



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 refered to the Orphanet's Orpha number.

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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.

Description





Infectious and parasitic diseases

Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Selection and synthesis of active compounds.
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Publications: www.ncbi.nlm.nih.gov/pubmed/?term=Luque+FJ%5BAuthor%5D

MULTIVARIANT AND COMPUTATIONAL STATISTICS (School of Biology)

IP: Antonio Monleón Getino

Diseases: Tuberculosis (ORPHA3389) (A15-A19)

Research Line: Study of tuberculosis incidence and trends.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Statistics, population analysis.

Publications: www.ncbi.nlm.nih.gov/pubmed/?term=Monleon-Getino%5BAuthor%5D

INFECTIOUS PATHOLOGY AND ANTIBIOTIC SENSITIVITY (School of Medicine)

Infectious Diseases Unit (Hospital of Bellvitge) IP: Miguel Ángel Santín Cerezales Diseases: Tuberculosis (ORPHA3389) (A15-A19)

esearch Line: Strategies to take therapeutic decisions during the following of tuberculosis cases.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Standard strategy based on different diagnostic methods.

Publications: www.ncbi.nlm.nih.gov/pubmed/?term=Santin+M%5BAuthor%5D

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.

Description





Infectious and parasitic diseases

Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Diagnosis, molecular classification and drug assay.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	 Nanotechnology for drug formulations. Patient data access. Parasitic diagnosis methods. Treatment efficacy. Genetic analysis. Population studies.

Publications: 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)

Research Line: Antimicrobial resistance, molecular mechanisms of action.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	<i>IN VITRO</i> MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	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.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	 Parasite culture and maintenance of T. cruzi strains. New lipid-based nanoformulations.

Publications: www.ncbi.nlm.nih.gov/pubmed/?term=Vinuesa+T%5BAuthor%5D





Neoplasms and oncological diseases

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

B: Emilia Mantagenet Costa

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

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Founder member of the International Workshop on Chronic Lymphocytic Leukemia and the European Task Force for Lymphomas.

Publications: 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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Molecular modeling
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	 In vitro and in vivo research and validation assays.

Publications: <u>www.ncbi.nlm.nih.gov/pubmed/?term=Rubio+JE%5BAuthor%5D</u>

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

IP: Luis Francisco Santamaría Babí

Diseases: Primary cutaneous T-cell lymphoma (CTCL) (ORPHA178533) (C84.4) Research Line: Translational immunology

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	Development and study of a CTCL animal model.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	 Access to clinical data and samples.

Publications: <u>www.ncbi.nlm.nih.gov/pubmed/?term=Santamaria-Bab%C3%AD+LF%5BAuthor%5D</u>





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).

Research Line: Antitumor drug transporters and transporter pharmacogenetics.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	<i>IN VITRO</i> MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Antitumor drug transporters and transporter pharmacogenetics
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	 Molecular analysis. Pancreatic xenografts Access to clinical data and samples.

Publications: www.ncbi.nlm.nih.gov/pubmed/?term=Pastor-Anglada+M%5BAuthor%5D



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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	<i>IN VITRO</i> MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Research on systemic autoimmune diseases. Giant cell arteritis registry.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	 Basic research on pathological mechanisms. Access to clinical data. International registries.

Publications: www.ncbi.nlm.nih.gov/pubmed/?term=Cervera+R%5BAuthor%5D

MOLECULAR PHYSIOLOGY (School of Biology)

IP: Antonio Felipe Campo

Diseases: Long QT syndrome (ORPHA768) (I45.8); Brugada syndrome (ORPHA130) (I47.2)

Research Line: Pharmacogenomic high-throughput screening. Genetical, functional and clinical studies.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	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

Publications: 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 (ORPHA64743) (Q27).

Research Line: Vascular hepatic diseases.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	<i>IN VITRO</i> MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Clinical data registries.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	 Access to international clinical data. Multicenter studies.

Publications: 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) **Research Line:** Pharmacoeconomics: studies of drug use suitability.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Equity access to orphan drugs.

