Van der Woude syndrome

Other Names: Lip-pit syndrome

Van der Woude syndrome is a genetic condition that results in an increased chance for developing malformations of the lips and palate. It is caused by mutations in the IRF6 gene that makes a protein called interferon regulatory factor 6.

Characteristics of Van der Woude syndrome
Van der Woude syndrome is an inherited condition that results in an increased risk for malformations of the lips and palate. People with this disorder may be born with a split in the lip (i.e., cleft lip), a split in the roof of the mouth (i.e., cleft palate) or both. Many individuals with Van der Woude syndrome also have depressions (pits) or mounds of tissue near the center of the lower lip. As with other conditions with cleft lip and/or palate, people with Van der Woude syndrome may have missing baby and/or permanent teeth. Although there is an increased risk of delayed language development, learning disabilities, or other mild cognitive problems in children with cleft lip and/or palate from any cause, the average IQ of individuals with this condition is not significantly different from that of the general population.

Diagnosis/Testing
Most individuals with Van der Woude syndrome have a change or mutation in a gene called IRF6. This gene makes a protein that is responsible for controlling cell growth and is involved in early development of the structures in the head and neck. Mutations in the IRF6 gene are thought to affect the development of these tissues, leading to the features seen in Van der Woude syndrome.

Management/Surveillance
Management of Van der Woude syndrome includes evaluations by a craniofacial team, and surgical interventions for clefts of the lip, palate and some lip pits. Like other individuals with cleft lip and/or palate, individuals with Van der Woude syndrome also require routine hearing and dental evaluations.

Mode of inheritance
Van der Woude syndrome is inherited in an autosomal dominant pattern. This means inheriting one IRF6 mutation is enough for an individual to be affected with Van der Woude syndrome. The mutation can be inherited from an affected parent or it can occur brand new (de novo) in an affected child.

Risk to family members
The risk to family members depends on whether or not the individual with Van der Woude syndrome has a parent affected with the condition. If a parent also has Van der Woude syndrome, the risk of having a child with Van der Woude syndrome is 50% with each pregnancy. If a parent does not have Van der Woude syndrome, the risk of other siblings being affected is very low.

Special considerations
In addition to Van der Woude syndrome, mutations in the IRF6 gene can cause a different condition known as
popliteal pterygium syndrome. This condition has the same features of lip pits and cleft lip and/or palate, but also involves webbing of the skin around the knees, eyelids, lips, fingers, and external genitalia. These two conditions occur independently, and a family with Van der Woude syndrome is not at risk to have a child with popliteal pterygium syndrome (and vice versa), but the two conditions are similar in nature, and as such babies should be carefully examined at diagnosis to ensure the correct diagnosis in the family.

Resources
Cleft Palate Foundation
http://www.cleftline.org/
Children’s Craniofacial Association
http://www.ccakids.com/
Genetics Home Reference: Van der Woude syndrome

References

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