of adipose tissue in the body. Establishment of the mechanisms by which changes in energy metabolism cause heart disorders, and of the endocrine factors involved and their potential as therapeutic targets.

Research team

Coordinator Francesc Villarroya
Researchers Marta Giralt, Anna Planavila, Roser Iglesias, Teresa Mampel, Aleix Galvaldà, Joan Villarroya, J. Miguel Gallego, Marion Peyrou, Ibon Redondo, Rubén Cerejio, Ricardo Moure, Tania Quesada, Laura Campderrós, Montserrat Cairó, Laia Cervantes, Isis Navarro, Albert Però

Projects

- Pathophysiology of obesity and nutrition. Spanish Ministry of Health and Consumption. 2006 - 2016. PI: Francesc Villarroya Gombau.
- Development of novel treatment strategies based on knowledge of cellular dysfunction in diabetes (BetaBat). European Union. 2011 - 2015. PI: Francesc Villarroya Gombau.
- Development of biotechnological products to increase glycogenesis in ruminants. Consejo Nacional de Ciencia y Tecnología de México (CONACYT). 2011 - 2016. PI: Francesc Villarroya Gombau.
- Identification of novel endocrine factors secreted by brown fat (batokines) with anti-diabetic action. European Foundation for the Study of Diabetes (EFSD). 2013 - 2015. PI: Francesc Villarroya Gombau.
- Lipotoxicity, liver steatosis and hepatocellular carcinoma (HCC): the function of the Armc10/Armcx family of mitochondrial proteins. Fundació La Marató de TV3. 2014 - 2016. PI: Francesc Villarroya Gombau.
- Genetics and molecular biology of mitochondrial proteins and associated pathologies. Agència de Gestió d'Ajuts Universitaris i de Recerca (AGAUR). 2014 - 2016. PI: Francesc Villarroya Gombau.
- Grant to promote and consolidate existing research of excellence in the public universities of Catalonia. Programa ICREA Academia 2014. Fundació Institució Catalana de Recerca i Estudis Avançats (ICREA). 2015 - 2019. PI: Francesc Villarroya Gombau.
- Collection of preliminary data, design of a research programme and preparation of a project on autophagy in brown adipose tissue in collaboration with the Albert Einstein College of Medicine. Universitat de Barcelona. 2014 - 2015. PI: Francesc Villarroya Gombau.
- FGF21, a new bioactive factor in breast milk. Instituto Danone. 2015 - 2016. PI: Francesc Villarroya Gombau.
- Involvement of the FGF21 system in cardiopathy associated with obesity. Fundación Banco Bilbao Vizcaya Argentaria (BBVA). 2015 - 2017. PI: Francesc Villarroya Gombau.
- Study of gene expression in the colon, small intestine and liver

Genetic, molecular biology and gene therapy (IBUB)

Molecular biology and gene regulation of adipose tissue and its pathologies

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Intestinal microbiota

in experimental studies. LUCTA, S.A. 2014 - 2019. PI: Francesc Villarroya Gombau

- Supplementary study of gene expression in the liver and intestine in experimental protocols carried out on *Sus scrofa*. LUCTA, S.A.. 2014 - 2019. PI: Francesc Villarroya Gombau
- Identification and characterisation of new factors with systemic and local action responsible for the beneficial metabolic effects of brown adipose tissue activation. Ministerio de Economía y Competitividad. 2015 - 2017. PI: Francesc Villarroya Gombau
- Study of gene expression in visceral and subcutaneous adipose tissues, muscle and small intestine, and circulating hormonal and metabolic parameters in experimental studies. LUCTA, S.A.. 2015 - 2016. PI: Francesc Villarroya Gombau
- Network of research on adipose tissue plasticity and its pathophysiological impact. Ministerio de Economía y Competitividad. 2015 - 2017. PI: Francesc Villarroya Gombau
- Course on "Emerging issues in diabetes/obesity". Lilly Portugal, Pharmaceutical Products. Lda. 2015. PI: Francesc Villarroya Gombau
- Identification and characterisation of new cardiomyokines: a new approach for the treatment and diagnosis of heart disease. MICINN - Ministerio de Economía y Competitividad. 2015 -2018. IP: Ana Planavila Porta
- Molecular and cellular bases of changes in the FGF23/alfa-Klotho system in relation to bone, adipose and metabolic dysfunctions in HIV patients. Ministerio de Economía y Competitividad. 2015 - 2017. IP: Marta Giralt Oms