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

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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Nucleosides transporters and nucleotides metabolism.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Molecular analysis.Metabolite analysis.Access to clinical data.

Publications: www.ncbi.nlm.nih.gov/pubmed/?term=Pastor-Anglada+M%5BAuthor%5D

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

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Member of <i>EUCERD</i> (European Committee of Experts in Rare Diseases) and Coordinator of <i>CATGLOBIN</i> (Catalan net for the study of Hemoglobinopathies).
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	 Metabolism and molecular pathology.

Publications: www.ncbi.nlm.nih.gov/pubmed/?term=Vives-Corrons+JL%5BAuthor%5D





Endocrine, nutritional and metabolic diseases

ADULT RARE DISEASES GROUP (School of Medicine)

IP: Francesc Cardellach López

Diseases: Congenital metabolic diseases (E70-90) and **Mithocondrial disorders. Research Line:** Diagnosis tests.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Physiopathological research studies.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	 Access to pediatric clinical data. Biochemical diagnosis method. Mitochondrial analysis.

Publications: 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)

Research Line: Folding and quality control of membrane proteins and associated pathologies.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Protein missfolding associated to disease mutations.

Publications: 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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	Drug discovery
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	 Biochemical assays and screening of new compounds. Mutation analysis and inhibitors evaluation.

Publications: www.ncbi.nlm.nih.gov/pubmed/?term=Delgado+A%5BAuthor%5D





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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	Genetic and biochemical assays. Animal models (murine, zebra fish). Phisiopathology studies. Astrocytic function evaluation.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	 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

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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	<i>IN VITRO</i> MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	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

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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	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





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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Chemical synthesis of molecular chaperones.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	 Computational chemistry Genetics, biochemistry and pharmacology.

Publications: 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) Research Line: Pharmacoeconomics: studies of drug use suitability.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Equity access to orphan drugs.

Publications: 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)

Research Line: Kidney aminoacid reabsorption. Generation of animal models. New gens and proteins implicated in the diseases.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	Animal model generation. Genetic and biochemical studies.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	 New gens description. RMN studies. In vivo models characterization. Transporters knowledge. Access to clinical data. Anatomopathology.

Publications: www.ncbi.nlm.nih.gov/pubmed/?term=Nunes+V%5BAuthor%5D





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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Molecular bases and potential therapeutic targets.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Access to clinical data and samples.Molecular characterization.

Publications: 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)

Research Line: Molecular mechanisms of action. Antimicrobial resistance.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	Susceptibility of <i>Pseudomonas</i> <i>aeruginosa</i> from CF patients to the treatment with nanoencapsulated antibiotics.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Gene therapy.Nanoparticles production.

Publications: www.ncbi.nlm.nih.gov/pubmed?term=Vi%C3%B1as%20M%5BAuthor%5D&cauthor=true&cauthor_uid=22702530





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).

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	Different approaches to HD physiopathology and therapy.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	 International collaborators for multiple experimental lines.

Publications: 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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Genetic, molecular and cellular studies.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Access to pediatric clinical data.Biomarker studies.Cell models.

Publications: 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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	New protocols to make clinical grade muscle stem cells and motor neuron stem cells.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	 In collabotion with the Cell teraphy program of the University of Barcelona.

Publications: www.ncbi.nlm.nih.gov/pubmed/?term=Edel+MJ%5BAuthor%5D



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 sydrome).

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Design and synthesis of compounds.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	<i>IN VITRO</i> MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	 In vivo studies. Formulation of compounds for in vivo administration.

Publications: 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)

							Description
						S	Research on structure and function of
Conscition		DIAGNOSTIC	IN VITRO	IN VIVO	CLINICAL	Ë	human nuclear receptors using X-ray
capacities		BIOMARKERS	MODELS	MODELS	DATA	E	crystallography and other structure-
	DISCOVERT					0	function approaches.

