IQ Motif-Containing Protein B1

Alternative Names
IQCB1
Nephrocystin 5
NPHP5
p53- and DNA Damage-Regulated IQ Motif Protein
PIQ
KIAA0036

Record Category
Gene locus

WHO-ICD
N.B.: Classification not applicable to gene loci.

Incidence per 100,000 Live Births
N/A to gene loci

OMIM Number
609237

Mode of Inheritance
N/A

Gene Map Locus
3q13.33

Description

The IQCB1 gene, also called NPHP5 encodes a protein called nephrocystin that is involved in ciliogenesis. The encoded protein is localized to the primary cilia of renal epithelial cells and connecting cilia of photoreceptor cells. It has a central coiled-coil region and two calmodulin-binding IQ domains and interacts with calmodulin and the retinitis pigmentosa GTPase regulator protein.

Defects in this protein are the cause of Senior-Loken syndrome 5 (SLSN5), characterized by nephronophthisis and retinal degeneration.

Molecular Genetics

The IQCB1 gene spans 65,676 bp on the long arm of chromosome 3. It consists of 15 exons, of which exon 1 and 2 are not translated. The full-length IQCB1 mRNA sequence encodes 598 amino acids with a molecular weight of 69 kDa.

Mutations in this gene have been associated with Senior-Loken syndrome (SLSN) and Leber congenital amaurosis.

Epidemiology in the Arab World

Saudi Arabia

Wang et al. (2011) performed homozygosity mapping, whole-exome sequencing and direct sequencing to identify mutations affecting a collection of six consanguineous Saudi families with Leber congenital amaurosis (LCA). There were 14 affected members who had vision defects since birth or as early as two years of age. A total of five disease-causing mutations located on four genes were identified in the six families. One of these was a novel nonsense mutation (c.1479C>A, p.Y493X) in the IQCB1 gene. This novel variant was rare and was absent in 200 normal matching controls. In addition, it had not been recorded in the dbSNP 130 database and the 1,000 Genome database. Patients with IQCB1 gene mutations had typical LCA phenotypes including; nystagmus, nonrecordable ERGs, and other visual defects.

References
Wang X, Wang H, Cao M, Li Z, Chen X, Patenia C, Gore A, Abboud EB, Al-Rajhi AA, Lewis RA, Lupski JR, Mardon G, Zhang K, Muzny D, Gibbs RA, Chen R. Whole-exome sequencing identifies ALMS1, IQCB1, CNGA3, and MYO7A mutations in patients with Leber congenital amaurosis. Hum Mutat. 2011; 32(12):1450-9. PMID: 21901789

Related CTGA Records
Leber Congenital Amaurosis 1

External Links
http://www.genecards.org/cgi-bin/carddisp.pl?gene=IQCB1
https://ghr.nlm.nih.gov/gene/IQCB1

Contributors