UB research on rare diseases brochure

We are pleased to present the brochure of rare diseases research of the University of Barcelona (UB) and associated centers, developed by the Bosch i Gimpera Foundation. In the following pages you will find the capabilities of more than 40 research groups classified by diseases according to the International Classification of Diseases (ICD) and also referred to the Orphanet’s Orpha number.

Summary:

2 Infectious and parasitic diseases
4 Neoplasms and oncological diseases
6 Diseases of the blood and disorders of the immune system
8 Endocrine, nutritional and metabolic diseases
12 Behavioral disorders and diseases of the nervous system
17 Diseases of the eye and ear and craniofacial malformations
20 Diseases of the respiratory system and the digestive system
21 Diseases of the musculoskeletal system and connective tissue
23 Congenital malformations and chromosomal abnormalities
25 Transversals

The Bosch i Gimpera Foundation is the Knowledge Transfer Office (KTO) of the University of Barcelona (UB)
Infectious and parasitic diseases

RESEARCH GROUPS

COMPUTATIONAL BIOLOGY AND DRUG DESIGN (School of Pharmacy)
IP: F. Javier Luque Garriga
Diseases: Tuberculosis (ORPHA3389) (A15-A19)
Research Line: Discovery of bioactive compounds through a computational and experimental own methodology.

<table>
<thead>
<tr>
<th>Capacities</th>
<th>MECHANISM AND TREATMENT DISCOVERY</th>
<th>DIAGNOSTIC BIOMARKERS</th>
<th>IN VITRO MODELS</th>
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<th>CLINICAL DATA</th>
<th>OTHERS</th>
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<tbody>
<tr>
<td>Description</td>
<td>Selection and synthesis of active compounds.</td>
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MULTIVARIANT AND COMPUTATIONAL STATISTICS (School of Biology)
IP: Antonio Monleón Getino
Diseases: Tuberculosis (ORPHA3389) (A15-A19)
Research Line: Study of tuberculosis incidence and trends.

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<tbody>
<tr>
<td>Description</td>
<td>Statistics, population analysis.</td>
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Publications: [www.ncbi.nlm.nih.gov/pubmed/?term=Monleon-Getino%5BAuthor%5D](www.ncbi.nlm.nih.gov/pubmed/?term=Monleon-Getino%5BAuthor%5D)

INFECTION PATHOLOGY AND ANTIBIOTIC SENSITIVITY (School of Medicine)
Infectious Diseases Unit (Hospital of Bellvitge)
IP: Miguel Ángel Santín Cerezales
Diseases: Tuberculosis (ORPHA3389) (A15-A19)
Research Line: Strategies to take therapeutic decisions during the following of tuberculosis cases.

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<tr>
<td>Description</td>
<td>Standard strategy based on different diagnostic methods.</td>
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Publications: [www.ncbi.nlm.nih.gov/pubmed/?term=Santin+M%5BAuthor%5D](www.ncbi.nlm.nih.gov/pubmed/?term=Santin+M%5BAuthor%5D)
Infectious and parasitic diseases

APPLICATION OF NEW TOOLS OF DIAGNOSIS, MOLECULAR CLASSIFICATION AND DRUG ASSAY FOR LEISHMANIA SPP AND TRYpanosoma CRUZI (School of Pharmacy)
Laboratory of Parasitology
IP: Roser Fisa and M. Cristina Riera.
Diseases: Leishmaniasis (ORPHA507) (B55); Chagas disease (ORPHA3386) (B57)
Research Line: Diagnosis, molecular classification and drug assay for leishmania spp and trypanosoma cruzi.

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<td>Diagnosis, molecular classification and drug assay.</td>
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<td>Nanotechnology for drug formulations.</td>
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<td>Patient data access. Parasitic diagnosis methods.</td>
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Publications: [www.ncbi.nlm.nih.gov/pubmed/?term=Fisa+R%5BAuthor%5D+OR+Riera+C%5BAuthor%5D](www.ncbi.nlm.nih.gov/pubmed/?term=Fisa+R%5BAuthor%5D+OR+Riera+C%5BAuthor%5D)

BENZNIDAZOL AND TRIAZOL RESEARCH GROUP FOR NANOMEDICINE AND INNOVATION ON CHAGAS DISEASE (School of Medicine)
IP: Teresa Vinuesa Aumedes
Diseases: Chagas disease (ORPHA3386) (B57)

<table>
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<td></td>
<td>Standardization of T. cruzi parasite cultures. Set up of cardiac and glial primary cultures. Toxicity and efficacy of the currently used drugs and comparison to optimized nanostructured agents.</td>
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<td>Parasite culture and maintenance of T. cruzi strains.</td>
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<td>New lipid-based nanoformulations.</td>
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Publications: [www.ncbi.nlm.nih.gov/pubmed/?term=Vinuesa+T%5BAuthor%5D](www.ncbi.nlm.nih.gov/pubmed/?term=Vinuesa+T%5BAuthor%5D)
# RARE AND ORPHAN DISEASES

**Neoplasms and oncological diseases**

**CLINICAL AND BIOLOGICAL UNIT FOR THE STUDY OF LYMPHOMAS AND LYMPHOPROLIFERATIVES SYNDROMES** (School of Medicine).

**IP:** Emilio Montserrat Costa

**Diseases:** Follicular lymphoma (ORPHA545) (C82); Diffuse large B-cell lymphoma (ORPHA544) (C83.3); MALT lymphoma (ORPHA52417) (C88.4)

**Research Line:** lymphomas and chronic lymphoid leukemias

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<td>Mechanism and treatment discovery</td>
<td>Founder member of the International Workshop on Chronic Lymphocytic Leukemia and the European Task Force for Lymphomas.</td>
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**Publications:** [www.ncbi.nlm.nih.gov/pubmed/?term=Montserrat+E%5BAuthor%5D](www.ncbi.nlm.nih.gov/pubmed/?term=Montserrat+E%5BAuthor%5D)

**MODELING OF BIOLOGICAL SYSTEMS AND DRUG DESIGN** (IQTC, School of Chemistry)

**IP:** Jaime Rubio Martínez

**Diseases:** Burkitt lymphoma (ORPHA543) (C83.7)

**Research Line:** Drug design by molecular modeling.

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<tr>
<td>Mechanism and treatment discovery</td>
<td>Molecular modeling</td>
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**Publications:** [www.ncbi.nlm.nih.gov/pubmed/?term=Rubio+JE%5BAuthor%5D](www.ncbi.nlm.nih.gov/pubmed/?term=Rubio+JE%5BAuthor%5D)