Publications

- Cereijo R, Gallego-Escuredo JM, Moure R, Villarroya J, Domingo JC, Fontdevila J, Martinez E, Gutierrez MD, Mateo MG, Giralt M. The Molecular Signature of HIV-1-Associated Lipomatosis Reveals Differential Involvement of Brown and Beige/Brite Adipocyte Cell Lineages. *PLOS ONE*. 2015; 10(8). IF: 3,234(Q1)
- Cereijo R, Giralt M, Villarroya F. Thermogenic brown and beige/brite adipogenesis in humans. *Ann Med*. 2015; 47(2): 169-177 IF: 3,886(Q1)
- Chung SY, Kao CH, Villarroya F, Chang HY, Chang HC, Hsiao SP, Liou GG, Chen SL. Bhlhe40 Represses PGC-1 alpha Activity on Metabolic Gene Promoters in Myogenic Cells. *Mol Cell Biol*. 2015;

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- Domingo P, Lamarca MK, Gallego-Escuredo JM, Torres F, Domingo JC, Villarroya J, Gutierrez MD, Mateo MG, Vidal F, Villarroya F, Giralt M. Circulating fibroblast growth factor 23 (FGF23) levels are associated with metabolic disturbances and fat distribution but not cardiovascular risk in HIV-infected patients. *J Antimicrob Chemother*. 2015; 70(6): 1825-1832 IF: 5,313(Q1)
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- Gavalda-Navarro A, Hondares E, Giralt M, Mampel T, Iglesias R, Villarroya F. Fibroblast growth factor 21 in breast milk controls neonatal intestine function. *Sci Rep*. 2015;5. IF: 5,578(Q1)
- Ghinis-Hozumi Y, Gonzalez-Davalos L, Antaramian A, Villarroya F, Pina E, Shimada A, Varela-Echavarría A, Mora O. Effect of resveratrol and lipoic acid on sirtuin-regulated expression of metabolic genes in bovine liver and muscle slice cultures. *J Anim Sci*. 2015; 93(8): 3820-3831. IF: 2,108(Q1)
- Giralt M, Gavalda-Navarro A, Villarroya F. Fibroblast growth factor-21, energy balance and obesity. *Mol Cell Endocrinol*. 2015; 418: 66-73. IF: 4,405(Q1)
- Malandrino MI, Fucho R, Weber M, Calderon-Dominguez M, Mir JF, Valcarcel L, Escote X, Gomez-Serrano M, Peral B, Salvado L, Fernandez-Veledo S, Casals N, Vazquez-Carrera M, Villarroya F, Vendrell JJ, Serra D, Herrero L. Enhanced fatty acid oxidation in adipocytes and macrophages reduces lipid-induced triglyceride accumulation and inflammation. *Am J Physiol Endocrinol Metab*. 2015; 308(9): E756-E769. IF: 3,785(Q1)
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Horizontal areas

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AREA

Genetic, molecular biology and gene therapy (IBUB)

Molecular biology and gene regulation of adipose tissue and its pathologies

Molecular pharmacology and experimental therapies
Human molecular genetics
Pharmacological targets in inflammation and metabolic diseases
Intestinal microbiota

