DOK7 gene

docking protein 7

Normal Function

The *DOK7* gene provides instructions for making a protein that is necessary for the formation of connections between nerve cells and muscle cells, which occur in the neuromuscular junction. The neuromuscular junction is the area between the ends of nerve cells and muscle cells where signals are relayed to trigger muscle movement. The Dok-7 protein participates in turning on (activating) a protein called MuSK that plays a key role in organizing the various proteins important for the development and maintenance of the neuromuscular junction. In particular, the MuSK protein is involved in concentrating a protein called the acetylcholine receptor (AChR) in the muscle membrane at the neuromuscular junction." The AChR protein is critical for signaling between nerve and muscle cells, which is necessary for movement.

Health Conditions Related to Genetic Changes

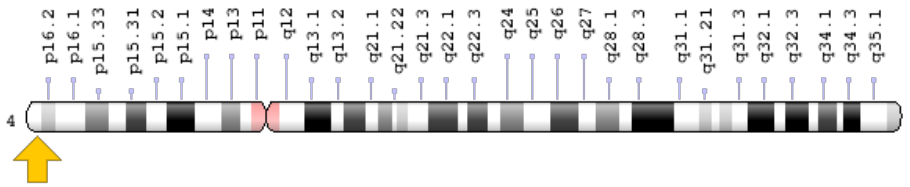
Congenital myasthenic syndrome

At least 45 mutations in the *DOK7* gene have been found to cause congenital myasthenic syndrome. A mutation that frequently occurs is the addition of four DNA building blocks (nucleotides) in the *DOK7* gene (written as 1124_1127dupTGCC). Mutations in this gene lead to the production of a defective Dok-7 protein that cannot activate the MuSK protein. As a result, less AChR is present in the neuromuscular junction, which reduces signaling between nerve and muscle cells. These signaling abnormalities lead to decreased muscle movement and the muscle weakness characteristic of congenital myasthenic syndrome. For reasons that are unclear, people with mutations in the *DOK7* gene tend to have muscle weakness in the shoulders, hips, and limbs, known as limb-girdle muscle weakness.

Chromosomal Location

Cytogenetic Location: 4p16.3, which is the short (p) arm of chromosome 4 at position 16.3

Molecular Location: base pairs 3,463,306 to 3,501,476 on chromosome 4 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- C4orf25
- CMS1B
- Dok-7
- DOK7_HUMAN
- downstream of tyrosine kinase 7

Additional Information & Resources

Educational Resources

- Molecular Cell Biology (fourth edition, 2000): Motor Neurons Induce Assembly of the Neuromuscular Junction
  https://www.ncbi.nlm.nih.gov/books/NBK21742/#A6877the
- Washington University, St. Louis: Neuromuscular Disease Center
  https://neuromuscular.wustl.edu/synmg.html#lgm

Clinical Information from GeneReviews

- Congenital Myasthenic Syndromes
  https://www.ncbi.nlm.nih.gov/books/NBK1168

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28DOK7%5BTIAB%5D%29+OR+%28Dok-7%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- DOWNSTREAM OF TYROSINE KINASE 7
  http://omim.org/entry/610285

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_DOK7.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=DOK7%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:26594
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:285489
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/285489
- UniProt
  https://www.uniprot.org/uniprot/Q18PE1

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