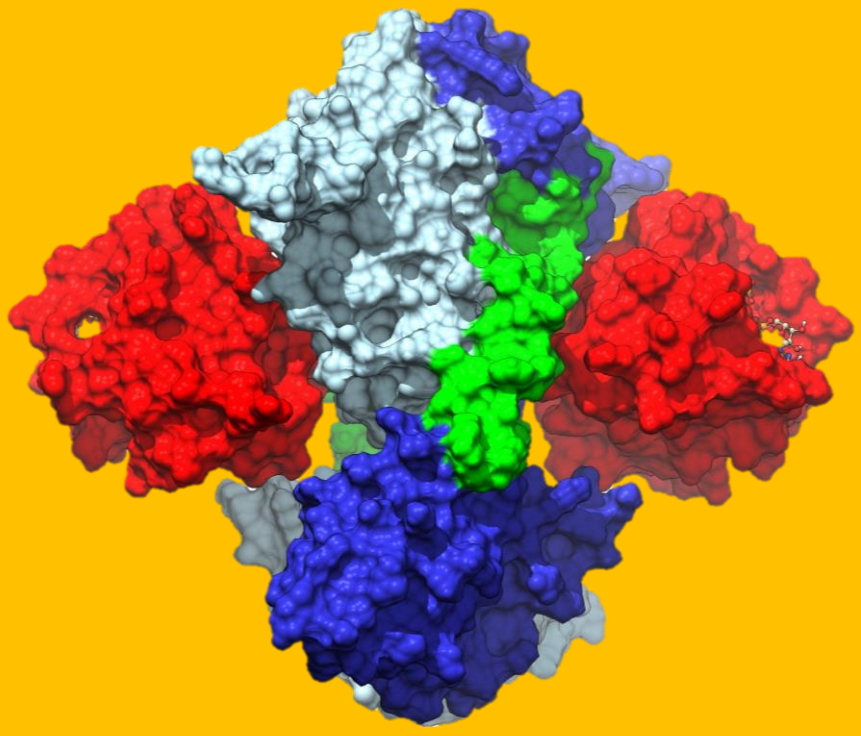
04/2021

FIGHT-CNNM 2

For Improving diagnostics and Grasping the disease mechanisms of rare Hypomagnesemia in patients with CNNM2 mutations



EUROPEAN JOINT PROGRAMME RARE DISEASES

FIGHT-CNNM2

Mutations in CNNM2 membrane proteins were identified to be causative for hypomagnesemia, intellectual disability and seizures. CNNM2 mutations are the most common genetic cause for rare dominant hypomagnesemia.

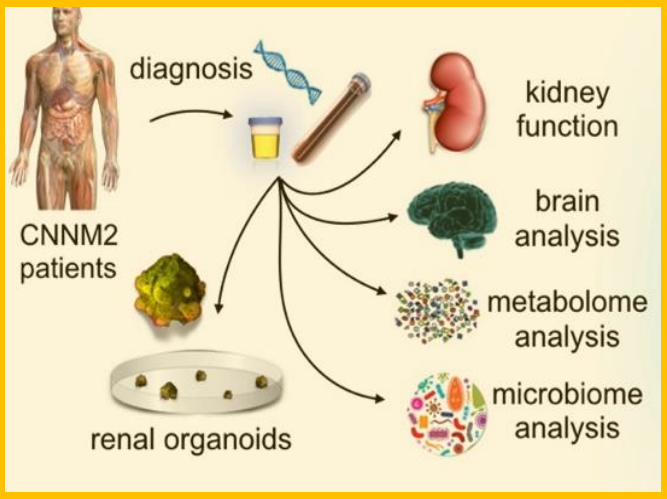
Currently, there are no clinical protocols for the diagnosis and follow-up of patients with CNNM2 mutations. Although it has been described that CNNM2 mutations impair renal magnesium reabsorption, the mechanisms that explain the disease are poorly understood. In this project proposal, we aim to improve diagnostics and the understanding of CNNM2-associated disease by integrated structural, functional and biochemical techniques.

WHAT OBJECTIVE DOES THE PROJECT PERSUE?