- Sanchez-Infantes D, Gallego-Escuredo JM, Diaz M, Aragonés G, Sebastiani G, Lopez-Bermejo A, de Zegher F, Domingo P, Villarroya F, Ibanez L. Circulating FGF19 and FGF21 surge in early infancy from infra- to supra-adult concentrations. *Int J Obes.* 2015; 39(5): 742-746 IF: 5(Q1)
- Schneider JL, Villarroya J, Diaz-Carretero A, Patel B, Urbanska AM, Thi MM, Villarroya F, Santambrogio L, Cuervo AM. Loss of hepatic chaperone-mediated autophagy accelerates proteostasis failure in aging. *Aging Cell.* 2015; 14(2): 249-264. IF: 6,34(Q1)
- Sheedfar F, Vermeer M, Paziienza V, Villarroya J, Rappa F, Cappello F, Mazzocchi G, Villarroya F, van der Molen H, Hofker MH, Koonen DP, Vinciguerra M. Genetic ablation of macrohistone H2A1 leads to increased leanness, glucose tolerance and energy expenditure in mice fed a high-fat diet. *Int J Obes.* 2015; 39(2): 331-338. IF: 5,004(Q1)
- Sheedfar F, Vermeer M, Paziienza V, Villarroya J, Rappa F, Cappello F, Mazzocchi G, Villarroya F, van der Molen H, Hofker MH. Genetic ablation of macrohistone H2A1 leads to increased leanness, glucose tolerance and energy expenditure in mice fed a high-fat diet. *Int J Obes.* 2015; 39(2): 331-338 IF:5,004(Q1)
- Villarroya F, Giralt M. The Beneficial Effects of Brown Fat Transplantation: Further Evidence of an Endocrine Role of Brown Adipose Tissue. *Edocrinology.* 2015; 156(7): 2368-2370. IF: 4,530(Q1)
- Villarroya J, Redondo-Angulo I, Iglesias R, Giralt M, Villarroya F, Planavila A. Sirt1 mediates the effects of a short-term high-fat diet on the heart. *J Nutr Biochem.* 2015; 26(11): 1328-1337. IF: 3,794(Q1)

Presented theses

- New molecular actors in the physiology of adipose tissue and associated pathologies. Universitat de Barcelona. Doctoral candidate: Cereijo, R. 2015.

ANNUAL SCIENTIFIC
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Institut de Recerca

Genetic, molecular biology and gene therapy (IBUB)

Molecular biology and gene regulation of adipose tissue and its pathologies

Molecular pharmacology and experimental therapies

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Pharmacological targets in inflammation and metabolic diseases

Intestinal microbiota

Molecular pharmacology and experimental therapies

Study of the possible role of various membrane proteins (essentially with biological function as transporters) in the bioavailability of drugs and also in various pathologies, covering both their role

in oncogenesis and in the pathophysiology of various disorders, including the rare diseases involving changes in the metabolism of nucleotides (cytosolic and mitochondrial).

Research team

Coordinator Marçal Pastor-Anglada
Researchers Paula Fernández-Calotti, Sandra Perez-Torras, Nerea Urtasun, Albert Viel, Olga Casulleras-Gras, Liska Caviedes, Sandra Moro-Villa, Hamsa Banjer, Ingrid Iglesias-Garanto

Projects

- Function dissection of human concentrative nucleoside transporter (hCNT) proteins. MINECO, SAF2014-52062-R. 2015 - 2017. PI: Dr. Marçal Pastor Anglada.
- Role of tumour miRNome in the expression of membrane transporters involved in the development of digestive tumours. Assessment of new therapeutic strategies. Acció Estratègica Intramural CIBER EHD. 2015 - 2016. PI: Dra. Sandra Pérez-Torras.

