TRAF3-Interacting Protein 2

Alternative Names
TRAF3IP2
Chromosome 6 Open Reading Frame 5
C6ORF5
Nuclear Factor Kappa-B Activator 1
ACT1
Connection To I-Kappa-B Kinase and Stress-Activated Protein Kinase
CIKS
Chromosome 6 Open Reading Frame 4
C6ORF4
Chromosome 6 Open Reading Frame 6
C6ORF6

Record Category
Gene locus

WHO-ICD
N/A to gene loci

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

OMIM Number
607043

Mode of Inheritance
N/A to gene loci

Gene Map Locus
6q21

Description
The TRAF3IP2 gene encodes the intracellular TRAF3-Interacting Protein 2. This protein plays an important role in the cellular response to inflammation, viral pathogens and stress. As the name suggests, it interacts with TRAF proteins (tumor necrosis factor receptor-associated factors) along with I-kappaB kinase to trigger the I-kappaB kinase/NF-kappaB signaling cascade. It is also involved in the activation of stress-activated protein kinase (SAPK)/JNK. TRAF3IP2 contains a helix-loop-helix domain at the N-terminus, which is essential for the activation of NF-kappaB, and a coiled-coil domain at the C-terminus which is involved in SAPK/JNK activation.
Homozygous mutations in the TRAF3IP2 gene have been implicated in Familial Candidiasis 8, a chronic mucocutaneous disorder characterized by recurrent infections with C. albicans. A coding SNP in the gene has also been associated with an increased susceptibility to Psoriasis 13, a skin condition characterized by raised, scaly, red patches often seen on the scalp, elbows and knees. This condition may also be associated with severe arthritis.

Molecular Genetics
The TRAF3IP2 gene is located on the long arm of chromosome 6 and spans a length of 51.4 kb of DNA. Its coding sequence is spread across 12 exons and it encodes a 64.6 kDa protein product comprised of 574 amino acids. Several additional isoforms of the TRAF3IP2 protein exist due to alternatively spliced transcript variants. One non-protein coding isoform, designated C6UAS, is transcribed in the anti-sense orientation and has been suggested to play a regulatory role in TRAF3IP2 gene expression. The gene is widely expressed in the human body. The homozygous missense mutation p.Thr536Ile has been associated with Familial Candidiasis 8.

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 6-year-old male patient suffering from severe eczema was subjected to a multigene panel for immunity disorders and was found to carry a homozygous mutation (c.200G>C:p.W67S) in exon 4 of the TRAF3IP2 gene. As a mutation in the gene had

previously been tentatively linked to eczema, this finding 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

External Links

http://www.genecards.org/cgi-bin/carddisp.pl?gene=TRAF3IP2

Contributors

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