**TRANSITIONAL IMMUNOLOGY IN DERMATOLOGY, ALLERGIES AND CHRONIC INFLAMMATION** (School of Biology)

**IP:** Luis Francisco Santamaria Babí

**Diseases:** Primary cutaneous T-cell lymphoma (CTCL) (ORPHA178533) (C84.4)

**Research Line:** Translational immunology

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<tr>
<td>Mechanism and treatment discovery</td>
<td>Development and study of a CTCL animal model.</td>
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**External collaborations**

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<tbody>
<tr>
<td>Mechanism and treatment discovery</td>
<td>Access to clinical data and samples.</td>
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**Publications:** [www.ncbi.nlm.nih.gov/pubmed/?term=Santamaria-Bab%C3%AD+LF%5BAuthor%5D](www.ncbi.nlm.nih.gov/pubmed/?term=Santamaria-Bab%C3%AD+LF%5BAuthor%5D)
Neoplasms and oncological diseases

MOLECULAR PHARMACOLOGY AND EXPERIMENTAL THERAPEUTICS (IBUB, School of Biology)

IP: Marçal Pastor Anglada

Diseases: Rare leukemias as Mantle cell lymphoma (ORPHA52416) (C83.1); Chronic lymphocytic leukemias (ORPHA67038) (C91); Acute lymphoblastic leukemia (ORPHA513) (C91.0).


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- Antitumor drug transporters and transporter pharmacogenetics
- Molecular analysis.
- Pancreatic xenografts.
- Access to clinical data and samples.

Publications: [www.ncbi.nlm.nih.gov/pubmed/?term=Pastor-Anglada+M%5BAuthor%5D](www.ncbi.nlm.nih.gov/pubmed/?term=Pastor-Anglada+M%5BAuthor%5D)
# RARE AND ORPHAN DISEASES

Diseases of the blood and disorders of the immune system

**SYSTEMIC AUTOIMMUNE DISEASES** (School of Medicine)
- **IP**: Ricard Cervera Segura
- **Diseases**: Sarcoidosis (ORPHA797) (D86); Systemic vasculitis (ORPHA52759) (M05.2); Autoinflammatory syndrome (ORPHA93665); Systemic lupus erythematosus (ORPHA536) (M32); Systemic sclerosis (ORPHA90291) (M34); Sjögren’s syndrome (ORPHA378) (M35.0).
- **Research Line**: Autoimmune diseases.

<table>
<thead>
<tr>
<th>Capacities</th>
<th>MECHANISM AND TREATMENT DISCOVERY</th>
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<td>OTHERS</td>
<td>Research on systemic autoimmune diseases. Giant cell arteritis registry.</td>
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**MOLECULAR PHYSIOLOGY** (School of Biology)
- **IP**: Antonio Felipe Campo
- **Diseases**: Long QT syndrome (ORPHA768) (I45.8); Brugada syndrome (ORPHA130) (I47.2)

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<tr>
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<td>CLINICAL DATA</td>
<td>OTHERS</td>
<td>Pharmacogenomic high-throughput screening. Ion channels analysis.</td>
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</table>

**Publications**: [www.ncbi.nlm.nih.gov/pubmed/?term=Felipe+Antonio%5BAuthor%5D](www.ncbi.nlm.nih.gov/pubmed/?term=Felipe+Antonio%5BAuthor%5D)

**HEPATIC HEMODYNAMICS UNIT** (School of Medicine)
- **IP**: Joan Carles García Pagan
- **Diseases**: Portal vein thrombosis, non neoplasic and non cirrotic (ORPHA854) (I81); Budd-Chiari syndrome (ORPHA131) (I82.0); Hepatoportal sclerosis (ORPHA4743) (Q27).
- **Research Line**: Vascular hepatic diseases.

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<td>CLINICAL DATA</td>
<td>OTHERS</td>
<td>Clinical data registries.</td>
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**Publications**: [www.ncbi.nlm.nih.gov/pubmed/?term=Garc%C3%ADa-Pag%C3%A1n+JC%5BAuthor%5D](www.ncbi.nlm.nih.gov/pubmed/?term=Garc%C3%ADa-Pag%C3%A1n+JC%5BAuthor%5D)
Diseases of the blood and disorders of the immune system

UNIT OF CLINICAL PHARMACY AND PHARMACOTHERAPY (School of Pharmacy)
IP: Eduardo L. Mariño Hernández
Diseases: Paroxysmal nocturnal hemoglobinuria (ORPHA447) (D59.5); Hereditary angioedema (ORPHA91378) (D84.1)

MOLECULAR PHARMACOLOGY AND EXPERIMENTAL THERAPEUTICS (IBUB, School of Biology)
IP: Marçal Pastor Anglada
Diseases: SLC29A3 disorders as H syndrome (ORPHA168569) (D76); Pigmented hypertrichosis with insulin dependent diabetes mellitus syndrome (ORPHA254723) (D76); Faisalabad histiocytosis (ORPHA254707) (D76.3)
Research Line: Nucleosides transporters and nucleotides metabolism.

RARE ANEMIAS GROUP (School of Medicine)
Red Cell Pathology Unit (Hospital Clínic). Official center for the diagnosis of Sickle cell.
IP: Juan Luis Vives Corrons
Diseases: Pyruvate kinase deficiency of erythrocytes (ORPHA766) (D55.2); Thalassemies (D56); Sickle cell (ORPHA251359) (D57.2); Hemoglobinopathy (ORPHA68364) (D58.2); Anemia related diseases (D60-64); Rare constitutional anemia (ORPHA183651) (D61.0)
Research Line: Molecular basis of hematology

Publications:
- www.ncbi.nlm.nih.gov/pubmed/?term=Marino+EL%5BAuthor%5D
- www.ncbi.nlm.nih.gov/pubmed/?term=Pastor-Anglada+M%5BAuthor%5D
- www.ncbi.nlm.nih.gov/pubmed/?term=Vives-Corrons+JL%5BAuthor%5D
ADULT RARE DISEASES GROUP (School of Medicine)
IP: Francesc Cardellach López
Diseases: Congenital metabolic diseases (E70-90) and Mitochondrial disorders.
Research Line: Diagnosis tests.

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Physiopathological research studies.

- Access to pediatric clinical data.
- Biochemical diagnosis method.
- Mitochondrial analysis.

Publications: [PubMed Search](https://www.ncbi.nlm.nih.gov/pubmed/?term=Cardellach+F%5BAuthor%5D)

MEMBRANE PROTEINS BIOGENESIS (School of Biology)
IP: Josep Chillarón Chaves
Diseases: Cystinuria (ORPHA214) (E72.0)

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Protein misfolding associated to disease mutations.

