## Vertical areas

### Area: Paedriatic neuroscience
- PKU and other intermediary metabolism defects
- Neurometabolic disorders with expression in the CNS
- Mitochondrial energy metabolism
- Applied research in neuromuscular disorders
- Movement disorders of genetic and neurometabolic origin
- Neurogenetics and molecular medicine

### Area: Molecular and cellular biology of developmental tumours
- Preclinical pharmacology and drug release in solid paediatrics tumours
- Paediatric Leukaemia
- Neuroblastoma and Personalized Medicine
- Translational genomics
- Ewing’s sarcoma

### Area: Paediatric Diseases originating in the early years of life
- Metabolic endocrinology
- Perinatal origin of adult diabetes: animal models
- Neonatal physiopathology

### Area: Infectious diseases and systemic inflammatory response in pediatrics
- Vaccine-preventable diseases
- Vertical transmitted infections and infections in vulnerable patients
- Tuberculosis
- Systemic inflammatory response in the paediatric age

### Emerging groups and groups under training
- Foetal/Paediatric Diseases and environment
  - Paediatric Intensive Care Unit research projects group
  - Influence of the milieu on the well-being of children and adolescents
  - Foetal Environment and obstetric complications

### Area: Biomedical engineering
- Bioinformatics and data analysis platform
- Medical imaging, 3D printing and serious games for diagnosis and rehabilitation
- Biomaterials and tissue engineering
- Processing and interpretation of biomedical signals
- Robotics and Vision
- Medical radiophysics
- Biomechanics
- Biomaterials
- Computer graphics
- Instrumentation and bioengineering

### Area: Genetic, molecular biology and gene therapy
- 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

### Area: Neuropsychology, cognitive and development neuroscience
- Neuropsychology
- Cognitive neuroscience
- Developmental neuroscience
- Behaviour modelling
Foreword

Introduction

Governing and advisory bodies
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

Governing council

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His Excellency Mr. Didac Ramírez i Sarrió
Ms. Esther Real Saladrigas
Dr. Jaume Pérez Payarols
Mr. Emili Bargalló Angerri
Dr. Francesc Palau Martínez

General Manager of Hospital Sant Joan de Déu in Esplugues de Llobregat
Vice-Rector for Research of the University of Barcelona
Vice-Rector for Knowledge Transfer of the Polytechnical University of Catalonia (UPC)

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Scientific deputy director of Institut de Recerca Sant Joan de Déu
University of Barcelona
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Mr. Emili Bargalló Angerri
Director of Research Foundation Sant Joan de Déu

Dr. Francesc Palau Martínez
Director of Research Committee IR-SJD

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Dr. Romà Pallarès Giner
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Genomic Regulation Centre of Barcelona
Director of the Neuropathology Institute of Bellvitge University Hospital
Dean of the Faculty of Economic and Business Sciences of Pompeu Fabra University (UPF)
Director of the Epidemiology and Cardiovascular Genetics Research Group Programme (IMIM)
Deputy Director of the General Management of the Biomedical Research Institute of Bellvitge (IDIBELL)
Agency for Health Information, Evaluation and Quality. Government of Catalonia
Director of the European Research Area (ERA) - European Commission
Professor of Psychiatry, University of Cantabria (UC)
General Director of BIOCAT

Ms. Júlia Ribot Ballabriga
Secretary

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Dr. Francesc Palau Martínez
Dr. Francesc Villarroya Gombar
Sra. Maite López Secanella, secretary
Vertical areas

2

AREA

Paedriatic neuroscience

Groups
Projects
Clinical Trials
Publications
Supervised theses
Networks
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, Silvia Meavilla, Carmen Fons, Camila García Volpe, Rose Colomer, Aida Ormazabal, Cristina Sierra
Vertical areas

2

Area

Paedriatic neuroscience

Groups
Projects
Clinical Trials
Publications
Supervised theses
Networks

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 Cortés-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 Álvarez
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 Clinic 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.

