



NCF4 gene

neutrophil cytosolic factor 4

Normal Function

The *NCF4* gene provides instructions for making a protein called neutrophil cytosolic factor 4 (also known as p40-phox). This protein is one part (subunit) of a group of proteins that forms an enzyme complex called NADPH oxidase, which plays an essential role in the immune system. Specifically, NADPH oxidase is primarily active in immune system cells called phagocytes. These cells catch and destroy foreign invaders such as bacteria and fungi. NADPH oxidase is also thought to regulate the activity of immune cells called neutrophils. These cells play a role in adjusting the inflammatory response to optimize healing and reduce injury to the body.

The presence of foreign invaders stimulates phagocytes and triggers the assembly of NADPH oxidase. This enzyme participates in a chemical reaction that converts oxygen to a toxic molecule called superoxide. Superoxide is used to generate several other compounds, including hydrogen peroxide (a strong disinfectant) and hypochlorous acid (the active ingredient in bleach). These highly reactive, toxic substances are known as reactive oxygen species. Phagocytes use these substances to kill foreign invaders, preventing them from reproducing in the body and causing illness.

Health Conditions Related to Genetic Changes

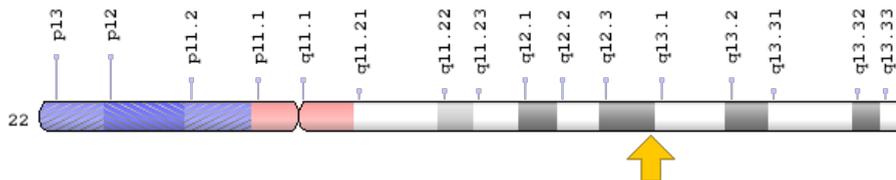
Chronic granulomatous disease

At least two mutations in the *NCF4* gene have been found to cause chronic granulomatous disease. People with this disorder are at increased risk of developing recurrent episodes of infection and inflammation due to a weakened immune system. Mutations in the *NCF4* gene are a rare cause of this condition. These mutations result in a decrease in functional NADPH oxidase. A shortage (deficiency) of this protein impairs the assembly or function of NADPH oxidase. As a result, phagocytes are unable to produce reactive oxygen species to kill foreign invaders, and neutrophil activity is not regulated. A lack of NADPH oxidase leaves affected individuals vulnerable to many types of infection and excessive inflammation.

Chromosomal Location

Cytogenetic Location: 22q12.3, which is the long (q) arm of chromosome 22 at position 12.3

Molecular Location: base pairs 36,860,988 to 36,878,017 on chromosome 22 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- NCF
- NCF-4
- NCF4_HUMAN
- neutrophil cytosol factor 4
- neutrophil cytosolic factor 4, 40kDa
- neutrophil NADPH oxidase factor 4
- p40-phox
- P40PHOX

Additional Information & Resources

Educational Resources

- Immunobiology: The Immune System in Health and Disease (2001, fifth edition): After entering tissues, many pathogens are recognized, ingested, and killed by phagocytes
<https://www.ncbi.nlm.nih.gov/books/NBK27105/#A156>
- Immunobiology: The Immune System in Health and Disease (2001, fifth edition): Defects in phagocytic cells permit widespread bacterial infections
<https://www.ncbi.nlm.nih.gov/books/NBK27109/#A1507>

Clinical Information from GeneReviews

- Chronic Granulomatous Disease
<https://www.ncbi.nlm.nih.gov/books/NBK99496>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28NCF4%5BTIAB%5D%29+OR+%28p40phox%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+1800+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- NEUTROPHIL CYTOSOLIC FACTOR 4
<http://omim.org/entry/601488>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_NCF4.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=NCF4%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:7662
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:4689>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/4689>
- UniProt
<https://www.uniprot.org/uniprot/Q15080>

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- OMIM: NEUTROPHIL CYTOSOLIC FACTOR 4
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