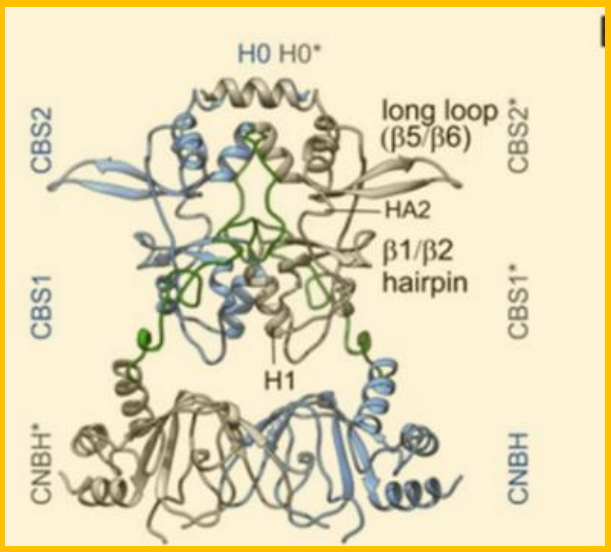
The aim is to improve the diagnosis and understanding of CNNM2 associated disease by the use of integrated structural, functional and biochemical techniques.

KEY OBJECTIVES

CNNM2 Diagnostics. By systematic phenotyping of CNNM2 patients, a novel diagnostics protocol will be developed. Patient-derived stem cells will be stored in a biobank.

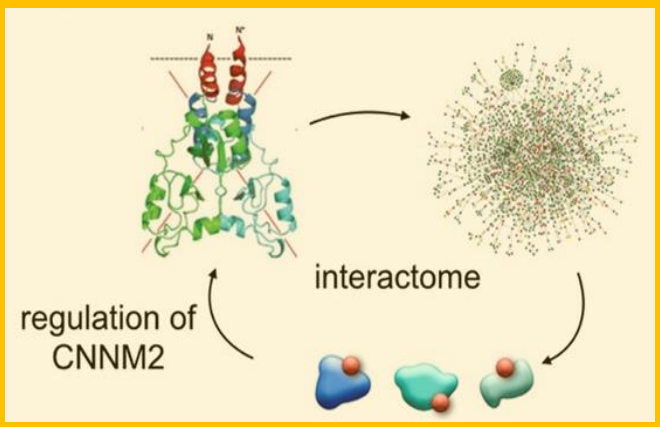


CNNM2 Structure. The CNNM2 protein structure will be uncovered to understand the impact of patient mutations on protein folding and activity.

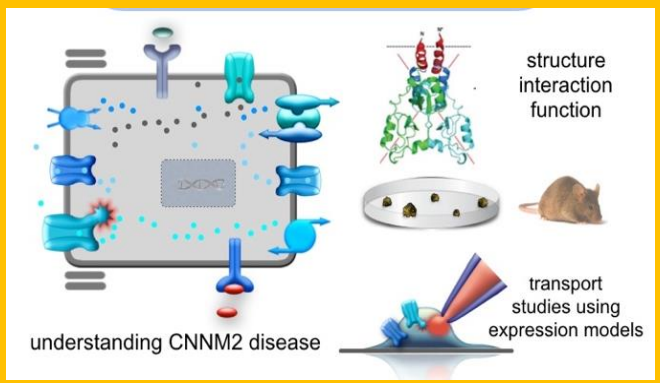


KEY OBJECTIVES

CNNM2 Interactome. By proteomic analysis, we will map the complete CNNM2 interactome and examine the role of interacting proteins on CNNM2 function and cell signalling. We hope to obtain important information on CNNM2 regulation that may explain the molecular defects caused by CNNM2 mutations in cell signaling.



CNNM2 Function. Using advanced cell, tissue and animal models, the CNNM2 function will be discovered to understand disturbed Mg2+ transport function in patients harboring mutations in CNNM2.



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ERKNet
The European Rare Kidney Disease Reference Network

Prof. Dr. Franz Schaefer. ERKNet coordinator

HOW CAN FIGHT-CNNM2 PROJECT HELP PATIENTS?

The improvement of the disease knowledge may open new opportunities in the future. The role of the Patient Association Organisation adds this vision to the management of the project and thus the objectives adapted to these needs.

FIGHT-CNMM2 allows to create a community for patients with this disease, where it will be possible to share experiences, ask questions, support among others. So on RareConnect a Community about Familiar Hypomagnesemia has been created, people around the world can be connected and get in touch with other patients and share data and information.

HOW CAN PATIENTS CONTRIBUTE TO FIGHT-CNNM2 PROJECT?

Dominant primary hypomagnesemia is a really rare disease and there is no registry of patients with a enough number of validate the results we obtain. The wider is the population-based of the project, the more reliable the results will be. For this reason we need to increase the number of patients.

If you are a patient with dominant primary hypomagnesemia or you are a doctor of a patient with this disease, please contact with FIGHT-CNNM2 project in order to be included and part of this project.

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