Van der Woude syndrome

Description

Van der Woude syndrome is a condition that affects the development of the face. Many people with this disorder are born with either a cleft lip or a cleft palate (an opening in the upper lip or roof of the mouth), or both. Affected individuals usually have depressions (pits) near the center of the lower lip, which may appear moist due to the presence of salivary and mucous glands in the pits. Small mounds of tissue on the lower lip may also occur. In some cases, people with van der Woude syndrome have missing teeth.

People with van der Woude syndrome who have cleft lip and/or palate, like other individuals with these facial conditions, have an increased risk of delayed language development, learning disabilities, or other mild cognitive problems. The average IQ of individuals with van der Woude syndrome is not significantly different from that of the general population.

Frequency

Van der Woude syndrome is believed to occur in 1 in 35,000 to 1 in 100,000 people, based on data from Europe, Asia, and Africa. Van der Woude syndrome is the most common cause of cleft lip and palate resulting from genetic variations.

Causes

Variations (also known as mutations) in the *IRF6* gene cause most cases of van der Woude syndrome. 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.

Variants in the *IRF6* gene that cause van der Woude syndrome 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 face, resulting in the signs and symptoms of van der Woude syndrome.

In about five percent of cases, van der Woude syndrome is caused by variants in the

GRHL3 gene.

Learn more about the gene associated with Van der Woude syndrome

- IRF6

Additional Information from NCBI Gene:

- GRHL3

Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. In most cases, an affected person has one parent with the condition. Occasionally, an individual who has a copy of the altered gene does not show any signs or symptoms of the disorder.

Other Names for This Condition

- Cleft lip and/or palate with mucous cysts of lower lip
- Lip-pit syndrome
- VDWS
- VWS

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Van der Woude syndrome 1 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C4551864/)

Genetic and Rare Diseases Information Center

- Van der Woude syndrome (https://rarediseases.info.nih.gov/diseases/8414/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Catalog of Genes and Diseases from OMIM

- VAN DER WOUDE SYNDROME 1; VWS1 (https://omim.org/entry/119300)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28van+der+woude+syndrome%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D)

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