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Research Details :

Research Title : *CLDN16 Genotype Predicts Renal Decline in Familia with Hypercalciuria and phrocalcinosisl*

CLDN16 Genotype Predicts Renal Decline in Familia with Hypercalciuria and phrocalcinosisl

Descriptipn : Familial hypomagnesemia with hypercalciuria and nephrocalcinosis (FHHNC) is a rare autosomal recessive tubular disorder caused by CLDN16 mutations. CLDN16 encodes the renal tight junction protein claudin-16, which is important for the paracellular reabsorption of calcium and magnesium in the thick ascending limb of Henle's loop. That FHHNC is frequently associated with progressive renal failure suggests additional roles for claudin-16 in the maintenance of tight junction integrity. An investigation of 32 patients with FHHNC and 17 different mutations was previously reported; here, the analysis is expanded to 39 additional patients and 12 new mutations. Expression studies revealed that five of the 12 new mutations led to partial loss of claudin-16 function and the remaining seven led to complete loss of function. The 23 patients who had mutations resulting in complete loss of function of both alleles were significantly younger at the onset of symptoms than the 46 patients who had at least one mutant allele providing partial function (2.2 versus 5.6 years; P 0.01). In addition, those with complete loss of function had a more rapid decline in GFR (7.3 versus 2.9 ml/min per 1.72 m2/y; P 0.01), leading to 54% requiring renal replacement therapy by age 15 compared with 20% of those with residual function (P 0.05). These data suggest that residual function of claudin-16 may delay the progression of renal failure in FHHNC

Research Type : Article

Added Date : Sunday, March 16, 2008

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Attatchments :

File Name	Type	Description
j am soc nephrol 2008 p171.pdf	pdf	مشاهدة المقالة العلمية كاملة