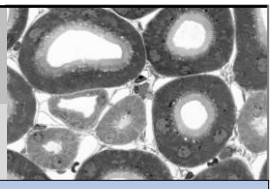
TRR 374

Tubular system and interstitium of the kidney:
(Patho-) physiology and crosstalk



Seminar

Ignacio Portales Castillo, MD

Division of Nephrology
John T. Milliken Department of Medicine
Washington University School of Medicine in St. Louis

Can we prevent renal failure in my clinic patient? Organoids and mice in GATM related renal disease

Ignacio Portales Castillo is an assistant professor of medicine in the Division of Nephrology at Washington University School of Medicine in St. Louis:

“My research is inspired directly by patients and focuses in two main areas 1) PTH receptor function, 2) Genetic causes of Fanconi Syndrome.
During my clinical nephrology fellowship, we studied patients with rare diseases affecting endochondral bone formation and we identified a role for impaired ß-arrestin recruitment leading to hypersensitive PTH receptor in Eiken syndrome (Portales Castillo et al., Communications Biology 2023). I also met a family with a rare, recently described form of Fanconi syndrome caused by mutations in a mitochondrial enzyme encoded by the gene GATM. Most of these patients progress to end-stage kidney disease. Motivated by the possibility of finding a treatment, I performed several in vitro studies to show regulation of the mutant protein by creatine in human cells including kidney organoids. We have now not only in vitro models, but also a novel mouse the mutation GatmP341L, which we use to study this disease and possible treatment options.“

Time: **Monday, February 17, 2025 13:00h**
Location: **Seminarraum Physiologie**
VKL 4.1.29
Universität Regensburg
and **Zoom**

To get the Zoom link please contact:
michaela.kritzenberger@ur.de

Universität Regensburg

Friedrich-Alexander-Universität Erlangen-Nürnberg