Publications

- Arimany-Nardi C, Koepsell H, Pastor-Anglada M. Role of SLC22A1 polymorphic variants in drug disposition, therapeutic responses, and drug-drug interactions. *Pharmacogenomics J.* 2015;15(6): 473-487 IF: 4,229(Q1)
- Arimany-Nardi C, Montraveta A, Lee-Verges E, Puente XS, Koepsell H, Campo E, Colomer D, Pastor-Anglada M. Human organic cation transporter 1 (hOCT1) as a mediator of bendamustine uptake and cytotoxicity in chronic lymphocytic leukemia (CLL) cells. *Pharmacogenomics J.* 2015;15(4): 363-371 IF: 4,229(Q1)

- Claudio-Montero A, Pinilla-Macua I, Fernandez-Calotti P, Sancho-Mateo C, Lostao MP, Colomer D, Grandas A, Pastor-Anglada M. Fluorescent Nucleoside Derivatives as a Tool for the Detection of Concentrative Nucleoside Transporter Activity Using Confocal Microscopy and Flow Cytometry. *Mol Pharm.* 2015;12(6): 2158-2166 IF: 4,384(Q1)
- Minami K, Shinsato Y, Yamamoto M, Takahashi H, Zhang S, Nishizawa Y, Tabata S, Ikeda R, Kawahara K, Tsujikawa K, Chijiwa K, Yamada K, Akiyama S, Pérez-Torras S, Pastor-Anglada M, Furukawa T, Yasuo T. Ribonucleotide reductase is an effective target to overcome gemcitabine resistance in gemcitabine-resistant pancreatic cancer cells with dual resistant factors. *J Pharmacol Sci.* 2015;127(3): 319-325 IF: 2,360(Q3)
- Pastor-Anglada M, Perez-Torras S. Nucleoside transporter proteins as biomarkers of drug responsiveness and drug targets. *Front Pharmacol.* 2015;6 IF: 3,802(Q1)

Genetic, molecular biology and gene therapy (IBUB)

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Human molecular genetics

The general goals include identification of genetic variants associated with pathologies, modelling of genetic variants associated with pathologies, and functional tests on cellular and/or animal models, as well as phenotypic rescue and therapeutic

strategy tests. All of them applied to Opitz C syndrome and to susceptibility to suffer atypical femoral fractures; Sanfilippo C, Niemann-Pick C y Gaucher disease; psychiatric disorders; retina and vision pathologies.

Research team

Coordinator Roser González Duarte
 Researchers Susana Balcells, Lluïsa Vilageliu, Bru Cormand, Roser Urreizti, Noelia Fernández, Mónica Cozar, Bárbara Torrico, Jenny Serra, Marta Gómez, Neus Roca, Judit Cabana, Gemma Marfany, Daniel Gringerg

Projects

- Mastering skills in the training Network for attention deficit hyperactivity and autism spectrum Disorders (MiND). European Commission, Horizon2020 (Research and Innovation Framework Programme: Training Networks). 2015 -2018. PI: Bru Cormand. Coordinator: Barbara Franke.
- Analysis of protein trafficking in retinal neurodegeneration via high-resolution microscopy in animal models and hiPSCs. Marató de TV3 (2013). 2015 - 2017. PI: Roser González-Duarte. Pablo Loza Álvarez. Institut de Ciències Fotòniques, Castelldefels. Slaven Erceg. Centro de investigación Príncipe Felipe. Valencia.
- Genetics of pigmentary retinitis. ASOCIACIÓN RETINA ASTURIAS. 2015. PI: Roser González-Duarte
- Identification of variants, generation of models, and therapeutic tests in three types of genetic pathologies: lysosomal disorders, osteoporosis, and Opitz C syndrome. MINECO. 2015 - 2016. PI: Daniel Grinberg / Susana Balcells.
- Search for the gene responsible for Opitz C. PRECIPITA - FECYT. 2015 - 2016. PI: Daniel Grinberg / Susana Balcells.
- Therapeutic strategy of substrate reduction based on short hairpin RNAs (shRNAs) for Sanfilippo disease. Asocaiación Stop Sanfilippo. 2015 - 2016. PI: Daniel Grinberg / Lluïsa Vilageliu.
- Use of a neuronal model derived from human induced pluripotent stem cells (iPSC) to test therapeutic strategies for lysosomal disorders. MPS-España. 2015 - 2016. PI: Daniel Grinberg / Lluïsa Vilageliu.
- Variants of directional change of DKK1 gene present in the general population: what is their role in the functionality of the protein?. FEIOMM. 2015 - 2016. PI: Susana Balcells.
- OPITZ C SYNDROME AND BOHRING-OPITZ SYNDROME. BeHeard. 2015. PI: Roser Urreizti.
- Support for research groups recognised by the Government of Catalonia. 2014. 2014 -2016. PI: Daniel Grinberg.
- Identification of therapeutic targets for retinal dystrophies: functional characterisation in vivo and in vitro of genes causing hereditary blindness. SAF2013-49069-C2-1-R. MICINN Proyectos I+D+I orientada a los retos de la sociedad. 2014 - 2016. PI: Roser González-Duarte, Gemma Marfany.
- Support for research of the Human Molecular Genetics Group (2014 SGR 932). AGAUR - Generalitat de Catalunya. 2014 - 2016. PI: Daniel Grinberg.
- Identification of genetic variants associated with bone mineral density (BMD) in the FLJ2280 gene. FEIOMM. 2014 - 2015. PI: Daniel Grinberg.

