

Nephrology

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DESCRIPTION

The Nephrology Research Group of Fundació Puigvert carries out translational research in the field of renal diseases. Most of our research is clinical (clinical trials, large cohorts of patients, collaborative studies, participation in clinical guidelines, observational studies in specific groups of renal patients, etc). The group's basic research focuses on the study of fibrosis and inflammation in kidney transplantation and glomerular diseases, and genomic as well as clinical studies in inherited kidney diseases.

The group actively participates in Spanish research networks in renal diseases, as well as some European ones.

MAIN LINES OF RESEARCH

- Inherited kidney diseases: research in this area is carried out from both the clinical and genomic perspective. The first one led by R. Torra and the second one by E. Ars. Both have led many research projects on these diseases, and have supervised several doctoral theses.
- Dr. R. Torra new line of research consist on the use of artificial intelligence for the diagnosis of inherited kidney diseases: 1) Use of a machine learning approach to develop a diagnostic tool and 2) use of natural Language processing to identify undiagnosed inherited kidney diseases within electronic health records. Both are funded projects.
- The hereditary renal diseases team is currently analyzing phenotype variability of autosomal dominant diseases, both in its genomic and its clinical dimension ad developing a prediction tool



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for autosomal dominant Alport syndrome. Also, several studies have been performed to test prediction tools for autosomal dominant polycystic kidney disease.

- Dr. E. Ars has led the setup of the genetic testing pipeline by developing a gene panel (massive parallel sequencing) containing more than 400 genes causing inherited kidney diseases. This panel has been used for several cohort studies. Her funded research right now deepens into negative genetic tests by using RNA analysis.
- Kidney Transplantation: led by Dr. L. Guirado and Dr. C. Facundo. The group is participating in or leading more than 20 studies, including clinical trials of drugs for the treatment of kidney rejection or associated viral infections. Their active participation and leadership in the investigation of biochemical, serological and genomic biomarkers of both rejection and viral infections associated with kidney transplantation is outstanding. They are also part of an EU initiative on the use of AI for kidney transplant prognosis.
- A new study based on HLA epitope and interferon elispot incompatibilities detected between donor and recipient prior to transplantation in the Living Donor Kidney Transplant Program is underway; it aims to individualize immunosuppression with the objective of maintaining a low rejection rate by minimizing the dose of immunosuppression administered.
- The group is starting a collaboration with the Microbiology and Infectious Diseases group to study the impact of microbiota on recurrent urinary tract infections in kidney transplant recipients.
- Glomerular diseases: This is an important area of nephrology led by Dr. M. Diaz with many RCTs, observational and interventional studies in different glomerulopathies. Basic research has also been done in this area by studying the process of fibrosis and inflammation. New projects on IgA nephropathy (led by Dr. H Marco) and onconephrology (led by Dr. X Barros) are on its way.
- Renal osteodystrophy: led by Dr. MJ. Lloret. She is conducting a pilot study to bone quality with bone microindentation (in addition to bone

quantity with DXA) in renal patients. Dialysis: Clinical studies are led by Dr. JM. Diaz and studies on genomic damage research and the effect of BPA and plastics on it are led by Dr. E. Coll.

- Cardiovascular risk in CKD. Led by Dr. P. Fernández-Llama studies the 24-hour central arterial pressure, its circadian rhythms and their relationship with organic damage. This group is part of RICORS coordinated by Dr. Elisa Llurba on the topic of pregnancy and CKD.
- Ms. Luz San Miguel, Dr. Cristina Canal, Ms. Judith Ballart, Ms. Irene Silva and Ms. Beatriz Bardají are the coordinators of Clinical studies and trials. Their work is essential and of the utmost importance for carrying out the high number of clinical trials performed by the group.

