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Why is identifying **22q11.2 deletion syndrome** (DiGeorge syndrome/VCFS/22q) important?

22q11.2 deletion syndrome is more common than cystic fibrosis, occurring in approximately 1 in 2,000 live births[1] (recent studies have shown incidence rates as high as 1/992[2]), and can cause a wide variety of congenital anomalies as well as mild-to-moderate intellectual disabilities.

Early medical and developmental intervention can be critical for improving the quality of life of the child.

1. **Hypocalcemia**. Low calcium levels, or hypocalcemia, is common with the 22q11.2 deletion, especially in newborns. Low calcium levels may lead to seizures. Often the cause of hypocalcemic seizures is overlooked and may go untreated, impacting the intellectual development of the child. ***Newborns with the 22q11.2 deletion should be monitored for hypocalcemia and, if identified, should be treated promptly.*** It is not uncommon for hypocalcemia to recur in affected individuals during growth spurts, puberty, illness or surgery.[3]

2. **Immune deficiency.** Almost 75% of individuals with the 22q11.2 deletion have immune problems. ***Because of the risk for immune deficiency, affected individuals should be evaluated prior to receiving live virus vaccines.***[4]

3. **Palatal abnormalities.** Almost 75% of individuals with the 22q11.2 deletion will have a palatal problem, either structural, functional, or a combination.[5] These problems can result in feeding and/or speech issues. If not corrected early, speech development may be impacted. ***Most of the time, these are treatable conditions.***

4. **Feeding difficulties.** Newborns with the 22q11.2 deletion often have difficulty feeding unrelated to palatal or cardiac abnormalities.[6] The underlying feeding problem may be caused by pharyngoesophageal dysmotility and commonly results in reflux and constipation. Rarely, intestinal malrotation and Hirschsprung disease have been reported.[7] ***Most of these problems are treatable.***

5. **Congenital cardiac defects.** About 75% of individuals with the 22q11.2 deletion have congenital heart defects; this finding is often the reason for diagnosis.[8] ***If an individual is diagnosed with the 22q11.2 deletion, they should be referred to a cardiologist.***

6. **There are other medical problems related to the 22q11.2 deletion** including renal anomalies, hearing loss, ENT complications, autoimmune diseases, growth delay and skeletal anomalies.[9,10,11]

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Treatment of the various conditions listed on the other side, and, in some cases, the addition of physical therapy, occupational therapy, and special education can help improve the outcomes for individuals with 22q11.2 deletion syndrome.

For more information on 22q11.2 deletion syndrome, please visit www.22q.org.

What is 22q11.2 deletion syndrome?

22q11.2 deletion syndrome, also called DiGeorge syndrome, Velo-Cardio-Facial syndrome (VCFS), or 22q, is caused by a missing piece of chromosome number 22. The resulting condition affects many parts of the body. The majority of children with this disorder have heart defects, immune system problems, and characteristic, though often subtle, facial features. Nearly all have mild-to-moderate intellectual disability and delayed speech and language. Some children will have low calcium levels, kidney problems, feeding problems, seizures, or other health problems. About one in five children with 22q11.2 deletion syndrome has autism spectrum disorder, and one in four young adults has a psychiatric illness, such as schizophrenia.

There is an increased risk of infant death in babies with severe heart or immune system problems. Individuals with the 22q11.2 deletion syndrome who survive childhood may have a shorter lifespan and may be at higher risk of sudden death.

What is the cause of 22q11.2 deletion syndrome?

The majority of individuals with the 22q11.2 deletion syndrome have a 3 MB deletion (encompassing, on average, 40 genes) on one of the copies of chromosome 22. The 22q11.2 deletion happens by chance and, in most cases, is not inherited. However, about 7% of children with the 22q11.2 deletion syndrome did inherit it from a parent who also has the condition. Testing of both parents can help determine the chance of this condition happening again in another pregnancy.

How should I address a high risk Panorama™ test result for 22q11.2 deletion syndrome?

The Panorama test is a screen, not a diagnostic test. Any high risk result should be confirmed with an amniocentesis or chorionic villus sampling (CVS) that includes a chromosome microarray (CMA) which specifically looks for the missing piece of chromosome 22 that causes the 22q11.2 deletion syndrome. If your patient elects not to have an amniocentesis or CVS, chromosome testing with a CMA can be done on the baby after birth.

Where can I send my patients for additional information?

You can send your patients to:
www.panoramatest.com/resources.
Several 22q11.2 deletion syndrome support groups' websites have links on this page. You can also contact Natera directly to ask our staff of genetic counselors any questions you or your patients may have. The phone number is 650-249-9090.

References available at www.panoramatest.com/references-07

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