



What is 22q11.2 Deletion Syndrome?

22q11.2 deletion syndrome is a genetic condition that affects learning, health, and physical traits. It occurs in males and females of all racial and ethnic backgrounds. 22q11.2 deletion syndrome occurs in about 1 in 4,000 to 6,000 people. It is sometimes known as Velocardiofacial or DiGeorge syndrome.

Michigan Resources & Support

Children's Special Health Care Services

Family Phone Line
Toll-free: 1-800-359-3722
E-mail: pcsp@michigan.gov
www.michigan.gov/cshcs

Early On[®] Michigan

Toll-free: 1-800-EARLY ON
www.1800earlyon.org

Michigan Birth Defects Program

Nurse Follow-up Coordinator
Toll-free: 1-866-852-1247
E-mail: BDRfollowup@michigan.gov

Michigan Genetics Connection

www.migeneticsconnection.org

National Resources & Support

Chromosome 22 Central

www.c22c.org

International 22q11.2 Deletion Syndrome Foundation

Toll-free: 1-877-739-1849
www.22q.org/

Velo-Cardio-Facial Syndrome Educational Foundation

Toll-free: 1-866-VCFSEF5
www.vcfsef.org

Family Village

www.familyvillage.wisc.edu

GeneReviews

www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=gr_22q11deletion

Genetic and Rare Diseases Information Center

Toll-free: 1-888-205-2311
E-mail: GARDinfo@nih.gov

Genetics Home Reference

www.ghr.nlm.nih.gov/condition=22q112deletionsyndrome

How may the syndrome affect my child?

Learning: There is a range of abilities. Some children with 22q11.2 deletion may have normal learning, while others have a developmental delay, learning disability or severe lifelong learning problems.

Behavior: Mental (psychiatric) illnesses including depression, bipolar disorder and schizophrenia have been reported in some people with 22q11.2 deletion syndrome.

Physical: Children with 22q11.2 deletion syndrome may have certain facial features, such as a prominent bridge of the nose and narrow eyes,

often noticed only by a medical professional. Short stature is also a common trait.

Medical: Heart defects are very common. They can be mild or severe enough to need surgery. Many children have palate abnormalities. These include an opening in the roof of the mouth (cleft palate) and a change in the way the throat closes (velopharyngeal incompetence). Other medical problems can include kidney abnormalities, problems with the immune system and low calcium levels.

How does the syndrome occur?

22q11.2 deletion syndrome is caused by a change in the #22 chromosome. A very small piece of the chromosome is missing (deleted), including some of the genes within it. The child is often the first and only family member affected, but sometimes the deleted chromosome is passed down from a parent. A person with the 22q11.2 deletion has a 1 in 2 (50%) chance of passing it on to each of his or her children. Genetic counseling is recommended for parents to learn about the genetic cause of the syndrome in their family, and possible health risks for other children.

How is the syndrome treated?

22q11.2 deletion syndrome cannot be cured, but many symptoms can be treated. Surgery may be needed to repair a heart defect or cleft palate. The immune system should be tested, and the kidneys should be checked by ultrasound in infancy. Calcium supplements may be needed. Infants and toddlers (birth to 3 years) should be connected with *Early On*[®] Michigan as soon as possible. If there are concerns about learning, speech, or behavior in a child over 3 years of age, a referral for special education services should be made. Other therapies or treatments may be available for problems as they arise. Children with 22q11.2 deletion syndrome and their families benefit from having a primary care physician who helps to coordinate their care with medical specialists and other community-based services.

For more information, call Michigan's Genetics & Birth Defects Program toll-free at 1-866-852-1247 or e-mail Genetics@michigan.gov

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