SCIENTIFIC CHALLENGES

- Strengthen our translational character and maintain our scientific production.
- To make the transfer of research results to society our main objective.
- To maintain and strengthen our relationship with patient associations in order to progressively integrate the RRI approach into our translational research, with the aim of better aligning the research process and its results with the values, needs and expectations of our patients.
- Enhance innovation by incorporating artificial intelligence tools and collaborating with Information and Communication Technologies (ICT) for educational purposes and patient empowerment.
- To continue the active collection of research samples from CKD and transplant patients.
- To work towards precision medicine, especially in the field of kidney transplantation.
- Strengthen collaborative research with other IR groups.
- To maintain and increase participation in national working groups and to increase international collaboration. To achieve access to European project funds in the field of rare inherited diseases.
- To consolidate our participation in the development of national and international clinical nephrology guidelines.



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- To increase our participation in relevant private pharmaceutical clinical trials, especially early stage clinical trials.
- Improve the statistical support for our scientific production.

ACTIVE GRANTS

- Ars Criach, Elisabet. Heterogeneidad clínica, genética y alélica de las enfermedades renales hereditarias de debut temprano: nuevas causas moleculares de anomalías congénitas del riñón y el tracto urinario. PI19/01633. Instituto de Salud Carlos III (ISCIII). Duration: 2020-2024. 93.170,00 €.
- Diaz Encarnacion, Maria Montserrat. Papel de la vía del succinato en la inflamación de los riñones procedentes de donantes cadáver. FMM 2019. Fundación Mutua Madrileña. Duration: 2019-2023. 99.997,00 €.
- Fernández De La Llama, Patricia. Primary care interventions to prevent maternal and child chronic diseases of perinatal and developmental origin. RD21/0012/0019. Instituto de Salud Carlos III (ISCIII). Duration: 2022-2024. 124.080,00 €.
- Fernández De La Llama, Patricia. Evolución de la variabilidad de la presión arterial posicional y temporal y su relación con el daño orgánico en ancianos hipertensos. Fundación Senefro. Ayudas 2019 a la investigación en Nefrología. Duration: 2019-2022. 24.000 € (F.PUIGVERT).
- Lloret Cora, Maria Jesus . Estudio del comportamiento de biomarcadores de recambio óseo durante el primer año post-trasplante renal. BECA FEIOMM 2022 INVESTIGACIÓN CLÍNICA. Sociedad Española de Investigación Ósea y Metabolismo Mineral. Duration: 2022-2022. 7.000,00 € (F.PUIGVERT).
- Torra Balcells, Roser. Use of artificial intelligence and systems biology for diagnosis and personalized risk assessment for inherited kidney diseases, focusing on alport syndrome. MARATO 202036-30. Fundació La Marató de TV3. Duration: 2021-2024. 112.500,00 €.
- Torra Balcells, Roser. Modelos de predicción en las nefropatías hereditarias autosómicas dominantes. PI18/00362. Instituto de Salud Carlos III (ISCIII). Duration: 2019-2022. 99.220,00 €.

- Torra Balcells, Roser. Búsqueda de enfermedades renales hereditarias no sospechadas: una aguja en un pajar. PI22/00361. Instituto de Salud Carlos III (ISCIII). Duration: 2023-2025. 123.420,00 €.
- Torra Balcells, Roser. Inflamación e inmunopatología de órganos y sistemas. RD21/0005/0006-1. Instituto de Salud Carlos III (ISCIII). Duration: 2022-2024. 105.765,00 €.

GRANTS AWARDED

- Ars Criach, Elisabet. Estudio genómico y transcriptómico de pacientes con nefropatía no filiada con sospecha de causa monogénica. PI23/00426. Instituto de Salud Carlos III (ISCIII). Duration: 2024-2026. 102.500,00 €.

DOCTORAL THESES DEFENDED

- Naranjo Muñoz, Javier. Modelos de predicción de rápida progresión en la poliquistosis renal autosómica dominante. 15/05/2023. Universidad de Cádiz. Supervisors: Torra Balcells, Roser; Remón Rodríguez, César.

SCIENTIFIC PRODUCTION

- Angerri O, Gracia S, Rousaud F, Kanashiro A, Emiliani E. Entering into 2.0 cystinuria management with a medical digital tool to monitor urine pH: A prospective, randomized study. Actas Urológicas Españolas. 2023; 47(9). DOI:10.1016/j.acuro.2023.02.004. IF:1,100 (Q4/9D). Document type: Article.
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