

Project: “Application of RenalTube to the needs of patients and clinical-experimental research on distal renal tubular acidosis and X-linked hypophosphatemic rickets”

Principal investigator: Dr. Helena Gil Peña

Institution: Pediatrics - Hospital Universitario Central de Asturias

General objective of the project: adapt RenalTube to the needs of the patients and maintain their exploitation as a clinical-genetic database and deepening methods for the diagnosis of distal renal tubular acidosis and therapeutic strategies applied to hereditary X-linked hypophosphatemia (XLH).

Specific objectives of the project: In coordination with other RenalTube subprojects, the following are sought: i) Develop transfer actions aimed at covering the needs of patients, in collaboration with the associations that represent them. ii) Maintain, exploit and promote the growth of RenalTube as a clinical-genetic database of primary tubulopathies. iii) In relation to the ATRD, look for children who have nephrocalcionosis, lithiasis or hypogrowth of an unclear cause, could present an incomplete ATRD clinical situation and, in the parents of children with ATRD, look for the same clinical situation. iv) In relation to XLH, find out the cardiac function in children with this tubulopathy and the influence on the disease of therapeutic strategies that normalize serum phosphorus levels.

Explanation of the project: The main objective of this project is to continue researching primary tubulopathies (rare diseases that debut in childhood and which, due to their low incidence, are still very unknown). It will focus mostly on distal renal tubular acidosis, and on hypophosphatemic rickets linked to the chromosome, developing, in addition to concrete research on these diseases, activities that meet the needs of patients. It will address this new challenge by expanding the present of primary tubulopathies to society through outreach activities, simple teaching material, non-scientific training sessions to the public. Likewise, RenalTube will try to learn about the daily situation of patients and their families, to look for strategies that improve their quality of life, health care processes, social equality and, ultimately, their health.

Funding is required for the hiring of research personnel capable of carrying out the study.

For additional information, please contact: **Helena Gil, helena.gil@sespa.es**