Fact Sheet

Division of Child and Adolescent Health
Pediatric Screening and Genetic Services
Virginia Genetics Program
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www.vahealth.org/genetics

VDH VIRGINIA DEPARTMENT OF HEALTH
Protecting You and Your Environment
www.vdh.virginia.gov

22q11.2 Deletion Syndrome

What is 22q11.2 Deletion Syndrome?
22q11.2 Deletion Syndrome means that a child is missing a small part of chromosome 22. It is sometimes called DiGeorge syndrome or Velocardiofacial (VCF) syndrome.

Found inside the cells of the body, chromosomes are tiny thread-like structures that house our genetic traits. Most of our body cells have 46 chromosomes. They come in pairs, for a total of 23 pairs. One member of each pair comes from our mother; the other comes from our father. The 23 pairs of chromosomes are numbered from largest to smallest, 1 to 22. The 23rd pair of chromosomes is the sex chromosomes, which determine whether we are a boy or a girl. Chromosomes are divided into two parts called arms. The top half of the chromosome is the short or "p" arm and the bottom half is called the long or "q" arm.

22q11.2 Deletion Syndrome occurs when a small section of one copy of chromosome 22 is missing (deleted). The “q11.2” tells doctors that the missing piece is in a very specific spot on the "q" arm. When part of a chromosome is missing, the child only has one copy of each of the genes within the section instead of the typical two copies.

What types of problems occur in 22q11.2 Deletion Syndrome?
Children with 22q11.2 Deletion Syndrome can have a wide range of physical and developmental problems. These problems can include heart defects, cleft palate, feeding problems, lower resistance to infection, growth problems, learning problems, and psychiatric illness. Not every person with 22q11.2 Deletion Syndrome will have the same problems.

How common is 22q11.2 Deletion Syndrome?
22q11.2 Deletion Syndrome occurs in about 1 of every 4,000 births in the United States. In Virginia, about 10 children are reported each year with 22q11.2 Deletion Syndrome.

What causes 22q11.2 Deletion Syndrome?
22q11.2 Deletion Syndrome is due to a missing section of chromosome 22. Most of the time, the deletion just happens by chance. About 7 out of 100 times, the child has inherited the deletion from one of his or her parents. The parents might not know if they have the deletion because they might not have any noticeable problems. Parents of a child with 22q11.2 Deletion Syndrome may have a higher chance of having another child with 22q11.2 Deletion Syndrome. If one of the parents has the deletion also, there would be a 50:50 chance of the next child having 22q11.2 Deletion Syndrome. A genetic counselor or geneticist can help you to determine the risks for your family and situation.

What can be done to help my child with 22q11.2 Deletion Syndrome?
Children with a heart defect or a cleft palate may require surgery to treat these issues. Children with 22q11.2 Deletion Syndrome can benefit from early intervention programs. These programs normally have staff to ad-

dress many of the babies' specific needs including learning specialists who will help the family understand how best to teach the child.

Where can I go for more information about 22q11.2 Deletion Syndrome?

Chromosome 22 Central
www.c22c.org
(919) 567-8167

22q and You Center
http://genetics.chop.edu/22q
(215) 590-1000

Chromosome Deletion Outreach, Inc.
www.chromodisorder.org
(561) 395-4252

March of Dimes Birth Defects Foundation
www.modimes.org
1-888-MODIMES (1-888-663-4637)

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