Publications: 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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	Genetic and biochemical assays. MLC1 interactome. Animal models (murine, zebra fish). Phisiopathology studies. Astrocytic function evaluation.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	 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





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) Research Line: Pharmacogenomic high-throughput screening. Genetical, functional and clinical studies.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Pharmacogenomic high-throughput screening. Ion channels analysis.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	 Access to clinical data

Publications: http://www.ncbi.nlm.nih.gov/pubmed/?term=Felipe+Antonio%5BAuthor%5D

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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Sensorial neuron alterations. Receptors and ion channels implicated in the diseases.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	 Identification of diseases-related ion channels alterations.

Publications: 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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	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

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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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	Genetic, molecular and cellular analyses.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	 Neurological and behavioral analysis.

Publications: 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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	<i>IN VITRO</i> MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	President 2011-2013, International Neurotoxicology Association (ina14.org)
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	 Vestibular physiopathology knowledge. In vitro models. Neuromuscular models.

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

VOLUME VISUALIZATION AND ARTIFITIAL 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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	3D Modeling and biomechanical simulation of malformations
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	 Medical knowledge and protocols and Patient data access. Biomedical data transfer Biomechanical simulations

Publications: www.maia.ub.es/~anna/home.html



Behavioral disorders and diseases of the nervous system

FUNCTIONAL CHARACTERIZATION OF HERC PROTEINS AND THEIR IMPLICATIONS IN NEURODEGENERATION

AND CANCER (School of Medicine)

IP: Jose Luís Rosa López

Diseases: Cerebellar ataxia (ORPHA4145) (G11).

Research Line: Integrating genetic, molecular and cellular analyses to better understand the mechanisms altered and therapeutic opportunities.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	Genetic, molecular and cellular analyses.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	 Multinational collaboration on genetic, cellular, molecular and pharmacologic analyses.

Publications: <u>www.ncbi.nlm.nih.gov/pubmed/?term=Rosa+JL%5BAuthor%5D</u>

MOLECULAR PATHOLOGY AND THERAPY FOR HETEROGENETIC AND POLIGENETIC DISEASES (IRB, School of

Biology)

IP: Antonio Zorzano

Diseases: Charcot-Marie-Tooth disease (ORPHA166) (G60.0).

Research Line: Study of the pathologic fission in type 2A Charcot-Marie-Tooth neuropathology and seeking of pharmacologic therapies.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	<i>IN VITRO</i> MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Genetic, molecular and cellular analyses.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	 Access to clinical data.

Publications: www.ncbi.nlm.nih.gov/pubmed/?term=Zorzano+A%5BAuthor%5D





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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Physiopathological research studies.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	 Access to pediatric clinical data. Biochemical diagnosis method. Mitochondrial analysis.

Publications: 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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	<i>IN VITRO</i> MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Sensorial neuron alterations. Receptors and ion channels implicated in the diseases.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	 Identification of diseases-related ion channels alterations.

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

HUMAN MOLECULAR GENETICS (IBUB, School of Biology)

IP: Roser Gonzàlez 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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	Genetic diagnosis. Genetic basis and functional in vivo studies.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	 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





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) **Research Line:** Genetic basis and functional studies on neurologic channel diseases.: migraine and ataxia. Functional genomics for retinitis pigmentosa studies. Design of genetic diagnosis chip for retina dystrophies.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	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

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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	President 2011-2013, International Neurotoxicology Association (ina14.org)
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	 Vestibular physiopathology knowledge. In vitro models. Neuromuscular models.

Publications: 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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	Early Diagnosis. Professional training. Scientific dissemination.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	 Genetic and proteomic studies. Developmental research in animal models.

Publications: <u>www.ncbi.nlm.nih.gov/pubmed/?term=Manzanares+MC%5BAuthor%5D</u>





Diseases of the eye and ear and craniofacial malformations

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.

-							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Spanish (REWBA) and European (EURO-WABB) registries.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	<i>IN VITRO</i> MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	 Access to international clinical data.

Publications: www.ncbi.nlm.nih.gov/pubmed/?term=Nunes+V%5BAuthor%5D





Diseases of the respiratory system and the digestive system

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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	Pneumology Hospital Unit
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	 Anatomical pathology analysis.

