Tyrosinase-Related Protein 1

Alternative Names
TYRP1
TYRP
TRP
Catalase B
CATB
CAS2
Glycoprotein 75
GP75
b-Protein

Record Category
Gene locus

WHO-ICD
N/A to gene loci

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

OMIM Number
115501

Mode of Inheritance
N/A to gene loci

Gene Map Locus
9p23

Description
The TYRP1 gene encodes Tyrosinase-Related Protein 1, an oxidoreductase enzyme found specifically in melanocyte cells. These cells are responsible for producing melanin, the pigment that gives skin, hair and eyes their color. The TYRP1 protein is involved in the biosynthesis of melanin, known as melanogenesis by playing a role in stabilizing the tyrosinase enzyme and in the organization of melanosomes, the organelles in which melanin is packaged.

Mutations in the gene have been linked to Oculocutaneous Albinism, type III, a brown albinism disorder characterized by hypopigmentation in light-skinned individuals and bright copper-red coloration of the skin and hair in dark-skinned individuals.

Molecular Genetics
The TYRP1 gene is located on the short arm of chromosome 9. It spans a length of 24.8 kb of DNA and its coding sequence is spread across 8 exons. The protein product encoded by this gene has a molecular mass of 60.7 kDa and consists of 537 amino acids. TYRP1 is found to be overexpressed in the skin, heart and serum. Homozygous TYRP1 mutations, including deletions, missense and nonsense variants have been implicated in Oculocutaneous Albinism, type III.

Epidemiology in the Arab World
Saudi Arabia
Monies et al. (2017) illustrated the genomic landscape of Saudi Arabia based on the findings of 1000 diagnostic panels and exomes. One patient, an 11-year-old male, suffered from hemimegalencephaly, developmental delay and ADHD. He also had abnormal pigmentation all over his body. Whole exome sequencing helped identify a dual molecular diagnosis in this patient. A heterozygous mutation (c.1557T>G, p.Y519X) was found in exon 8 of the patient's TYRP1 gene, associated with oculocutaneous albinism type 3, and a heterozygous variant (c.90G>A, p.W30X) was uncovered in exon 1 of the TGIF1 gene, associated with holoprosencephaly 4. Such dual molecular diagnoses were rare and only occurred in 1.5% of the cohort.

References
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Related CTGA Records

Albinism, Oculocutaneous, Type III (OMIM 203290)

External Links

http://www.genecards.org/cgi-bin/carddisp.pl?gene=TYRP1

Contributors

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