Research team

Coordinator: Dr. Janet Hoenicka
Principal Investigators: Francesc Palau, Janet Hoenicka, Judith Armstrong, Mercedes Serrano
Researchers: Laura Dominguez, Lara Cantarero, Xotchilt Castro, Loreto Martorell, Maria del Mar Pérez Iribarne, Isabel Plensa, Antonio Martinez Monseny, Nuria Brandi, Laura Blasco, Azahara Civera, Silvia Vidal
Technicians: Dolores López, Ana López, Lidia Álvarez, Paola Pacheco, Jordi Genovés, Montserrat Naudó, Daniel Castillo, Patricia Peral, Teresa Zabala, Lluisa Ramirez
Projects


- 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.


• 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: Mª Pilar Poo.


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 Sanfilippo 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: Mª 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.

- 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: Maria Julieta.


- 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 Ramirez.

- 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.

Publications


- Patterson MC, Mengel E, Vanier MT, Schwierin B, Muller A, Cornelles P, Pineda M. Stable or improved neurological manifestations during miglustat therapy in patients from the international disease registry for Niemann-Pick disease type C: an observational cohort study. Orphanet J. Rare Dis. 2015. 10: 65-0. FI: 3,358(Q2).


Supervised theses


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.
Vertical areas

Molecular and cellular biology of developmental tumours

Groups
Projects
Clinical Trials
Publications
Networks
Preclinical pharmacology and drug release in solid paediatrics tumours


Research team
Coordinator Ángel Montero-Carcaboso
Researchers Sonia Paco, Carles Monterrubio, 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 Torrebader Burriel, Anna Ruiz-Llobet, Rubén Berrueco Moreno, Albert Català Temprano, Julia Marsal Ricomà, Isabel Badell Serra, Susana Rives Solà
Technician Camino Estella Aguado
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
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: Mª Inmaculada Hernández Muñoz, Sara Sanchez Molina, Elisabet Figuerola Bou, Sara Perez Jaume, Estela Prada Varela, Veronica Celis, Moira Garraus
Technician: Mª Jesús Nagel Moyano
Projects

- 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.
- CHD5, PAFAH1B1 and NME1: Markers for prognosis and prediction of chemoresistance mechanisms. Hospital Sant Joan de Déu - Esplugues HSJD. April 2012 - March 2015. PI: Cinzia Lavarino.
- Neuroblastoma NEN. Associació de familiars i amics de pacients amb neuroblastoma. January 2012 -. PI: Cinzia Lavarino.
Clinical trials


- Placebo-controlled randomised phase III clinical trial to assess the effectiveness and safety of MK 0617/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 Berrueco.

- Open multi-centre phase 2 study to assess the pharmacokinetics, pharmacodynamics, safety and activity of 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.


- 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.


- 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 Berrueco.

- 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 Berrueco.

- 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 Berrueco.

- 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.


- Multi-centre open phase I increasing-dose study of a single group, with monitoring of efficacy, to evaluate vemurafenib...
(ro5185426) in children and adolescents with surgically incurable and unresectable melanoma in stage iiic or iv presenting BRAF V600 mutations. F. Hoffmann- La Roche Ltd; Hospital Sant Joan de Déu - Esplugues HSJD; Quintiles. February 2014 -. PI: Ofelia Cruz.


- 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.


- 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 Farmacéutica. 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 Berrueco.

- 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.


- 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.


Publications

Molecular and cellular biology of developmental tumours


Networks

- Innovative Therapies for Children with Cancer. European consortium.
Vertical areas

AREA 4

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, Silvia Ribó, Ivonne Palacios
Technician: Judith Cebrià
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: periconceptional, 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 García-Alix
Technician Thais Agut, Marta Camprubi, Isabel Iglesias, Martin Iriondo, Ana Martin, Africa Pertierra, Ana Riverola
Projects


• Study to determine the suitability of preparing a practical clinical guide on hypoxic-ischaemic encephalopathy. Agencia Información, Avaluació i Qualitat en Salut. December 2011 -. PI: Alfredo Garcia-Alix.


Clinical trials


• Treatment with recombinant growth hormone (rGH) in short patients born small-for-gestational-age (SGA) followed at Hospital Sant Joan de Déu: Analysis of early predictors of good response useful in clinical practice. W1191603. Fundación Privada per a la Recerca i la Docencia Sant Joan de Déu. November 2014 -. PI: Lourdes Ibáñez.

