

PARK7 gene

Parkinsonism associated deglycase

Normal Function

The *PARK7* gene provides instructions for making the DJ-1 protein. This protein is found in many tissues and organs, including the brain. Studies indicate that the DJ-1 protein has several functions, although none are fully understood. One of the protein's functions may be to help protect cells, particularly brain cells, from oxidative stress. Oxidative stress occurs when unstable molecules called free radicals accumulate to levels that can damage or kill cells. Additionally, the DJ-1 protein may serve as a chaperone molecule that helps fold newly produced proteins into the proper 3-dimensional shape and helps refold damaged proteins. Like other chaperone molecules, the DJ-1 protein may assist in delivering selected proteins to proteasomes, which are structures within cells that break down unneeded molecules. Researchers suggest that the DJ-1 protein may also play a role in activities that produce and process RNA, a chemical cousin of DNA.

Health Conditions Related to Genetic Changes

Parkinson's disease

Researchers have identified more than 25 *PARK7* gene variants (also called mutations) that can cause Parkinson's disease, a condition characterized by progressive problems with movement and balance. These variants are associated with the early-onset form of the disorder, which begins before age 50. Some *PARK7* gene variants lead to an abnormally small DJ-1 protein or change the building blocks (amino acids) used to make the protein. The altered protein is unstable and does not function properly, if at all. Other variants delete a large portion of the *PARK7* gene, preventing the production of any functional DJ-1 protein.

It is unclear how loss of functional DJ-1 protein leads to Parkinson's disease. Some studies suggest that *PARK7* gene variants disrupt the protein's chaperone function, which leads to a toxic buildup of misfolded or damaged proteins and eventually to cell death. Another possibility is that *PARK7* gene variants impair the protein's ability to protect cells from destructive oxidative stress. Nerve cells that make the chemical messenger dopamine are particularly vulnerable to oxidative stress. With diminished protection, free radicals may cause enough damage to kill these nerve cells. Progressive loss of dopamine-producing nerve cells is a characteristic feature of

Parkinson's disease. The death of these cells weakens communication between the brain and muscles, and ultimately the brain becomes unable to control muscle movement.

Other Names for This Gene

- DJ-1
- DJ1
- PARK7_HUMAN
- Parkinson disease (autosomal recessive, early onset) 7
- parkinson protein 7

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PARK7 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=11315\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=11315[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28PARK7%5BTIAB%5D%29+OR+%28DJ1%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%29%29%29>)

Catalog of Genes and Diseases from OMIM

- ONCOGENE DJ1; DJ1 (<https://omim.org/entry/602533>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/11315>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=PARK7\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=PARK7[gene]))

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Genomic Location

The *PARK7* gene is found on chromosome 1 (<https://medlineplus.gov/genetics/chromosome/1/>).

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