Publications: [PubMed Search](https://www.ncbi.nlm.nih.gov/pubmed/?term=Chillar%C3%B3n+J%5BAuthor%5D)

RESEARCH UNIT ON BIOACTIVE MOLECULES (RUBAM) (School of Pharmacy)
IP: Antonio Delgado Cirilo
Diseases: Farber Disease (ORPHA333) (E75.2) and Gaucher Disease (ORPHA355) (E75.2)
Research Line: Design and synthesis of pharmacological chaperones.

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Drug discovery

- Biochemical assays and screening of new compounds.
- Mutation analysis and inhibitors evaluation.

Publications: [PubMed Search](https://www.ncbi.nlm.nih.gov/pubmed/?term=Delgado+A%5BAuthor%5D)
RARE AND ORPHAN DISEASES
Endocrine, nutritional and metabolic diseases

TRANSPORT DISEASES PHYSIOLOGY (School of Medicine)
IP: Raúl Estévez Povedano
Diseases: Bartter syndrome (ORPHA112) (E26.8)
Research Line: Molecular bases and therapy discovery.

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- Biochemical assays and screening of new compounds.
- Mutation analysis and inhibitors evaluation.

Publications: [www.ncbi.nlm.nih.gov/pubmed/?term=Est%C3%A9vez+R%5BAuthor%5D](www.ncbi.nlm.nih.gov/pubmed/?term=Est%C3%A9vez+R%5BAuthor%5D)

NEUROPHYSIOLOGY LAB (School of Medicine)
IP: Xavier Gasull Casanova
Diseases: Megalencephalic leukoencephalopathy with subcortical cysts (ORPHA2478) (E75.2); Trigeminal neuralgia (ORPHA221091) (G50.0); Peripheral neuropathy (ORPHA98496) (G90.0); Glaucomas (ORPHA359) (H40); Sjögren’s syndrome (ORPHA378) (M35.0)
Research Line: Gene and protein therapies to hereditary retinal dystrophies: a neuroprotective approach.

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- Sensorial neuron alterations. Receptors and ion channels implicated in the diseases.
- Identification of diseases-related ion channels alterations.

Publications: [www.ncbi.nlm.nih.gov/pubmed/?term=Gasull+X%5BAuthor%5D](www.ncbi.nlm.nih.gov/pubmed/?term=Gasull+X%5BAuthor%5D)

HUMAN MOLECULAR GENETICS (School of Biology)
IP: Daniel Grinberg Vaisman
Diseases: MTHFR deficiency (ORPHA395) (E72.1); Classical homocystinuria (ORPHA394) (E72.1); GM1 gangliosidosis (ORPHA354) (E75.1); Gaucher disease (ORPHA355) (E75.2); Krabbe disease (ORPHA487) (E75.2); Niemann-Pick disease type A, b and C (ORPHA77292) (E75.2); Maroteaux-Lamy disease (ORPHA583) (E76.2); Sanfilippo syndrome type A and B (ORPHA79269/ORPHA79270) (E76.2).
Research Line: Genetic basis and functional studies on neurologic channel diseases.

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- Genetic basis and functional studies. Genetic diagnosis.
- Design and synthesis of active compounds and chaperones.
- Computational biology.
- Access to clinical data.

Publications: [www.ncbi.nlm.nih.gov/pubmed/?term=Grinberg+D%5BAuthor%5D](www.ncbi.nlm.nih.gov/pubmed/?term=Grinberg+D%5BAuthor%5D)
RARE AND ORPHAN DISEASES
Endocrine, nutritional and metabolic diseases

BIOSYNER (School of Pharmacy)
IP: Rodolfo Lavilla Grifols
Diseases: Neurometabolic diseases: congenital glicosilation defects. (ORPHA68385) (M32)
Research Line: Precompetitive ChemBioMedical Knowledge on Rare Diseases and its Application to Drug Repurposing and Orphan Drug Design.

<table>
<thead>
<tr>
<th>Capacities</th>
<th>MECHANISM AND TREATMENT DISCOVERY</th>
<th>DIAGNOSTIC BIOMARKERS</th>
<th>IN VITRO MODELS</th>
<th>IN VIVO MODELS</th>
<th>CLINICAL DATA</th>
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<td>External collaborations</td>
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<td>DIAGNOSTIC BIOMARKERS</td>
<td>IN VITRO MODELS</td>
<td>IN VIVO MODELS</td>
<td>CLINICAL DATA</td>
<td>OTHERS: Chemical synthesis of molecular chaperones.</td>
</tr>
</tbody>
</table>

Publications: [www.ncbi.nlm.nih.gov/pubmed/?term=Lavilla+R%5BAuthor%5D](www.ncbi.nlm.nih.gov/pubmed/?term=Lavilla+R%5BAuthor%5D)

UNIT OF CLINICAL PHARMACY AND PHARMACOTHERAPY (School of Pharmacy)
IP: Eduardo L. Mariño Hernández
Diseases: Fabry disease (ORPHA324) (E75.2); Mucopolysaccharidosis (ORPHA79213) (E77.8)

<table>
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<th>Capacities</th>
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<th>DIAGNOSTIC BIOMARKERS</th>
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<th>OTHERS: Equity access to orphan drugs.</th>
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<td>DIAGNOSTIC BIOMARKERS</td>
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<td>IN VIVO MODELS</td>
<td>CLINICAL DATA</td>
<td>OTHERS: New gens description. RMN studies.</td>
</tr>
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</table>

Publications: [www.ncbi.nlm.nih.gov/pubmed/?term=Marino+EL%5BAuthor%5D](www.ncbi.nlm.nih.gov/pubmed/?term=Marino+EL%5BAuthor%5D)

HUMAN MOLECULAR GENETICS GROUP (GGMH) (School of Medicine)
IP: Virginia Nunes Martínez
Diseases: Cystinuria (ORPHA214) (E72.0); Lysinuric protein intolerance (ORPHA470) (E72.0); Megalencephalic leukoencephalopathy with subcortical cysts (ORPHA2478) (E75.2)

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Publications: [www.ncbi.nlm.nih.gov/pubmed/?term=Nunes+V%5BAuthor%5D](www.ncbi.nlm.nih.gov/pubmed/?term=Nunes+V%5BAuthor%5D)
RARE AND ORPHAN DISEASES
Endocrine, nutritional and metabolic diseases

GENETICS AND MOLECULAR BIOLOGY OF MITOCHONDRIAL PROTEINS AND RELATED PATHOLOGIES (IBUB, School of Biology)
IP: Francesc Villarroya Gombau
Diseases: Congenital lipodystrophies (ORPHA528) (E88.1)
Research Line: Molecular bases and potential therapeutic targets of this group of rare diseases.

