Adiponectin Receptor 1

Alternative Names
ADIPOR1
CGI45
Progestin and AdipoQ Receptor Family, Member 1
PAQR1

Record Category
Gene locus

WHO-ICD
N/A to gene loci

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

OMIM Number
607945

Mode of Inheritance
N/A to gene loci

Gene Map Locus
1q32.1

Description
Adiponectin is a hormone secreted by adipocytes responsible for metabolizing fatty acids and maintaining glucose homeostasis. It has thus been identified as a key anti-diabetic, anti-atherogenic and anti-inflammatory factor. Adiponectin carries out its functions by binding to the adiponectin receptor, encoded by the ADIPOR1 gene. Binding of the globular or full-length adiponectin to the ADIPOR1 receptor triggers the AMP-activated kinase signaling pathway, thereby resulting in increased levels of fatty acid catabolism, enhanced glucose utilization and reduced gluconeogenesis.

There have been individual reports of ADIPOR1 gene mutations in patients with Syndromic Retinitis Pigmentosa as well as Intellectual Disability. However, a direct causal link between these disorders and ADIPOR1 mutations has not yet been established.

Molecular Genetics
The ADIPOR1 gene is located on the long arm of chromosome 1. It spans a length of 17.7 kb of DNA and its coding sequence is spread across 11 exons. The gene encodes a 42.6 kDa protein product comprised of 375 amino acids. While the gene is widely expressed, highest expression is seen in the heart and skeletal muscle, followed by the brain, spleen, kidney, liver, placenta, lung and peripheral blood leukocytes.

Epidemiology in the Arab World
Saudi Arabia
Monies et al. (2017) outlined the genomic landscape of Saudi Arabia based on the findings of 1000 diagnostic panels and exomes. One patient, a 13-year-old female, suffered from microcephaly, fine/gross motor delay, speech delay, intellectual disability, learning disability, autistic features, ataxia, high blindness and retinitis pigmentosa. Whole exome sequencing helped identify a dual molecular diagnosis in this patient. A homozygous mutation (c.1898C>T, p.T633M) was found in exon 5 of the patient's CRB1 gene, associated with retinitis pigmentosa 12, and a homozygous variant (c.346C>A, p.P116T) was uncovered in exon 4 of the ADIPOR1 gene, associated with intellectual disability (ID). Such dual molecular diagnoses were rare and only occurred in 1.5% of the cohort. Further, as the ADIPOR1 gene had previously been tentatively linked to ID, this report helped confirm its association with the disorder.

References
Monies D, Abouelhoda M, AlSayed M, Alhassnan Z, Alotaibi M, Kayyali H, Al-Owain M, Shah A, Rahbeeni Z, Al-Muhaizea MA, Alzaidan HI, Cupler E, Bohlega S, Faqeih E, Faden M, Alyounes B, Jaroudi D, Goljan E, Elbardisy H, Akilan A,

Albar R, Aldhalaan H, Gulab S, Chedrawi A, Al Saud BK, Kurdi W, Makhseed N, Alqasim T, El Khashab HY, Al-Mousa H, Alhashem A, Kanaan I, Algoufi T, Alsaleem K, Basha TA, Al-Murshedi F, Khan S, Al-Kindy A, Alnemer M, Al-Hajjar S, Alyamani S, Aldhekri H, Al-Mehaidib A, Arnaout R, Dabbagh O, Shagrani M, Broering D, Tulbah M, Alqassmi A, Almugbel M, AlQuaiz M, Alsaman A, Al-Thihli K, Sulaiman RA, Al-Dekhail W, Alsaegh A, Bashiri FA, Qari A, Alhomadi S, Alkuraya H, Alsebayel M, Hamad MH, Szonyi L, Abaalkhail F, Al-Mayouf SM, Almojalli H, Alqadi KS, Elsiesy H, Shuaib TM, Seidahmed MZ, Abosoudah I, Akleh H, AlGhonaium A, Alkharfy TM, Al Mutairi F, Eyaid W, Alshanbary A, Sheikh FR, Alsohaibani FI, Alsonbul A, Al Tala S, Balkhy S, Bassiouni R, Alenizi AS, Hussein MH, Hassan S, Khalil M, Tabarki B, Alshahwan S, Oshi A, Sabr Y, Alsaadoun S, Salih MA, Mohamed S, Sultana H, Tamim A, El-Haj M, Alshahrani S, Bubshait DK, Alfadhel M, Faquih T, El-Kalioby M, Subhani S, Shah Z, Moghrabi N, Meyer BF, Alkuraya FS. The landscape of genetic diseases in Saudi Arabia based on the first 1000 diagnostic panels and exomes. Hum Genet. 2017 Aug;136(8):921-939. PMID: 28600779.

Related CTGA Records

Adipocyte-, C1q-, and Collagen Domain-Containing (OMIM 605441)

External Links

http://www.genecards.org/cgi-bin/carddisp.pl?gene=ADIPOR1

Contributors

Sayeeda Hana
15.09.2017