Published on *My46* (https://www.my46.org) 01/20/2022
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My46 Trait Profile

22q11.2 deletion syndrome

Other Names: Velocardiofacial syndrome, DiGeorge syndrome, Shprintzen syndrome

22q11.2 deletion syndrome results from a deletion of genetic material on one of the long arms of chromosome 22. This deletion results in the loss of approximately 40 genes.

Characteristics of 22q11.2 deletion syndrome

Individuals with 22q11.2 deletion syndrome often have characteristic facial features, multiple congenital anomalies (i.e., birth defects), as well as developmental, speech, and psychiatric problems.

The distinct, yet subtle facial characteristics of this condition include facial asymmetry when smiling or crying, a small mouth with a thin downturned upper lip, small nostrils and full nasal tip, narrow opening of the eye-lids, and distinctive ear positioning or shape (e.g., low-set, small, cupped, peaked). Individuals with 22q11.2 deletion syndrome are often slightly shorter as compared to other children of the same age.

Common birth defects seen in this syndrome include heart defects, kidney defects, and problems with the palate (roof of the mouth) and trachea (windpipe). Individuals with 22q11.2 deletion syndrome often have a history of feeding and swallowing difficulties in infancy, mild hearing loss, and velopharyngeal insufficiency (i.e., an inability to close the connection between the throat and the nose), leading to the escape of liquids, food, and air through the nose, as well as nasal-sounding speech. Immune system weakness leading to recurrent infections (T-cell deficiency), low calcium levels in the blood due to an underactive or weak parathyroid gland, thyroid problems, and blot clotting problems are frequently seen in this condition.

Speech and communication delay as well as difficulty with articulation (making speech sounds) and expressive language (putting thoughts and ideas into complex sentences) are seen in the majority of individuals with 22q11.2 deletion syndrome. Learning disability or mild intellectual disability is seen in over 90% of individuals with the condition. Additionally, individuals with 22q11.2 deletion syndrome can have delayed social maturity and awareness, as well as anxiety disorders. Individuals with this condition have an increased chance (approximately 25-30%) of developing a psychiatric illness such as depression, bipolar disorder, schizophrenia in adulthood.

Diagnosis/Testing

This syndrome is caused by a deletion (missing piece) of genetic material on one of the two copies of chromosome 22 in each cell. A microarray (also known as an oligoarray, SNP array or arrayCGH) is a blood test that can simultaneously evaluate the cells for small pieces of genetic material that may be missing or extra on each chromosome (the packages of genetic material). A blood test known as FISH (fluorescence in situ hybridization) involves attaching fluorescent probes to the specific area of interest and is frequently used for testing family members of affected individuals. Several genes within this region have been linked to some of the key features observed for individuals with 22q11.2 deletion syndrome. The TBX1 gene is though to be responsible for many of the associated features seen in this condition, including the heart defects.

Management/Surveillance

Management of 22q11.2 deletion syndrome often involves a team of healthcare providers. Management often

involves medical follow-up or surgical correction of the birth defects. It is important to be aware of the surgical risks associated with this condition, which include airway compromise, abnormally-positioned carotid arteries, increased risk for post-surgical hypocalcemia, and immune deficiency. Surveillance also includes lifelong yearly monitoring of thyroid, parathyroid and platelet function, avoiding live viral vaccines (until cleared by a specialist), lifelong adherence to a diet rich in vitamin D and calcium, and prescription medication to treat hypocalcemia, if necessary. Regular dental appointments with preventative treatment from a young age, and regular hearing and vision assessments are also recommended. Educational services, physical and occupational therapies may be helpful for the feeding and motor issues seen in this condition. Periodic assessment of mental health status and advanced planning for transition into the adult health care system is essential.

Mode of inheritance

22q11.2 deletion syndrome is inherited in an autosomal dominant pattern. This means inheriting one 22q11.2 deletion is enough for an individual to be affected and show signs of 22q11.2 deletion syndrome. The deletion 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 22q11.2 deletion syndrome has a parent with the deletion. If a parent also has the deletion, the risk for that parent to have another child with the deletion is 50% with each pregnancy. If a parent does not have the deletion, the risk of other siblings being affected is very low.

Special considerations

None

Resources

Children's Hospital of Philadelphia: 22q and You Center
http://www.chop.edu/service/22q-and-you-center/
VCFS Foundation of NSW: Parent's Guide to Deletion 22q11
http://www.vcfsfa.org.au/media/documents/vcfs%20parents%20guide.pdf
SickKids: 22q11 Fact Sheets
http://www.sickkids.ca/CGenetics/What-we-do/22q-deletion-syndrome-clinic/Meet-the-22q-team/index.html
Genetics Home Reference: 22q11.2 deletion syndrome
http://ghr.nlm.nih.gov/condition/22q112-deletion-syndrome
Unique: Understanding chromosome disorders
http://www.rarechromo.org/information/Chromosome%2022/22q11.2%20deletions%20syndrome%20(Velo-Cardio-Facial%20Syndrome)%20FTNW.pdf

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Created: 06/2013 **Created by:** Andrea Shugar, MS, CGC
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