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Molecular bases and potential therapeutic targets.

- Access to clinical data and samples.
- Molecular characterization.

Publications: [www.ncbi.nlm.nih.gov/pubmed/?term=Villarroya+F%5BAuthor%5D](www.ncbi.nlm.nih.gov/pubmed/?term=Villarroya+F%5BAuthor%5D)

COMPREHENSIVE RESEARCH IN EFFECTIVE THERAPIES FOR THE TREATMENT OF CYSTIC FIBROSIS AND RELATED DISEASES (School of Medicine)
IP: Miguel Viñas Ciordia
Diseases: Cystic fibrosis (ORPHA568) (E84)

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Susceptibility of *Pseudomonas aeruginosa* from CF patients to the treatment with nanoencapsulated antibiotics.

- Gene therapy.
- Nanoparticles production.

Publications: [www.ncbi.nlm.nih.gov/pubmed?term=Vi%C3%B1as%20M%5BAuthor%5D&cauthor=true&cauthor_uid=22702530](www.ncbi.nlm.nih.gov/pubmed?term=Vi%C3%B1as%20M%5BAuthor%5D&cauthor=true&cauthor_uid=22702530)
### RARE AND ORPHAN DISEASES

**Behavioral disorders and diseases of the nervous system**

**PHYSIOPATHOLOGY OF NEURODEGENERATIVE DISEASES** (IDIBAPS, School of Medicine)
- **IP:** Jordi Alberch
- **Diseases:** Huntington chorea (ORPHA399) (G10)
- **Research Line:** Neuroprotection (Jordi Alberch); Cell therapy (Josep M. Canals); New pharmacological targets (Silvia Ginés); Altered molecular mechanisms (Esther Pérez-Navarro).

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<td>Different approaches to HD physiopathology and therapy.</td>
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<td>International collaborators for multiple experimental lines.</td>
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**Publications:** [http://www.ncbi.nlm.nih.gov/pubmed/?term=Alberch+J%5BAuthor%5D](http://www.ncbi.nlm.nih.gov/pubmed/?term=Alberch+J%5BAuthor%5D)

**NEURAL DEVELOPMENT RESEARCH GROUP** (School of Medicine)
- **IP:** Soledad Alcantara Horrillo
- **Diseases:** Autism (ORPHA106) (F84.0); Rett Syndrome (ORPHA778) (F84.2)
- **Research Line:** Potential implication of new BDNF signaling effector gens.

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<tr>
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<td>Genetic, molecular and cellular studies.</td>
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<td>Access to pediatric clinical data.</td>
<td>Biomarker studies.</td>
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**Publications:** [http://www.ncbi.nlm.nih.gov/pubmed/?term=Alc%C3%A1ntara+S%5BAuthor%5D](http://www.ncbi.nlm.nih.gov/pubmed/?term=Alc%C3%A1ntara+S%5BAuthor%5D)

**MOLECULAR GENETICS GROUP** (School of Medicine)
- **IP:** Michael John Edel
- **Diseases:** Rett syndrome (ORPHA778) (F84.2); Spinal muscular atrophy (ORPHA139557) (G12.2); Spinal cord injury (ORPHA90058) (T09.3) and Mitochondrial respiratory chain disorder (ORPHA309136).
- **Research Line:** Induced pluripotent stem cell technology (iPS cells) as a model to study rare diseases. New protocols to make clinical grade muscle stem cells and motor neuron stem cells.

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<td><strong>Description</strong></td>
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<td>In collaboration with the Cell therapy program of the University of Barcelona.</td>
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**Publications:** [www.ncbi.nlm.nih.gov/pubmed/?term=Edel+MJ%5BAuthor%5D](http://www.ncbi.nlm.nih.gov/pubmed/?term=Edel+MJ%5BAuthor%5D)
RARE AND ORPHAN DISEASES
Behavioral disorders and diseases of the nervous system

SYNTHESIS OF COMPOUNDS WITH POTENTIAL BIOLOGICAL ACTIVITY (School of Pharmacy)
IP: María Carmen Escolan Mirón
Diseases: De Vivo disease (ORPHA71277) (G93.4).
Research Line: Therapeutic potential of triheptanoin anaplerotic diet in a mouse model of de vivo's disease (Glut-1 deficiency syndrome).

<table>
<thead>
<tr>
<th>Capacities</th>
<th>Design and synthesis of compounds.</th>
</tr>
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<td>MECHANISM AND TREATMENT DISCOVERY</td>
<td>DIAGNOSTIC BIOMARKERS</td>
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<tr>
<td>OTHERS</td>
<td>In vivo studies.</td>
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<td>Formulation of compounds for in vivo administration.</td>
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Publications: [www.ncbi.nlm.nih.gov/pubmed/?term=Escolano+C%5BAuthor%5D](www.ncbi.nlm.nih.gov/pubmed/?term=Escolano+C%5BAuthor%5D)

NUCLEAR RECEPTORS INTERACTION SURFACES AS NOVEL THERAPEUTIC TARGETS (School of Biology)
IP: Eva Estébanez Perpiñá
Diseases: Kennedy's Disease (ORPHA481) (G12.2).
Research Line: Identification of the androgen receptor aggregates causing Spinal and Bulbar Muscular Atrophy (SBMA, Kennedy’s disease)

<table>
<thead>
<tr>
<th>Capacities</th>
<th>Research on structure and function of human nuclear receptors using X-ray crystallography and other structure-function approaches.</th>
</tr>
</thead>
<tbody>
<tr>
<td>MECHANISM AND TREATMENT DISCOVERY</td>
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<td>OTHERS</td>
<td>Biochemical assays and screening of new compounds.</td>
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<td>Mutation analysis and inhibitors evaluation.</td>
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Publications: [www.ub.edu/ibub/eva_estebanez.html](www.ub.edu/ibub/eva_estebanez.html)

TRANSPORT DISEASES PHYSIOLOGY (School of Medicine)
IP: Raúl Estévez Povedano
Diseases: Megalencephalic leukoencephalopathy with subcortical cysts (ORPHA2478) (E75.2); Congenital myotonia (ORPHA206973) (G71.1)
Research Line: Molecular bases and therapy discovery.