• Treatment with recombinant growth hormone (rGH) in patients with GH deficiency, controlled at Hospital Sant Joan de Déu: analysis of early markers of good response which are useful in clinical practice. October 2015 -. PI: Marta Ramon.

• Newborns with extremely low gestational age assigned at random, blindly, to be resuscitated with 21% vs 60% oxygen: influence on mortality and chronic disorders in the neonatal period. October 2015 -. PI: Marta Ramon.


• AC201503 - NN304-4093 26-week open randomised multi-centre trial on 2 parallel groups to investigate the effectiveness and safety of insulin detemir versus NPH insulin (neutral protamin Hagedorn insulin) in combination with metformin and diet/exercise. November 2015 -. PI: Marta Ramon-Krauel.

Vertical areas

Paediatric Diseases originating in early life

Groups
Projects
Clinical Trials
Publications
Supervised theses
Networks

Publications


Martín-Ancel A, Balaguer A, García-Alix A. Duration of resuscitation...


Supervised theses


Networks

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

5

AREA

Infectious diseases and systemic inflammatory response in pediatrics

Groups
Projects
Clinical Trials
Publications
Supervised theses
Networks
Infectious diseases and systemic inflammatory response in pediatrics

Groups
Projects
Clinical Trials
Publications
Supervised theses
Networks

Vertical areas

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
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 Vincent Genestar, Eneritz Velasco Arnaitz, Maria Goretti López Ramos, Aina Martinez Planas, Eva Maria Guix Cormellas, 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, Estíbaliz Iglesias, Judith Sánchez, Joan Calzada, Violeta Bitterman, Angela Deyà, Mónica Piquer, Anna Esteve-Solé
Projects


- 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.


• Study of the primary and secondary defects of the IL12-IFNg-TNFα pathway with susceptibility to intracellular microorganism infection. Hospital Sant Joan de Déu Barcelona. March 2014 - February 2017. PI: Laia Alsina.


• 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.


• 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. PICT14/00228. January 2014 - December 2016. PI: Antoni Noguera.


• Use of dried-blood spot (DBS) samples for detection and molecular surveillance of Streptococcus pneumoniae in developing countries. Hospital Sant Joan de Déu - Esplugues HJD. April 2012 - March 2015. PI: Carmen Muñoz-Almagro.


Clinical trials


- 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.


- 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 Mª 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 Mª 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.


- 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 Guash.

- 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: Mª Asunción Vicente Villa.


- 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.


• 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 Mª 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 II, multi-centre, open study on a single group 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.

• Prospective, multi-centre, single-arm study to evaluate the efficacy, safety and pharmacokinetics of denosumab in children with osteogenesis imperfecta. October 2014 -. PI: Rosa Bo.

• 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 Mª 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 Farmacéutica. 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 Mª 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.


• 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.
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. 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.


- 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.
Publications


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 Claudia 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: Claudia Fortuny. July 2015.

Networks

• Centre for Biomedical Network Research of Epidemiology and Public Health (CIBERESP). Ministry of Economy and Competitiveness. Carlos III Health Institute.
Emerging groups and groups under training

Foetal/Paediatric Diseases and environment

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


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 Vilarrubi

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 Boguñà, Sergi Cabrè, Carolina Esteve, Silvia Ferrero, Edda Marimón, Edurne Mazarico, Jaume Miñano, Elisabeth Palacios, Miriam Perez, Joan Sabrià, Maria José Tojo
Projects


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 Vilarubi.

- 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, Sagüé 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).


Supervised theses


Networks

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, Mónica Rojas, Sergio Romero, Alexander Vallmitjana, Montserrat Vallverdu, 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 Eguia, 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 Diez,Montserrat Español, Roberta Fraioli, Francesc Xavier Gil, Jordi Guillem, Mireia Hoyos, Kanupriya Khurana, Cédric Labay, Jose Maria Manero, Carles Mas, Meritxell Molmeneu, Mónica Ortiz, Marta Pegueroles, Miquel Punset, Daniel Rodríguez, Elisa Rupérez, Joanna Maria Sadowska, Romain Hegues Marie Schieber, Sergi Torrent, Elia 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.