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 Pharmacological targets in inflammation and metabolic diseases
 Intestinal microbiota

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- Cross-disorder analysis of psychiatric disorders: Contribution of rare and common genetic variants to autism, ADHD and substance use disorders. Ministerio de Economía y Competitividad (MINECO), Spain. 2013 - 2015. PI: Bru Cormand.
- Development of pharmacological chaperones for the treatment of rare neurometabolic disorders. MINECO. 2012 -2015. PI: Marc Martinell (Minoryx Therapeutics) - Participant: Daniel Grinberg / Lluïsa Vilageliu.
- European network and registry for homocystinurias and methylation defects. European Commission. 2012 - 2015. PI: Henk Blom - Participant: Daniel Grinberg /Susana Balcells.

Publications

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Genetic, molecular biology and gene therapy (IBUB)

Molecular biology and gene regulation of adipose tissue and its pathologies
Molecular pharmacology and experimental therapies
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- Molecular genetics of autism: identification of rare and common variants of susceptibility. Universitat de Barcelona. Doctoral candidate: Bàrbara Torrico Avilés. Directors: Bru Cormand Rifà, Claudio Toma. July 2015.
- Genes and migraine: mutational analysis and association studies in patients, and transcriptomics in an animal model. Universitat de Barcelona. Doctoral candidate: Cèlia Sintas Vives. Director: Bru Cormand Rifà. 2015.
- Genetic and molecular analysis of Sanfilippo C syndrome. Generation of a neuronal model using human induced pluripotent stem (ipS) cells and therapeutic strategies. Universitat de Barcelona. Doctoral candidate: Isaac Canals Montferrer. Directors: Daniel Grinberg / Lluïsa Vilageliu. 2015.
- Generation of an osteoblastic cell model and therapeutic approaches for Gaucher disease. Universitat de Barcelona. Doctoral candidate: Jenny Serra Vinardell. Directors: Daniel Grinberg / Lluïsa Vilageliu. 2015.

Horizontal areas

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AREA

Genetic, molecular biology and gene therapy (IBUB)

Molecular biology and gene regulation of adipose tissue and its pathologies
Molecular pharmacology and experimental therapies
Human molecular genetics
Pharmacological targets in inflammation and metabolic diseases
Intestinal microbiota

- Models and therapeutic approaches for Niemann-Pick (A/B and C) and other lysosomal storage disorders. Universitat de Barcelona. Doctoral candidate: Marta Gómez Grau. Directors: Daniel Grinberg / Lluïsa Vilageliu. 2015.

Patents

METHOD FOR THE DETERMINATION OF THE RISK OF ATYPICAL FRACTURES IN PATIENTS TREATED WITH BONE REMODELLING INHIBITORS. Patent number (application date): EP 15382064.2(2015).