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<tr>
<td>MECHANISM AND TREATMENT DISCOVERY</td>
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Publications: [www.ncbi.nlm.nih.gov/pubmed/?term=Est%C3%A9vez+R%5BAuthor%5D](www.ncbi.nlm.nih.gov/pubmed/?term=Est%C3%A9vez+R%5BAuthor%5D)
RARE AND ORPHAN DISEASES
Behavioral disorders and diseases of the nervous system

MOLECULAR PHYSIOLOGY (School of Biology)
IP: Antonio Felipe Campo
Diseases: Multiple sclerosis (ORPHA802) (G35); Dravet Syndrome (ORPHA33069) (G40.4)

<table>
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<tr>
<th>Capacities</th>
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<td>Mechanism and treatment discovery</td>
<td>Diagnosis biomarkers</td>
<td>In vitro models</td>
<td>In vivo models</td>
<td>Clinical data</td>
<td>Others</td>
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</table>

Description:
Pharmacogenomic high-throughput screening. Ion channels analysis.

External collaborations
MECHANISM AND TREATMENT DISCOVERY
DIAGNOSTIC BIOMARKERS
IN VITRO MODELS
IN VIVO MODELS
CLINICAL DATA
OTHERS

* Access to clinical data


NEUROPHYSIOLOGY LAB (School of Medicine)
IP: Xavier Gasull Casanova
Diseases: Trigeminal neuralgia (ORPHA221091) (G50.0) and Peripheral neuropathy (ORPHA98496) (G90.0)
Research Line: Gene and protein therapies to hereditary retinal dystrophies: a neuroprotective approach.

<table>
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</table>

Description:
Sensorial neuron alterations. Receptors and ion channels implicated in the diseases.

External collaborations
MECHANISM AND TREATMENT DISCOVERY
DIAGNOSTIC BIOMARKERS
IN VITRO MODELS
IN VIVO MODELS
CLINICAL DATA
OTHERS

* Identification of diseases-related ion channels alterations.

Publications: [www.ncbi.nlm.nih.gov/pubmed/?term=Gasull+X%5BAuthor%5D](http://www.ncbi.nlm.nih.gov/pubmed/?term=Gasull+X%5BAuthor%5D)

HUMAN MOLECULAR GENETICS (School of Biology)
IP: Daniel Grinberg Vaisman
Diseases: Familial paroxysmal ataxia (ORPHA97) (G11.8); Tyrosine hydroxylase deficiency (ORPHA101150) (G24.1); Familial or sporadic hemiplegic migraine (ORPHA569) (G43.1).
Research Line: Genetic basis and functional studies on neurologic channel diseases: migraine and ataxia.

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<tr>
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</table>

Description:
Genetic basis and functional studies. Genetic diagnosis.

External collaborations
MECHANISM AND TREATMENT DISCOVERY
DIAGNOSTIC BIOMARKERS
IN VITRO MODELS
IN VIVO MODELS
CLINICAL DATA
OTHERS

* Design and synthesis of active compounds and chaperones.
* Computational biology.
* Access to clinical data.

Publications: [www.ncbi.nlm.nih.gov/pubmed/?term=Grinberg+D%5BAuthor%5D](http://www.ncbi.nlm.nih.gov/pubmed/?term=Grinberg+D%5BAuthor%5D)
RARE AND ORPHAN DISEASES

Behavioral disorders and diseases of the nervous system

**METABOLIC ENGINEERING AND DIABETES** (IRB, School of Biology)
IP: Joan J. Guinovart Cirera
Diseases: Lafora disease (ORPHA501) (G40.3).
Research Line: Glycogen metabolism and its dysfunctions in Lafora disease.

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Description: Genetic, molecular and cellular analyses.

Publications: [www.ncbi.nlm.nih.gov/pubmed/?term=Guinovart+JJ%5BAuthor%5D](http://www.ncbi.nlm.nih.gov/pubmed/?term=Guinovart+JJ%5BAuthor%5D)

**NEUROTOXICITY OF SENSORIAL AND MOTOR SYSTEMS** (School of Medicine)
IP: Jordi Llorens Baucells
Diseases: Amyotrophic lateral sclerosis (ORPHA803) (G12.2)
Research Line: Neurotoxicology.

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Description: President 2011-2013, International Neurotoxicology Association (ina14.org)

Publications: [www.ncbi.nlm.nih.gov/pubmed/?term=Llorens+J%5BAuthor%5D](http://www.ncbi.nlm.nih.gov/pubmed/?term=Llorens+J%5BAuthor%5D)

**VOLUME VISUALIZATION AND ARTIFICIAL INTELLIGENCE, WAI** (School of Mathematics)
IP: Anna Puig Puig
Diseases: Arnold Chiari malformation type 1 (ORPHA268882) (G95.0); Idiopathic Syringomyelia (ORPHA99858) (G95.0)
Research Line: Identification and classification of cerebral malformations presents in the disease by artificial intelligence. The medical imaging technology provided by the group is also applicable to identification and diagnosis of several conditions coursing with malformations.

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</table>

Description: 3D Modeling and biomechanical simulation of malformations

Publications: [www.maia.ub.es/~anna/home.html](http://www.maia.ub.es/~anna/home.html)
FUNCTIONAL CHARACTERIZATION OF HERC PROTEINS AND THEIR IMPLICATIONS IN NEURODEGENERATION AND CANCER (School of Medicine)
IP: Jose Luis Rosa López
Diseases: Cerebellar ataxia (ORPHA4145) (G11).
Research Line: Integrating genetic, molecular and cellular analyses to better understand the mechanisms altered and therapeutic opportunities.

<table>
<thead>
<tr>
<th>Capacities</th>
<th>MECHANISM AND TREATMENT DISCOVERY</th>
<th>DIAGNOSTIC BIOMARKERS</th>
<th>IN VITRO MODELS</th>
<th>IN VIVO MODELS</th>
<th>CLINICAL DATA</th>
<th>OTHERS</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>External collaborations</td>
<td>MECHANISM AND TREATMENT DISCOVERY</td>
<td>DIAGNOSTIC BIOMARKERS</td>
<td>IN VITRO MODELS</td>
<td>IN VIVO MODELS</td>
<td>CLINICAL DATA</td>
<td>OTHERS</td>
<td>• Multinational collaboration on genetic, cellular, molecular and pharmacologic analyses.</td>
</tr>
</tbody>
</table>


MOLECULAR PATHOLOGY AND THERAPY FOR HETEROGENETIC AND POLIGENETIC DISEASES (IRB, School of Biology)
IP: Antonio Zorzano
Diseases: Charcot-Marie-Tooth disease (ORPHA166) (G60.0).

