Finding the mutation in hypomagnesemia: the genome as grab bag

Clinical relevance

As many as 65% percent of the patients in the intensive care unit suffer from hypomagnesemia, causing epilepsy, muscle weakness and cardiac arrhythmia. Therefore, tight regulation of the magnesium (Mg^{2+}) balance by the intestine and the kidney is of major importance. In a small but important group of patients the hypomagnesemia is caused by genetic defects.

Background

Over the last decade, several genes have been identified that are involved in renal Mg^{2+} transport. Mutations in these genes cause hypomagnesemia. However there are still families with hypomagnesemia of which the genetic cause is unknown. These families are now subjected to whole exome sequencing to find new genes involved in Mg^{2+} homeostasis.

Goals

Currently we are analyzing the results of the whole exome sequencing approach and we are identifying new candidates in Mg^{2+} homeostasis. In this internship we will elucidate the role of some of these candidates using a broad range of biochemical and molecular techniques. Within this project we aim to answer the following questions:

- Which gene mutations cause hypomagnesemia?
- Where are these genes expressed and can we link this to the phenotype of the patients?
- How are these genes involved in (renal) Mg^{2+} transport?

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 35 colleagues in a well-equipped department. You will work on this project under the supervision of an excellent PhD. You will have the opportunity to learn a broad range of techniques, such as molecular cloning, cell culture, immunohistochemistry, bioinformatics, real time PCR and western blot.

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