Publications: www.ncbi.nlm.nih.gov/pubmed/?term=Molina-Molina+M%5BAuthor%5D



Diseases of the musculoskeletal system and connective tissue

VOLUME VISUALIZATION AND ARTIFITIAL 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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	3D Modeling and biomechanical simulation of malformations
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	 Medical knowledge and protocols and Patient data access. Biomedical data transfer Biomechanical simulations

Publications: 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 (ORPHA536) (M32); Systemic sclerosis (ORPHA90291) (M34); Sjögren's syndrome (ORPHA378) (M35.0). Research Line: Autoimmune diseases.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	<i>IN VITRO</i> MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Research on systemic autoimmune diseases. Giant cell arteritis registry.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	<i>IN VITRO</i> MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	 Basic research on pathological mechanisms. Access to clinical data. International registries.

Publications: www.ncbi.nlm.nih.gov/pubmed/?term=Cervera+R%5BAuthor%5D

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.

Description MECHANISM Sensorial neuron alterations. OTHERS DIAGNOSTIC Capacities AND TREATMENT Receptors and ion channels implicated **BIOMARKERS** DISCOVERY in the diseases. MECHANISM AND External DIAGNOSTIC IN VITRO IN VIVO CLINICAL Identification of diseases-related ion TREATMENT collaborations BIOMARKERS MODELS MODELS DATA channels alterations. DISCOVERY

Publications: 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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Nutrition and food science. Epidemiology and symtomatological evaluation.

Publications: <u>www.ncbi.nlm.nih.gov/pubmed/?term=Rafecas+M%5BAuthor%5D</u>





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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

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	K-Ras4B activity regulation

Publications: 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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	Interaction of transport intermediaries, actin cytoskeleton and its associated proteins and regulators.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	 Access to clinical data

Publications: <u>www.ncbi.nlm.nih.gov/pubmed/?term=Egea+G%5BAuthor%5D</u>

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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	<i>IN VITRO</i> MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	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





Congenital malformations and chromosomal abnormalities

HUMAN GENETICS GRUP (School of Medicine)

IP: Rafael Oliva Virgili

Diseases: Male infertility due to microdeletions in Y cromosome (ORPHA217034) (N46). **Research Line:** Male infertility

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Genome, transcriptome and proteome sperm analysis.

Publications: www.ncbi.nlm.nih.gov/pubmed/?term=Oliva+R%5BAuthor%

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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	<i>IN VIVO</i> MODELS	CLINICAL DATA	OTHERS	Genetic, molecular and cellular analyses.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	 Multinational collaboration on genetic, cellular, molecular and pharmacologic analyses.

Publications: www.ncbi.nlm.nih.gov/pubmed/?term=Rosa+JL%5BAuthor%5D





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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	GMP facilities for clinical application. Culture rooms for preclinical research. New clinical protocols for stem cells. Packaging and cryopreservation areas. High qualified technicians.

Publications: www.ub.edu/TCUB

http://www.ub.edu/web/ub/en/menu eines/noticies/2013/05/096.html?

COMPUTATIONAL BIOLOGY AND DRUG DESIGN (School of Pharmacy)

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

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Computational tools for active compounds identification. Protein stability assays.

Publications: 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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Modified siRNAs for silencing gene expression, mainly focused on inflammatory diseases.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	 Validation of the modified siRNAs efficacy.

Publications: www.ncbi.nlm.nih.gov/pubmed/?term=Perales+JC%5BAuthor%5D





Transversals

ONCOLOGICAL THERAPY, INFLAMMATION AND IMMUNOMODULATION (School of Pharmacy)

IP: Carlos Ciudad Gómez

Research Line: Antisense therapy.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Coding polypurine hairpins (PPRHs) for gene expression targeting, mainly focused on oncologic diseases.
External collaborations	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	 Validation of the PPRHs efficacy.

Publications: 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.

							Description
Capacities	MECHANISM AND TREATMENT DISCOVERY	DIAGNOSTIC BIOMARKERS	IN VITRO MODELS	IN VIVO MODELS	CLINICAL DATA	OTHERS	Drug formulation

Publications: www.ub.edu/sdm/in index.htm



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