<table>
<thead>
<tr>
<th>Capacities</th>
<th>MECHANISM AND TREATMENT DISCOVERY</th>
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<td>External collaborations</td>
<td>MECHANISM AND TREATMENT DISCOVERY</td>
<td>DIAGNOSTIC BIOMARKERS</td>
<td>IN VITRO MODELS</td>
<td>IN VIVO MODELS</td>
<td>CLINICAL DATA</td>
<td>OTHERS</td>
<td>• Access to clinical data.</td>
</tr>
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</table>

Publications: [www.ncbi.nlm.nih.gov/pubmed/?term=Zorzano+A%5BAuthor%5D](http://www.ncbi.nlm.nih.gov/pubmed/?term=Zorzano+A%5BAuthor%5D)
## RARE AND ORPHAN DISEASES

### Diseases of the eye and ear and craniofacial malformations

#### ADULT RARE DISEASES GROUP (School of Medicine)
**IP:** Francesc Cardellach López  
**Diseases:** Kearns-Sayre syndrome (ORPHA480) (H49.8)  
**Research Line:** Molecular and biochemical diagnosis for Kearns-Sayre syndrome by MT-ATP8 gene and eight more.

<table>
<thead>
<tr>
<th>Capacities</th>
<th>MECHANISM AND TREATMENT DISCOVERY</th>
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<td>CLINICAL DATA</td>
<td>OTHERS</td>
</tr>
</tbody>
</table>

- Access to pediatric clinical data.
- Biochemical diagnosis method.
- Mitochondrial analysis.

**Publications:** [www.ncbi.nlm.nih.gov/pubmed/?term=Cardellach+F%5BAuthor%5D](https://www.ncbi.nlm.nih.gov/pubmed/?term=Cardellach+F%5BAuthor%5D)

#### NEUROPHYSIOLOGY LAB (School of Medicine)
**IP:** Xavier Gasull Casanova  
**Diseases:** Glaucomas (ORPHA359) (H40)  
**Research Line:** Gene and protein therapies to hereditary retinal dystrophies: a neuroprotective approach.

<table>
<thead>
<tr>
<th>Capacities</th>
<th>MECHANISM AND TREATMENT DISCOVERY</th>
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</table>

- Identification of diseases-related ion channels alterations.

**Publications:** [www.ncbi.nlm.nih.gov/pubmed/?term=Gasull+X%5BAuthor%5D](https://www.ncbi.nlm.nih.gov/pubmed/?term=Gasull+X%5BAuthor%5D)

#### HUMAN MOLECULAR GENETICS (IBUB, School of Biology)
**IP:** Roser González Duarte  
**Diseases:** Hereditary retinal dystrophies (H35.5): Retinitis pigmentosa (ORPHA791); Cone rod dystrophy (ORPHA791).  
**Research Line:** Development of DNA chips for the diagnosis of hereditary retinal dystrophies. Genomic approaches to identify new genes implicated in retinal dystrophies. Generation and study of new animal models of Retinitis pigmentosa and Cone rod dystrophy.

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<td>CLINICAL DATA</td>
<td>OTHERS</td>
</tr>
</tbody>
</table>

- Identification and functional validation of new gens.
- Generation and study of animal models
- Retina Lipidomics.

**Publications:** [www.ncbi.nlm.nih.gov/pubmed/?term=Gonz%C3%A0lez-Duarte+R%5BAuthor%5D](https://www.ncbi.nlm.nih.gov/pubmed/?term=Gonz%C3%A0lez-Duarte+R%5BAuthor%5D)

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## RARE AND ORPHAN DISEASES

### Diseases of the eye and ear and craniofacial malformations

**HUMAN MOLECULAR GENETICS** (School of Biology)

**IP:** Daniel Grinberg Vaisman

**Diseases:** Opitz-Caltabiano syndrome (ORPHA1786) (Q75.4); Multiple osteochondromas (ORPHA321) (Q78.6)


**External collaborations**

**Capacities**

<table>
<thead>
<tr>
<th>MECHANISM AND TREATMENT DISCOVERY</th>
<th>DIAGNOSTIC BIOMARKERS</th>
<th>IN VITRO MODELS</th>
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<th>CLINICAL DATA</th>
<th>OTHERS</th>
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</thead>
<tbody>
<tr>
<td>Genetic basis and functional studies. Genetic diagnosis.</td>
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</tbody>
</table>

**Description**

- Design and synthesis of active compounds and chaperones.
- Computational biology.
- Access to clinical data.

**Publications:** [www.ncbi.nlm.nih.gov/pubmed/?term=Grinberg+D%5BAuthor%5D](www.ncbi.nlm.nih.gov/pubmed/?term=Grinberg+D%5BAuthor%5D)

**NEUROTOXICITY OF SENSORIAL AND MOTOR SYSTEMS** (School of Medicine)

**IP:** Jordi Llorens Baucells

**Diseases:** Meniere disease (ORPHA45360) (H81.0); Audio-vestibular degeneration (H81); Vestibular pathology (H81).

**Research Line:** Neurotoxicology.

**External collaborations**

**Capacities**

<table>
<thead>
<tr>
<th>MECHANISM AND TREATMENT DISCOVERY</th>
<th>DIAGNOSTIC BIOMARKERS</th>
<th>IN VITRO MODELS</th>
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<th>CLINICAL DATA</th>
<th>OTHERS</th>
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</thead>
<tbody>
<tr>
<td>President 2011-2013, International Neurotoxicology Association (ina14.org)</td>
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</table>

**Description**

- Vestibular physiopathology knowledge.
- In vitro models.
- Neuromuscular models.

**Publications:** [www.ncbi.nlm.nih.gov/pubmed/?term=Llorens+J%5BAuthor%5D](www.ncbi.nlm.nih.gov/pubmed/?term=Llorens+J%5BAuthor%5D)

**CRANIOFACIAL PHENOTYPE AND RARE DISEASES** (School of Medicine)

**IP:** M. Cristina Manzanares Céspedes

**Diseases:** Hypodontia (ORPHA2227) (K00.0); Oligodontia (ORPHA99798) (K00.0); Amelogenesis imperfecta (ORPHA88661) (K00.5); Dentinogenesis imperfecta (ORPHA49042) (K00.5).