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

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.
Robots 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
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 Diez, Montserrat Español, Roberta Fraioli, Jordi Guillem, Mireia Hoyos, Kanupriya Khurana, Cédric Labay, Jose Maria Manero, Carles Mas, Meritxell Molmeneu, Mònica Ortiz, Marta Pegueroles, Miquel Pusset, Daniel Rodríguez, Elisa Rupérez, Joanna Maria Sadowska, Romain Hegues Marie Schieber, Sergi Torrent, Elia 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.

Research team

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

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


- 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.


- SeniorLudens. INDRA Sistemas S.A. April 2014 - September 2016. PI: Daniela Tost Pardell.


- 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.


- 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.


- Serious games for heart surgery training. MICINN. January 2012 - March 2015. PI: Daniela Tost Pardell.


- 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.


Publications


• Nucl Eng Des/Fusion. 2015; 98-99: 2206-2209. IF: 1.301(Q1).
Supervised theses


Patents


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
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 Cereijo, Ricardo Moure, Tania Quesada, Laura Campderrós, Montserrat Cairó, Laia Cervantes, Isis Navarro, Albert Però

Projects


• 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.


• Study of gene expression in the colon, small intestine and liver
• 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

Publications

• Díaz M, Gallego-Escured JM, de Zegher F, Villarroya F, Ibáñez L. Effects of ethinylestradiol-cyproterone acetate vs. pioglitazone-flutamide-metformin on plasma FGF21 levels in adolescent girls with androgen excess. Diabetes Metab. 2015; IF: 3.267(Q2)

Horizontal areas

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
Horizontal areas

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

Presented theses

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

Projects


Publications


• 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)
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 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, Bàrbara Torrico, Jenny Serra, Marta Gómez, Neus Roca, Judit Cabana, Gemma Marfany, Daniel Grinberg

Projects


• Genetics of pigmentary retinitis. ASOCIACIÓN RETINA ASTURIAS. 2015. PI: Roser Gonzàlez-Duarte


• 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. Asociación 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 / Lluisa 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.


Publications


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


Presented theses

Horizontal areas
8

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


Patents

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 Impulse. 2015 - 2016. PI: Rosana Leiva.

Publications

• Magliano DC, Penna-de-Carvalho A, Vazquez-Carrera M, Mandarim-de-Lacerda CA, Aguilera MB. Short-term administration of GW501516 improves inflammatory state in white adipose tissue and liver damage in high-fructose-fed mice through modulation of the renin-angiotensin system. Endocrine. 2015;50(2): 365-367 IF: 3.878(Q2).


• Rodriguez-Calvo R, Vázquez-Carrera M, Masana L, Neumann D. AICAR Protects against High Palmitate/High Insulin-Induced Intramyocellular Lipid Accumulation and Insulin Resistance in HL-1 Cardiac Cells by Inducing PPAR-Target Gene Expression. PPAR Res. 2015 IF: 2,509(Q2).


Presented theses


• 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

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, Mª José Fábrega, Mª Alexandra Cañas, Carina Shianya Alvarez

Projects


Publications


Horizontal areas

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 Cornellà, Natàlia Gorina, Imma Clemente, Maribel Nuñez, Marc Via, Hans Supèr, Alejandro Lerer
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

- 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.
- EEG amplifier. MINECO. 2015-2016. PI: Escera, Carles.
- Subcortical contributions to auditory cognition. MINECO. 2015-2016. PI: Escera, Carles.
- 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 Angeles.
- 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.
Publications


- do Vale S, Selinger L, Martins JM, Bicho M, do Carmo L, Escera C. Dehydroepiandrosterone (DHEA) and dehydroepiandrosterone-sulfate (DHEAS) and emotional processing - A behavioral and electrophysiological approach. Horm Behav. 2015; 73:94-103 IF: 4.632(Q1).


- Igualada A, Bosch L, Prieto P. Language development at 18 months is related to multimodal communicative strategies at 12 months. Infant Dev Behav. 2015; 39:42-52. IF: 1.349(Q3).


Supervised theses