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Genetic, molecular biology and gene therapy (IBUB)

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Human molecular genetics
Pharmacological targets in inflammation and metabolic diseases
Intestinal microbiota

Pharmacological targets in inflammation and metabolic diseases

The general goal is the search for new pharmacological targets which will allow the blocking of the connection between inflammation and the development of pathologies such as insulin resistance and cardiomyopathies.

Research team

Coordinator Manuel Vazquez Carrera
Researchers Xavier Palomer, Emma Barroso, Gaia Botteri, Mohammad Zarei

Projects

- Drugs for inflammatory and cardiovascular disorders. Caixa Impulso. 2015 - 2016. PI: Rosana Leiva.
- Synthesis and pharmacological assessment of new molecules with antidiabetic properties, SAFNAD. ACCIÓ. Agència de Suport a l'Empresa Catalana. 2014 - 2016. PI: Santiago Vázquez.
- Hyperinsulinaemia and inflammation as negative regulators of insulin sensitivity: study of the effects of PPARb/d and oleic acid activators in in-vitro and in-vivo models of insulin resistance. Ministerio de Economía y Competitividad. 2013 - 2015. PI: Manuel Vázquez Carrera.

Publications

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- Masana L, Cabré A, Heras M, Amigó N, Correig X, Martínez-Hervás S, Real JT, Ascaso JF, Quesada H, Julve J, Palomer X, Vázquez-Carrera M, Girona J, Plana N, Blanco-Vaca F. Remarkable quantitative and qualitative differences in HDL after niacin or fenofibrate therapy in type 2 diabetic patients. *Atherosclerosis*. 2015;238(2): 213-219 IF:3,994(Q1).

Horizontal areas

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AREA

Genetic, molecular biology and gene therapy (IBUB)

Molecular biology and gene regulation of adipose tissue and its pathologies
Molecular pharmacology and experimental therapies
Human molecular genetics
Pharmacological targets in inflammation and metabolic diseases
Intestinal microbiota

- Palomer X, Capdevila-Busquets E, Botteri G, Davidson MM, Rodríguez C, Martínez-González J, Vidal F, Barroso E, Chan TO, Feldman AM, Vázquez-Carrera M. miR-146a targets Fos expression in human cardiac cells. *Dis Model Mech.* 2015;8(9): 1081-1091 IF:4.973(Q1).
- Petrov D, Pedrós I, Artiach G, Sureda FX, Barroso E, Pallás M, Casadesús G, Beas-Zarate C, Carro E, Ferrer I, Vazquez-Carrera M, Folch J, Camins A. High-fat diet-induced deregulation of hippocampal insulin signaling and mitochondrial homeostasis deficiencies contribute to Alzheimer disease pathology in rodents. *Biochim Biophys Acta.* 2015;1852(9): 1687-1699 IF: 4,882(Q1).
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Presented theses

- Exploring Adamentane-like scaffolds for a wide range of therapeutic targets. Doctoral candidate: Elena Valverde Murillo. Directors: Santiago Vázquez Cruz and Manuel Vázquez Carrera. 30 October 2015.
- Short-term administration of GW501516 improves the inflammatory condition of white adipose tissue, liver damage and renal inflammation of mice fed with fructose-rich diet. Doctoral candidate: D'Angelo Carlo Magliano. Directors: Profa. Dra. Márcia Barbosa Águila Mandarim de Lacerda (Universidade do Estado do Rio de Janeiro) and Prof. Dr. Manuel Vázquez-Carrera. 15 August 201.

Patent

- ANALOGS OF ADAMANTYLUREAS AS SOLUBLE EPOXIDE HYDROLASE INHIBITORS. Patent number (application date): Patent: ZBM ref: P3392EP00. Inventors: Santiago Vázquez Cruz, Elena Valverde Murillo and Manuel Vázquez Carrera.

