The genetic loss of Mg^{2+} transporter, "SLC41A1" causes inherited cystic kidney disease

Professor Takaaki Abe

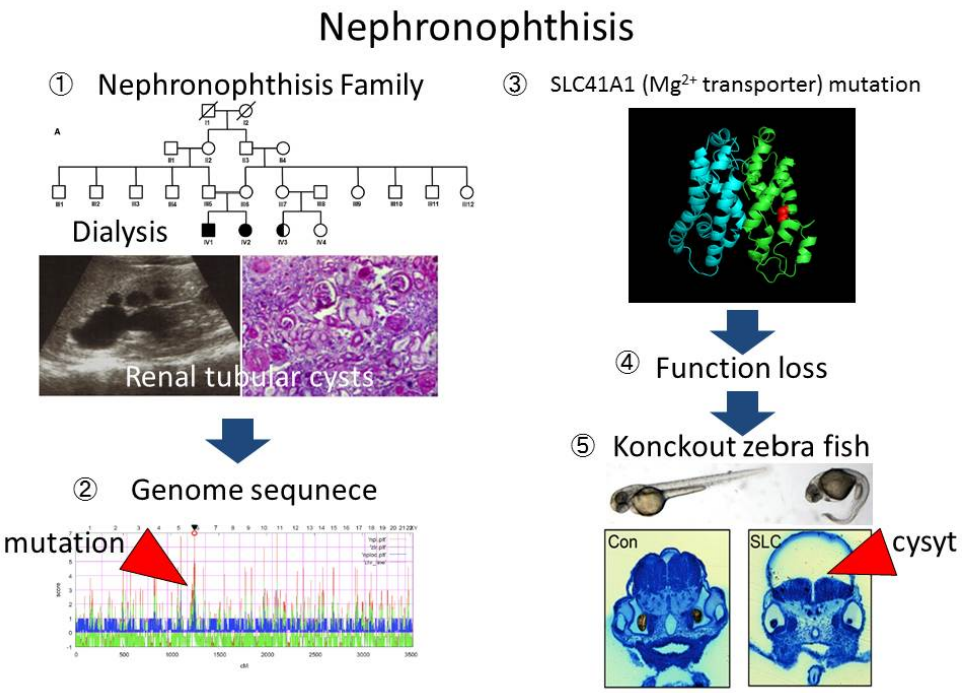
Professor ABE, Takaaki
Clinical Bilology and Hormonal Regulation
takaabe@med.tohoku.ac.jp
http://www.med.tohoku.ac.jp/english/org/cooperate/136/

The research group consisting of Professor Takaaki Abe of the Graduate School of Biomedical Engineering Regenerative and Biomedical Engineering, Medical Science, Humoral Pathology Control Medicine at Tohoku University School of Medicine, Professor Hildebrant of the University of Michigan, and Professor Masato Konishi of the Tokyo Medical University discovered that genetic mutation in Mg^{2+} transporter, "SLC41A1" causes congenital cystic kidney disease as nephronophthisis.

Nephronophthisis is the most common in hereditary juvenile kidney diseases and there are no known existing fundamental medical treatments, causing it to be the primary cause for the use of kidney dialysis or transplant treatments as the disease progresses.

These results indicate that the reduced function of Mg^{2+} transport causes structural abnormalities in the renal tubules and subsequent renal dysfunction followed by hemodialysis.
This is an important discovery in development of therapies to treat cystic kidney diseases including nephronophthisis and in understanding its pathophysiology.

The results of this research were published in the electronic edition of the American nephrological journal, the Journal of the American Society of Nephrology.



Mutation of the Mg^{2+} Transporter SLC41A1 Results in a Nephronophthisis-Like Phenotype and causes dialysis in childhood.

"Mutation of the Mg^{2+} Transporter SLC41A1 Results in a Nephronophthisis-Like Phenotype."
Hurd TW, Otto EA, Mishima E, Gee HY, Inoue H, Inazu M, Yamada H, Halbritter J, Seki G, Konishi M, Zhou W, Yamane T, Murakami S, Caridi G, Ghiggeri G, Abe T, Hildebrandt F.
J Am Soc Nephrol. 2013 May;24(6):967-77. doi: 10.1681/ASN.2012101034. Epub 2013 May 9.
PMID: 23661805 [PubMed - in process] PMCID: PMC3665398[Available on 2014/5/31]