

IRF6 gene

interferon regulatory factor 6

Normal Function

The *IRF6* gene provides instructions for making a protein that plays an important role in early development. This protein is a transcription factor, which means that it attaches (binds) to specific regions of DNA and helps control the activity of particular genes. The IRF6 protein is active in cells that give rise to tissues in the head and face. It is also involved in the development of other parts of the body, including the skin and genitals.

Health Conditions Related to Genetic Changes

Popliteal pterygium syndrome

Variants (also known as mutations) in the *IRF6* gene cause popliteal pterygium syndrome. This condition affects the development of the face, skin, and genitals.

The *IRF6* gene variants that cause this condition may change the transcription factor's effects on the activity of certain genes. This affects the development and maturation of tissues in the face, skin, and genitals, resulting in the facial and genital abnormalities, skin webbing, and fusion of the fingers or toes (syndactyly) seen in popliteal pterygium syndrome.

Van der Woude syndrome

Variants in the *IRF6* gene cause van der Woude syndrome. This condition affects the development of the face and often causes cleft lip, cleft palate (an opening in the roof of the mouth, or both). *IRF6* gene variants that cause this condition prevent one copy of the gene in each cell from making any functional protein. A shortage of the IRF6 protein affects the development and maturation of tissues in the skull and face. These abnormalities underlie the signs and symptoms of van der Woude syndrome.

Other disorders

Certain variations in the *IRF6* gene have been associated with increased risk of cleft lip, cleft palate, or both. When these features appear without other signs or symptoms (such as in the conditions described above), the condition is called isolated cleft lip and/or palate. The *IRF6* gene variations are believed to affect the function of the IRF6

protein in its role as a transcription factor, which may interfere with the normal development of the face.

Other Names for This Gene

- IRF6_HUMAN
- LPS
- OFC6
- PIT
- PPS
- VWS
- VWS1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of IRF6 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3664\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3664[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28IRF6%5BTIAB%5D%29+OR+%28interferon+regulatory+factor+6%5BTIAB%5D%29%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%5D>)

Catalog of Genes and Diseases from OMIM

- OROFACIAL CLEFT 6, SUSCEPTIBILITY TO; OFC6 (<https://omim.org/entry/608864>)
- INTERFERON REGULATORY FACTOR 6; IRF6 (<https://omim.org/entry/607199>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/3664>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=IRF6\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=IRF6[gene]))

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Genomic Location

The *IRF6* gene is found on chromosome 1 (<https://medlineplus.gov/genetics/chromosome>)

me/1/).

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