## Are SNPs in the calcium channel TRPV5 causing hypercalciuria?

### Clinical relevance

Kidney stone formation is a major socioeconomic problem in humans, involving pain, recurrent treatment and renal insufficiency. A positive family history predisposes individuals to an increased risk of stone formation, which strongly indicates the involvement of genetic susceptibility factors. As most renal precipitates contain calcium as a major component, hypercalciuria is the main risk factor for renal stone formation.

### Background

In the kidney, the renal epithelial calcium channel TRPV5 act as the gatekeeper protein in active calcium reabsorption. TRPV5 gene ablation in mice leads to severe hypercalciuria, implying that TRPV5 is an interesting candidate gene for renal hypercalciuria in humans. Recently, two single nucleotide polymorphisms (SNPs) A561T and A563T were shown to be highly enriched in patient cohorts with a disturbed calcium balance.

### Goals

This study aims to functionally characterize and identify the TRPV5 A561T and A563T SNPs in patients with renal hypercalciuria. Within this project we aim to answer the following questions:

- How do these SNPs alter TRPV5 channel activity?
- How do these SNPs alter TRPV5 channel structure?
- Are TRPV5 SNPs prognostic markers in idiopathic hypercalciuria?

### We offer

We offer the possibility to perform and present high-quality clinically-oriented research in a professional, multicultural and highly-motivating working environment with about 20 colleagues in a well-equipped department. You will work on this project under the supervision of an excellent Postdoc/PhD. You will have the opportunity to learn a broad range of techniques including: life-cell calcium imaging, site-directed mutagenesis, molecular cloning, cell culture, western blot and bioinformatics.

### Contact:


| | |
|---|---|
| Department: | Physiology; Group Ion Transport |
| Supervisor: | Dr. Sjoerd Verkaart |
| Contact person: | Dr. Milene Catoire |
| Telephone number: | 024-3092772 |
| Email address: | milene.catoire@radboudumc.nl |
| Website: | www.physiomics.eu |