Genetic, molecular biology and gene therapy (IBUB)

Molecular biology and gene regulation of adipose tissue and its pathologies
Molecular pharmacology and experimental therapies
Human molecular genetics
Pharmacological targets in inflammation and metabolic diseases
Intestinal microbiota

Intestinal microbiota

Widening of the knowledge of the molecular mechanisms involved in the communication between the gut flora and the cells of the intestinal mucosa. Study of proteins secreted by bacteria and their

function in the interaction with the host. Study of outer membrane vesicles (OMV) of commensal microbiota and probiotics as signalers in intestinal homeostasis.

Research team

Coordinator Laura Baldomà
Researchers Josefa Badia, Rosa Giménez, Laura Aguilera, Lorena Toloza,
M^a José Fábrega, M^a Alexandra Cañas, Carina Shianya Alvarez

Projects

- We belong to the Integrative Biochemistry Consolidated Group. Agència de Gestió d'Ajuts Universitaris i de Recerca (AGAUR). 2014 -2016. PI: Marta Cascante Serratosa.
- Function of external membrane vesicles of commensal macrobiota and gram-negative probiotics in the regulation of intestinal homeostasis. Ministerio de Economía y Competitividad. 2013 - 2015. PI: Laura Baldoma Llavines.
- Developing an efficient e-infrastructure, standards and data-flow for metabolomics and its interface to biomedical and life science e-infrastructures in Europe and world-wide (COSMOS). Unió Europea. 2012 - 2015. PI: Marta Cascante Serratosa; Pedro Ramon de Atauri Carulla.

Publications

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- Albert J, D'Andrea L, Granell J, Pla-Vilanova P, Quirante J, Khosa MK, Calvis C, Messeguer R, Badia J, Baldoma L. Cyclopalladated and cycloplatinated benzophenone imines: Antitumor, antibacterial and antioxidant activities, DNA interaction and cathepsin B inhibition (vol 140, pg 80, 2014). J Inorg Biochem. 2015; 150:189-189 IF: 3.444(Q1).
- Ferreira E, Gimenez R, Cañas MA, Aguilera L, Aguilar J, Badia J, Baldoma L. Glyceraldehyde-3-phosphate dehydrogenase is required for efficient repair of cytotoxic DNA lesions in Escherichia coli. Int J Biochem Cell Biol. 2015;60:202-212 IF: 4.046(Q2).
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Horizontal areas

9

Neuropsychology, cognitive
and development neuroscience

Groups
Projects
Publications
Supervised theses

Horizontal areas

9

AREA

Neuropsychology,
cognitive and
development
neuroscience

Groups

Projects

Publications

Supervised theses

Neuropsychology

Characterisation of the neuroanatomical and neurofunctional correlates of the various disorders and of their recovery. Determination of the neurobiological effects of physical exercise and cognitive stimulation. Identification of the changes of the white and grey matter and of cerebral activation in neurodevelopmental

disorders and disorders of the neurobiological mechanisms associated with intervention strategies. Study of the changes of cerebral connectivity and neuropsychological performance after brain damage. Characterisation of brain networks in obesity and overweight.

Research team

Coordinator Maria Mataró
Researchers Maria Angeles Jurado, Jonatan Ottino, Roser Pueyo, Júlia Ballester,
Olga Laporta, Xavier Caldú

Cognitive neuroscience

Identification of specific brain mechanisms on the levels of system, brain region, circuit and neuronal population, as well as on genetic level, which contribute to the cognitive functions, such as auditory

perception, predictive perception in response to a stimulus, and cognitive processes in numerical cognition.

Research team

Coordinator Carles Escera i Mico
Researchers Maria José Corral, Iria San Miguel, Raffaele Cacciaglia, Miriam Cornella, Natàlia Gorina,
Imma Clemente, Maribel Nuñez, Marc Via, Hans Supèr, Alejandro Lerer

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Developmental neuroscience

Study of early capacity to recognise the mother tongue, of skills in discrimination of languages and/or dialectal variants, of phonetic categorisation of speech sounds (native and non-native), of

segmentation of speech, of word-meaning association (mapping and learning of words), of phonological representation of lexicon items, and of recognition of familiar words.

