

New approach to early diagnosis of chronic renal diseases in children

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Credit: AI-generated image ([disclaimer](#))

New mutations of the different pathologies found in renal diseases have been discovered by researchers from the University of Oviedo, Spain.

Primary tubulopathies - renal diseases categorized as congenital or genetic disorders that usually affect children- have no known cure and

often negatively affect the quality of life of patients. The disease can impede early development through stunted growth, [renal failure](#) or rickets. Aiming to address the debilitating effects on child development that primary tubulopathies carry, the RenalTube research project designed a clinical database to house diagnostic information on patients affected by the disease.

The team coordinated by Doctor Fernando Santos and Doctor Helena Gil-Peña, from the University of Oviedo and the Central University Hospital of Asturias (HUCA), worked with 410 international clinical researchers to form the RenalTube portal. In the three years since its conception the portal has gathered detailed information on more than 400 patients from all around the globe and published results in child nephrology [scientific journals](#).

Laboratories that participated in the project conducted an automatic sequencing of the genes, which allowed for a [genetic diagnosis](#) of the patients studied. The team's research led to the discovery of new [mutations](#) in some primary tubulopathies.

The experts are spread between three laboratories working with a set of patients affected by 23 different types of pathologies, for instance research spearheaded by the Central University Hospital of Asturias (HUCA) focused on the different forms of hereditary rickets.

Patients diagnosed with a form of primary tubulopathy are able to provide clinical data to the portal. To do so, the doctor overseeing the patient's care must register to the RenalTube portal and answer a survey that covers several aspects of the patient's clinical history.

Provided by CORDIS

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