**Research Line:** Identification of oral and craniofacial phenotypes for rare diseases diagnostic.

**External collaborations**

**Capacities**

<table>
<thead>
<tr>
<th>MECHANISM AND TREATMENT DISCOVERY</th>
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<th>CLINICAL DATA</th>
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<tbody>
<tr>
<td>Early Diagnosis. Professional training. Scientific dissemination.</td>
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</table>

**Description**

- Genetic and proteomic studies.
- Developmental research in animal models.

**Publications:** [www.ncbi.nlm.nih.gov/pubmed/?term=Manzanares+MC%5BAuthor%5D](www.ncbi.nlm.nih.gov/pubmed/?term=Manzanares+MC%5BAuthor%5D)
HUMAN MOLECULAR GENETICS GROUP (GGMH) (School of Medicine)

IP: Virginia Nunes Martínez

Diseases: Wolfram syndrome (ORPHA3463) (H48.0)

Research Line: Molecular characterization and generation of disease registry.

<table>
<thead>
<tr>
<th>Capacities</th>
<th>MECHANISM AND TREATMENT DISCOVERY</th>
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<td>Spanish (REWBA) and European</td>
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<td>(EURO-WABB) registries.</td>
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<td>• Access to international clinical data.</td>
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External collaborations

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Publications: [www.ncbi.nlm.nih.gov/pubmed/?term=Nunes+V%5BAuthor%5D](www.ncbi.nlm.nih.gov/pubmed/?term=Nunes+V%5BAuthor%5D)
PNEUMOLOGY RESEARCH GROUP (School of Medicine)

IP: Maria Molina Molina

Diseases: Idiopathic pulmonary fibrosis (ORPHA2032) (J84.1)

Research Line: Study of cellular and extracellular interactions, possible paths to inhibit progression.


- Anatomical pathology analysis.
Diseases of the musculoskeletal system and connective tissue

VOLUME VISUALIZATION AND ARTIFICIAL INTELLIGENCE, WAI (School of Mathematics)
IP: Anna Puig Puig
Diseases: Scoliosis diseases (M41).
Research Line: Identification and classification of cerebral malformations presents in the disease by artificial intelligence. The medical imaging technology provided by the group is also applicable to identification and diagnosis of several conditions coursing with malformations.

**Capacities**

- MECHANISM AND TREATMENT DISCOVERY
- DIAGNOSTIC BIOMARKERS
- IN VITRO MODELS
- IN VIVO MODELS
- CLINICAL DATA

**External collaborations**

- MECHANISM AND TREATMENT DISCOVERY
- DIAGNOSTIC BIOMARKERS
- IN VITRO MODELS
- IN VIVO MODELS
- CLINICAL DATA

**Description**
- 3D Modeling and biomechanical simulation of malformations
- Medical knowledge and protocols and Patient data access.
- Biomedical data transfer
- Biomechanical simulations

Publications: [www.maia.ub.es/~anna/home.html](http://www.maia.ub.es/~anna/home.html)

SYSTEMIC AUTOIMMUNE DISEASES (School of Medicine)
IP: Ricard Cervera Segura
Diseases: Systemic vasculitis (ORPHA52759) (M05.2); Autoinflammatory syndrome (ORPHA93665); Systemic lupus erythematosus (ORPHA36) (M32); Systemic sclerosis (ORPHA90291) (M34); Sjögren’s syndrome (ORPHA378) (M35.0).
Research Line: Autoimmune diseases.

**Capacities**

- MECHANISM AND TREATMENT DISCOVERY
- DIAGNOSTIC BIOMARKERS
- IN VITRO MODELS
- IN VIVO MODELS
- CLINICAL DATA

**External collaborations**

- MECHANISM AND TREATMENT DISCOVERY
- DIAGNOSTIC BIOMARKERS
- IN VITRO MODELS
- IN VIVO MODELS
- CLINICAL DATA

**Description**
- Research on systemic autoimmune diseases. Giant cell arteritis registry.
- Basic research on pathological mechanisms.


NEUROPHYSIOLOGY LAB (School of Medicine)
IP: Xavier Gasull Casanova
Diseases: Sjögren’s syndrome (ORPHA378) (M35.0)
Research Line: Gene and protein therapies to hereditary retinal dystrophies: a neuroprotective approach.

**Capacities**

- MECHANISM AND TREATMENT DISCOVERY
- DIAGNOSTIC BIOMARKERS
- IN VITRO MODELS
- IN VIVO MODELS
- CLINICAL DATA

**External collaborations**

- MECHANISM AND TREATMENT DISCOVERY
- DIAGNOSTIC BIOMARKERS
- IN VITRO MODELS
- IN VIVO MODELS
- CLINICAL DATA

**Description**
- Sensorial neuron alterations. Receptors and ion channels implicated in the diseases.
- Identification of diseases-related ion channels alterations.

Publications: [www.ncbi.nlm.nih.gov/pubmed/?term=Gasull+X%5BAuthor%5D](http://www.ncbi.nlm.nih.gov/pubmed/?term=Gasull+X%5BAuthor%5D)
Diseases of the musculoskeletal system and connective tissue

NUTRITIONAL QUALITY AND LIPID TECHNOLOGY (School of Pharmacy)

IP: Magdalena Rafecas Martínez

Diseases: Fibromyalgia (ORPHA41842) (M79.7)

Research Line: Bioactive compounds intake and symptomatological improvement. Epidemiologic studies.

<table>
<thead>
<tr>
<th>Capacities</th>
<th>Mechanism and Treatment Discovery</th>
<th>Diagnostic Biomarkers</th>
<th>In Vitro Models</th>
<th>In Vivo Models</th>
<th>Clinical Data</th>
<th>Others</th>
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</thead>
<tbody>
<tr>
<td>Description</td>
<td>Nutrition and food science. Epidemiology and symptomatological evaluation.</td>
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</table>

Publications: [www.ncbi.nlm.nih.gov/pubmed/?term=Rafecas+M%5BAuthor%5D](https://www.ncbi.nlm.nih.gov/pubmed/?term=Rafecas+M%5BAuthor%5D)
RARE AND ORPHAN DISEASES

Congenital malformations and chromosomal abnormalities

CELL CYCLE SIGNALING AND CHECKPOINTS (School of Medicine)
IP: Neus Agell Jané
Diseases: Noonan syndrome (ORPHA648) (Q87.1).
Research Line: K-Ras4B activity regulation in the Noonan Syndrome

<table>
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<tr>
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<tbody>
<tr>
<td>Description</td>
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<td>K-Ras4B activity regulation</td>
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Publications: [www.ncbi.nlm.nih.gov/pubmed/?term=Agell+N%5BAuthor%5D](www.ncbi.nlm.nih.gov/pubmed/?term=Agell+N%5BAuthor%5D)

MEMBRANE TRAFFIC (School of Medicine)
IP: Gustavo Egea Guri
Diseases: Marfan syndrome (ORPHA558) (Q87.4)
Research Line: Transport of the membrane components (lipids and proteins) within the cell. Physiopathological bases.