Research team

Coordinator Laura Bosch Galceran
Researchers Marta Ramon, Joan Birulés, Clément François, Maria Teixidó,
Jordina Solé, Ferran Pons

Behaviour modelling

Collection of quantitative descriptions of emergence and maintenance of coordinated collective motion (CCM), development of analytical techniques for detection of subgroups in CCM and applying them both to simulated data and to empirical data,

tracking of paths to make empirical descriptions of the paths of the individuals who move by means of a CCM and assessment of the adjustment between data from simulations (in silico) and data from observation (in vivo).

Research team

Coordinator Francesc Salvador I Beltran
Researchers Elisabet Gimeno, Ruth Dolado

Projects

- Exploration of the sensory patterns involved in predictive processing by means of sensory-motor associations. MINECO. 2015-2017. PI: San Miguel Insua, Iria.
- Exploring the novel role of ocular vergence in visual attention and perception. MINECO. 2015-2017. PI: Supér, Hendrik Anne.
- Multimodal information at the start of language learning: use of audiovisual keys in normal/typical development and in neurocognitive risk populations. MINECO. 2015-2017. PI: Bosch Galceran, Laura.
- Subcortical Cognition. MINECO. 2015-2016. PI: Escera, Carles.
- Phasic boosting of attentional capabilities with Forbrain®. Innovation and Technology Transfer agreement with Soundev, SA (Luxemburg). 2015-2016. PI: Escera, Carles.
- A single-case study of phasic boosting of auditory processing and attentional strengthening with Forbrain®. Innovation and Technology Transfer agreement with Soundev, SA (Luxemburg). 2015. PI: Escera, Carles.
- Mastering skills in the training Network for ADHD and autism spectrum Disorders. European Commission. 2015-2016. PI: Jan Buitelaar.
- Simple screening of cognitive development in infants by low-cost remote eye measurements. Bill Gates Foundation. 2015-2016. PI: Super, Hans.
- EEG amplifier. MINECO. 2015-2016. PI: Escera, Carles.
- Subcortical contributions to auditory cognition. MINECO. 2015-2016. PI: Escera, Carles.
- Networks of Excellence. MINECO. 2015-2018. PI: Otazy Porter, Xavier.
- Brainlab-Cognitive Neuroscience Research Group. AGAUR, Generalitat de Catalunya. 2014-2016. PI: Escera, Carles.
- Improving Cognitive Audition. University of Barcelona. 2014-2015. PI: Escera, Carles.
- Vision and Control of Action lab. AGAUR, Generalitat de Catalunya. 2014-2016. PI: López-Moliner, Joan.
- Neuroplasticity in the adult age: physical exercise and cognitive training. MINECO. 2013-2016. PI: Mataró, Maria.
- Obesity and overweight: neuronal correlates over the course of life. Characterisation of brain networks. MINECO. 2013-2016. PI: Jurado, Maria Ángeles.
- Regularity encoding and deviance detection on a distributed auditory subcortico-cortical network. MINECO. 2013-2015. PI: Escera, Carles.
- PredictiveCodingTime - Interactions between predictive coding and predictive timing in audition: characterizing the role of rhythm in repetition suppression through entrained brain oscillations. European Commission. 2013-2016. PI: Costa, Jordi.
- Executive and brain connectivity dysfunctions in dyskinetic cerebral palsy. MINECO. 2012-2015. PI: Pueyo Benito, Roser.
- Grant to promote and consolidate existing research of excellence in the public universities of Catalonia. ICREA Academia 2015 programme. ICREA. 2010-2015. PI: Escera, Carles.

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- Solé Puig M, Pérez Zapata L, Puigcerver L, Esperalba Iglesias N, Sanchez Garcia C, Romeo A, Cañete Crespillo J, Supèr H. Attention-Related Eye Vergence Measured in Children with Attention Deficit Hyperactivity Disorder. *PLoS One.* 2015; 10(12):e0145281. IF: 3.234(Q1).
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