Tectonic Family, Member 2

Alternative Names
TCTN2
Tectonic 2
TECT2

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
613846

Mode of Inheritance

Gene Map Locus
12q24.31

Description
The TCTN2 gene encodes a type I membrane protein that belongs to the tectonic family. This gene is expressed in all tissues, with significant expression in brain, kidney, and eye. Studies in mice suggest that this protein may be involved in hedgehog signaling, and is essential for ciliogenesis.

Mutations in this gene are associated with Meckel syndrome type 8 and Joubert syndrome type 24.

Molecular Genetics
The TCTN2 gene is located on the long (q) arm of chromosome 12 at position 24.31. It spans 37,293 bases and contains 18 exons. The deduced mouse Tctn2 protein contains 700 amino acids. It has an N-terminal signal peptide and a C-terminal transmembrane domain that is conserved in Drosophila tectonic. A splice site mutation in the TCTN2 gene was identified in one family. Different homozygous mutations in the TCTN2 gene have been identified as well.

Epidemiology in the Arab World

Saudi Arabia

Shaheen et al. (2011) studied a consanguineous family of Arab origin affected with Meckel Syndrome. Homozygosity scan in the first family revealed the presence of only one overlapping ROH of 7Mb in size on chromosome 12. A splice site mutation (c.1506_2A4G; NM_024809.3) in the TCTN2 gene was identified in this family, which was is predicted in silico to fully abolish the original acceptor. This mutation was found to segregate with the disease in the family and was not observed in 192 ethnically matched controls. This defined TCTN2 as a novel MKS locus.

References
Shaheen R, Faqeih E, Seidahmed MZ, Sunker A, Alali FE, AlQahtani K, Alkuraya FS. A TCTN2 mutation defines a novel Meckel Gruber syndrome locus. Hum Mutat. 2011; 32(6):573-8. PMID: 21462283

Related CTGA Records
Meckel Syndrome 8

External Links
http://www.genecards.org/cgi-bin/carddisp.pl?gene=TCTN2

Contributors
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