

EXPERTISE OFFER to join a consortia to address the JTC2026 “Personalised Medicine for CARdiovascular, Metabolic, and kidNey diseases” (CARMEN2026)

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We are interested in participating in a project that is looking for PM strategies for patients with kidney diseases. Namely, projects that share a genetic aspect of translational research on either common or rare kidney disease, including the renal transplant population.

In the last decade, we have gained expertise in clinical evaluation of adult and pediatric kidney patients of distinct ancestries with inherited kidney diseases, actively recruiting for ERKregistry, and characterising patients with chronic kidney disease (CKD) of unknown etiology in the transplantation setting. We have also implemented molecular genetic tools aiming at research and diagnosis, including genotyping Taqman assays (APOL1 and CYP3A5) and next-generation sequencing gene panels for mendelian kidney phenotypes. Our group has pioneered in Portugal the introduction of *personalized* as well as outcome-modifying therapeutics for hemolytic uremic syndrome mediated by the alternative complement pathway (Eculizumab) and Autosomal Dominant Polycystic Kidney Disease-ADPKD (Tolvaptan). Presently, we are studying metabolic and inflammatory profiles in Tolvaptan *treatment response* in ADPKD. Finally, and for the APOL1-mediated kidney, early disease *risk prediction* is being implemented through the APOL1 G1/G2 genotyping in at-risk populations.

As an MD, PhD, we have access to cohorts of patient samples.