Chromosome 17 Open Reading Frame 28

Alternative Names
C17ORF28
Downregulated in Multiple Cancers 1
DMC1
HID1

Record Category
Gene locus

WHO-ICD
N/A to gene loci

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

OMIM Number
605752

Mode of Inheritance
N/A to gene loci

Gene Map Locus
17q25.1

Description
The C17ORF28 gene encodes an integral membrane protein belonging to the 'inside-out' class of membrane proteins. It consists of a cytoplasmic domain at its N-terminus, a single transmembrane peptide and an extracellular domain at its C-terminus. It is also found in the cytoplasm where it shuttles between the Golgi apparatus and the cytosol. As expression of the gene is seen to be reduced in several cancer cell lines, it has been suggested to play a role in tumor suppression. The gene is thus also designated as DMC1 or 'Downregulated in Multiple Cancers-1'.

While the C17ORF28 gene is yet to be fully characterized, recent studies on lower orthologs have identified its role in the peptidergic signaling pathway, intracellular trafficking within the Golgi region and maturation of secretory granules generated at the trans-Golgi network.

Molecular Genetics
The C17ORF28 gene, located on the long arm of chromosome 17, spans a length of just 22.4 kb of DNA. Its coding sequence is spread across 19 exons and it encodes an 88.7 kDa protein product composed of 788 amino acids. Two additional isoforms of the C17ORF28 protein exist due to alternatively spliced transcript variants. While the gene is expressed in the heart, skeletal muscle, colon, spleen, kidney, liver, small intestine and lung, highest expression is seen in the brain and placenta.

Epidemiology in the Arab World
Saudi Arabia
Monies et al. (2017) discussed the findings of 1000 diagnostic panels and exomes carried out at a next generation sequencing lab in Saudi Arabia. One patient, a 2-year-old male, presented with a failure to thrive, growth retardation, intellectual disability, agenesis of the corpus callosum, hypotonia and panhypopituitarism. The family also reported a similar phenotype in several male maternal cousins, indicating a possible X-linked inheritance. Two of his cousins suffering from hypoglycemia, central hypothyroidism, central adrenal insufficiency and post meningitis had both succumbed before being diagnosed and he had lost a brother at six months of age. Using whole exome sequencing, a homozygous mutation (c.2318dupC, p.P773fs) was identified in exon 19 of the patient's C17ORF28 (HID1) gene. This gene mutation was considered a candidate for pathogenicity as it was a novel variant located within the autozygome and was predicted to be deleterious; and the C17ORF28 (HID1) protein was known to be required for homotypic fusion of immature secretory granules during maturation. The authors hoped that further studies would help confirm this association.

References

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Related CTGA Records

External Links

http://www.genecards.org/cgi-bin/carddisp.pl?gene=HID1

Contributors

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