tRNA Methyltransferase 1, S. Cerevisiae, Homolog of

Alternative Names
TRMT1
N2,N2-Dimethylguanosine-26 tRNA Methyltransferase
tRNA(m(2,2)G26)Dimethyltransferase

Record Category
Gene locus

WHO-ICD
N/A to gene loci

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

OMIM Number
611669

Mode of Inheritance
N/A to gene loci

Gene Map Locus
19p13.13

Description
The TRMT1 gene encodes a methyltransferase enzyme that acts on tRNA. This enzyme, which consists of a zinc finger motif and an arginine/proline rich region at its C-terminus, is responsible for catalyzing the dimethylation of a guanine residue located at position 26 of most tRNAs. TRMT1 uses S-adenosyl-L-methionine as a substrate and has both mono and dimethylase activity.

Recent studies have suggested a possible link between TRMT1 gene mutations and Autosomal Recessive Intellectual Disability (ARID). Patients with TRMT1 mutations have been reported to suffer from moderate to severe intellectual disability, mild facial dysmorphia, progressive spasticity of the upper and lower limbs and pes planus. These findings are further bolstered by the fact that mutations in two other RNA-methyltransferase genes, NSUN2 and FTSJ1, have been associated with intellectual disability.

Molecular Genetics
The TRMT1 gene is located on the short arm of chromosome 19 and spans a length of 12.6 kb of DNA. Its coding sequence is spread across 18 exons and it encodes a 72.2 kDa protein product comprised of 659 amino acids. An additional 69.3 kDa isoform of the TRMT1 protein exists due to an alternatively spliced transcript variant. The gene is widely expressed in the human body, particularly in the nervous system, intestine, spleen, kidney and lung.

Epidemiology in the Arab World
Saudi Arabia
Monies et al. (2017) studied the findings of 1000 diagnostic panels and exomes carried out at a next generation sequencing lab in Saudi Arabia. One patient, a 13-year-old male, presented with speech delay, intellectual disability, learning disability, hypotonia and seizures. Using whole exome sequencing, a homozygous mutation (c.1245_1246del, p.L415fs) was identified in exon 10 of the patient's TRMT1 gene. As a mutation in the gene had previously been tentatively linked to intellectual disability, this finding helped confirm its association with the disorder.

References
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Related CTGA Records

External Links

http://www.genecards.org/cgi-bin/carddisp.pl?gene=TRMT1

Contributors

Sayeeda Hana
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