Glutamate Receptor, Ionotropic, Kainate 4

Alternative Names
GRIK4
KA1

Record Category
Gene locus

WHO-ICD
N/A to gene loci

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

OMIM Number
600282

Mode of Inheritance
N/A to gene loci

Gene Map Locus
11q23.3

Description
The GRIK4 gene encodes a protein that forms a functional heteromeric ligand-gated ionic channel along with the subunits encoded by the GRIK1, GRIK2 and GRIK3 genes. This channel functions as a kainate receptor which is activated by the major excitatory neurotransmitter, glutamate. By carrying out its function, the GRIK4 kainate receptor contributes to excitatory post-synaptic currents in the central nervous system.

Recent studies in evolutionary biology have further implicated the role of GRIK4 in the development of the nervous system. This was indicated by the significantly higher rates of GRIK4 protein evolution in primates than in rodents. The acceleration of GRIK4 evolution was particularly pronounced in the lineage leading from ancestral primates to humans, which correlates with the phenotypic evolution of the human nervous system into a larger and more complex structure.

Molecular Genetics
The GRIK4 gene is located on the long arm of chromosome 11. It spans a length of 477.2 kb of DNA and its coding sequence is spread across 24 exons. The gene encodes a 107.2 kDa protein product consisting of 956 amino acids. GRIK4 is found to be expressed in the nervous system and kidney.

Epidemiology in the Arab World
Saudi Arabia
Monies et al. (2017) depicted the genomic landscape of Saudi Arabia based on the findings of 1000 diagnostic panels and exomes. One patient, a 5-year-old female, presented with microcephaly and developmental regression. She had a niece with severe global developmental delay (GDD) and a sister with GDD that had died at 3 years of age. Using whole exome sequencing, a homozygous mutation (c.2479T>G, p.F827V) was identified in exon 19 of the patient's GRIK4 gene. This gene mutation was considered a candidate for pathogenicity as it was a novel variant located within the autozygome that was predicted to be deleterious, and the gene had been implicated in the pathogenesis of autism. The authors noted that further studies were required to independently confirm this association.

References
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Related CTGA Records

External Links

http://www.genecards.org/cgi-bin/carddisp.pl?gene=GRIK4

Contributors

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