<table>
<thead>
<tr>
<th>Capacities</th>
<th>MECHANISM AND TREATMENT DISCOVERY</th>
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<tbody>
<tr>
<td>Description</td>
<td></td>
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<td></td>
<td>Interaction of transport intermediaries, actin cytoskeleton and its associated proteins and regulators.</td>
</tr>
</tbody>
</table>

External collaborations

- Access to clinical data

Publications: [www.ncbi.nlm.nih.gov/pubmed/?term=Egea+G%5BAuthor%5D](www.ncbi.nlm.nih.gov/pubmed/?term=Egea+G%5BAuthor%5D)

HUMAN MOLECULAR GENETICS (School of Biology)
IP: Daniel Grinberg Vaisman
Diseases: Costello syndrome (ORPHA3071) (Q87.8).
Research Line: Genetic basis and functional studies on neurologic channel diseases.

<table>
<thead>
<tr>
<th>Capacities</th>
<th>MECHANISM AND TREATMENT DISCOVERY</th>
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<td>Description</td>
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<td></td>
<td>Genetic basis and functional studies. Genetic diagnosis.</td>
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External collaborations

- Design and synthesis of active compounds and chaperones.
- Computational biology.
- Access to clinical data.

Publications: [www.ncbi.nlm.nih.gov/pubmed/?term=Grinberg+D%5BAuthor%5D](www.ncbi.nlm.nih.gov/pubmed/?term=Grinberg+D%5BAuthor%5D)
RARE AND ORPHAN DISEASES

Congenital malformations and chromosomal abnormalities

HUMAN GENETICS GRUP (School of Medicine)
IP: Rafael Oliva Virgili
Diseases: Male infertility due to microdeletions in Y chromosome (ORPHA217034) (N46).
Research Line: Male infertility

<table>
<thead>
<tr>
<th>Capacities</th>
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<tbody>
<tr>
<td>Genome, transcriptome and proteome sperm analysis.</td>
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FUNCTIONAL CHARACTERIZATION OF HERC PROTEINS AND THEIR IMPLICATIONS IN NEURODEGENERATION AND CANCER (School of Medicine)
IP: Jose Luís Rosa López
Diseases: Tuberous sclerosis (ORPHA805) (Q85.1); Angelman syndrome (ORPHA72) (Q93.5)
Research Line: Integrating genetic, molecular and cellular analyses to better understand the mechanisms altered and therapeutic opportunities.

<table>
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<tr>
<th>Capacities</th>
<th>MECHANISM AND TREATMENT DISCOVERY</th>
<th>DIAGNOSTIC BIOMARKERS</th>
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<th>IN VIVO MODELS</th>
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<tbody>
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<td>Genetic, molecular and cellular analyses.</td>
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<th>CLINICAL DATA</th>
<th>OTHERS</th>
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<tbody>
<tr>
<td>Multinational collaboration on genetic, cellular, molecular and pharmacologic analyses.</td>
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## RARE AND ORPHAN DISEASES

### Transversals

**CELLULAR THERAPY PROGRAM, TCUB** (School of Medicine)

**IP:** Josep Maria Canals Coll  
**Research Line:** Research and Development Unit with a new technology platform to produce cell or gene based medicinal products for research and clinical applications.

**Publications:**  
[www.ub.edu/TCUB](http://www.ub.edu/TCUB)  

### Computational Biology and Drug Design

**School of Pharmacy**

**IP:** Xavier Barril  
**Diseases:** Diseases with protein instability or missfolding.  
**Research Line:** Drug discovery

**Publications:**  
[www.ncbi.nlm.nih.gov/pubmed/?term=Barril+X%5BAuthor%5D](http://www.ncbi.nlm.nih.gov/pubmed/?term=Barril+X%5BAuthor%5D)

### Growth Factors, Hormones and Metabolism

**School of Medicine**

**IP:** José Carlos Perales Losa  
**Research Line:** Antisense therapy.

**Publications:**  
[www.ncbi.nlm.nih.gov/pubmed/?term=Perales+JC%5BAuthor%5D](http://www.ncbi.nlm.nih.gov/pubmed/?term=Perales+JC%5BAuthor%5D)

### Capacities

<table>
<thead>
<tr>
<th>Capabilities</th>
<th>Mechanism and Treatment Discovery</th>
<th>Diagnostic Biomarkers</th>
<th>In Vitro Models</th>
<th>In Vivo Models</th>
<th>Clinical Data</th>
<th>Others</th>
</tr>
</thead>
</table>

- **GMP facilities for clinical application.**
- **Culture rooms for preclinical research.**
- **New clinical protocols for stem cells.**
- **Packaging and cryopreservation areas.**
- **High qualified technicians.**

- **Computational tools for active compounds identification.**
- **Protein stability assays.**

- **Modified siRNAs for silencing gene expression, mainly focused on inflammatory diseases.**

- **Validation of the modified siRNAs efficacy.**

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ONCOLOGICAL THERAPY, INFLAMMATION AND IMMUNOMODULATION (School of Pharmacy)
IP: Carlos Ciudad Gómez
Research Line: Antisense therapy.

<table>
<thead>
<tr>
<th>Capacities</th>
<th>MECHANISM AND TREATMENT DISCOVERY</th>
<th>DIAGNOSTIC BIOMARKERS</th>
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<th>CLINICAL DATA</th>
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<tr>
<td>Description</td>
<td>Coding polyurine hairpins (PPRHs) for gene expression targeting, mainly focused on oncologic diseases.</td>
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External collaborations

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<tr>
<td>Validation of the PPRHs efficacy.</td>
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Publications: [www.ncbi.nlm.nih.gov/pubmed/?term=Ciudad+CJ%5BAuthor%5D](www.ncbi.nlm.nih.gov/pubmed/?term=Ciudad+CJ%5BAuthor%5D)

SERVICE OF DEVELOPMENT OF MEDICINES (SDM) (IDIBELL, School of Pharmacy)
IP: Josep Maria Suñé Negre
Research Line: Technical, scientific and research services, to develop medical devices and medicines for both human and animal use.

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Publications: [www.ub.edu/sdm/in_index.htm](www.ub.edu/sdm/in_index.htm)