



22 Questions about 22q11.2 Deletion Syndrome

1. What is 22q11.2 Deletion Syndrome?

22q11.2 deletion syndrome is a chromosomal abnormality that can cause a wide range of health and developmental issues, including heart defects, breathing issues, gastrointestinal tract problems, immune and endocrine systems issues, differences in the palate, slow growth, autism/developmental delays or learning disabilities in some individuals. Children with this condition may have many or only a few of the symptoms, with varying severity.

2. How common is 22q11.2 Deletion Syndrome?

The deletion occurs in an estimated 1 of 2,000 live births, although this is possibly an underestimate of the prevalence of this deletion, making it almost as common as Down Syndrome. In addition, it is the most frequent cause of syndromic palatal defects and it is found in 1 of 68 children born with a heart defect. Despite this prevalence, many physicians are still not familiar with the diagnosis or its extreme variability. Because of this, some patients are in their late teens or even adulthood before the diagnosis is made.

3. What tests are used to identify the deletion?

Today there are many tests that can identify the deletion and they are most readily performed as blood tests. They include: FISH (fluorescence in situ hybridization), comparative genomic hybridization (CGH), whole genome or SNP array, and multiplex ligation-probe amplification (MLPA). In general, a regular chromosome study will only identify this very small deletion about 25% of the time so one of the above more specialized tests would need to be requested in order to find the deletion.

4. Is 22q11.2 Deletion Syndrome hereditary?

Most times the deletion is not hereditary or "running in the family" but once it is present the person with the deletion has a 50% chance of passing it on to his or her child. It is important to know that the eggs and sperm have no memory so every pregnancy has the same 50% chance of having a child with the deletion even if there is an affected child already in the family. In addition, as the findings in people with the 22q11.2 deletion are variable it is impossible to predict how mildly or significantly the child will be affected.

5. What are the symptoms of 22q Deletion Syndrome?

Most affected areas:

Cardio-vascular, Cleft Palate, Feeding difficulties, Immunization Problems, Growth hormone deficiencies, Delayed neurological and psychological developments, Speech problems, