Zinc Finger, MYM-Type 5

Alternative Names
ZMYM5
Zinc Finger Protein 237
ZNF237
Zinc Finger Protein 198-Like 1
ZNF198L1

Record Category
Gene locus

WHO-ICD
N/A to gene loci

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

OMIM Number
616443

Mode of Inheritance
N/A to gene loci

Gene Map Locus
13q12.11

Description
The ZMYM5 gene encodes Zinc Finger, MYM-Type 5, a protein belonging to the MYM (myeloproliferative and mental retardation motif) family. The protein binds to DNA and functions as a DNA templated, RNA polymerase II transcription factor. Specifically, the protein has been found to bind to the transcription factor ERM and repress the expression of the PS1 reporter gene by attaching to the PS1 promoter region.

Molecular Genetics
The ZMYM5 gene is located on the long arm of chromosome 13 at position 13q12.11. The gene spans a length of 40 kb and its coding sequence is spread across 9 exons. The protein product encoded by ZMYM5 has a molecular mass of 74.8 kDa and consists of 669 amino acids. Several additional isoforms of the ZMYM5 protein exist due to alternative splicing. While the gene is expressed in the spleen, thymus, prostate, testis, ovary, small intestine, colon and peripheral blood leukocytes, highest expression is seen in the bone.

Epidemiology in the Arab World
Saudi Arabia
Anazi et al. (2016) described the effectiveness of genomic tools as a first-tier test in the diagnosis of Intellectual Disability (ID) cases. A cohort of 337 ID patients were subjected to molecular karyotyping, exome sequencing and a multi-gene panel comprised of neurologically associated genes. Genomic tools were found to have a higher diagnostic yield than standard clinical evaluations (58% vs 16%). This approach helped discover a homozygous c.362_363insT (p.Asp33Valfs*13) mutation in the ZMYM5 gene of a 7 year old girl. The patient suffered from global developmental delay, dysmorphic features, bone lytic lesions and anhidrosis. The authors noted that ZMYM5 regulates the expression of PSEN1, a gene associated with dementia and Alzheimer's.

References
Anazi S, Maddirevula S, Faqeih E, Alsedairy H, Alzahrani F, Shamseldin HE, Patel N, Hashem M, Ibrahim N, Abdulwahab F, Ewida N, Alsaif HS, Al Sharif H, Alamoudi W, Kentab A, Bashiri FA, Alnaser M, AlWadei AH, Alfadhel M, Eyaid W, Hashem A, Al Asmari A, Saleh MM, AlSaman A, Alhasan KA, Alsughayir M, Al Shammari M, Mahmoud A, Al-Hassnan ZN, Al-Husain M, Osama Khalil R, Abd El Meguid N, Masri A, Ali R, Ben-Omran T, El Fishway P, Hashish A, Ercan Sencicek A, State M, Alazami AM, Salih MA, Altassan N, Arold ST, Abouelhoda M, Wakil SM, Monies D, Shaheen R, Alkuraya FS. Clinical genomics expands the morbid genome of intellectual disability and offers a high diagnostic yield. Mol Psychiatry. 2016 Jul 19. PMID: 27431290.

Related CTGA Records

External Links
http://www.genecards.org/cgi-bin/carddisp.pl?gene=ZMYM5

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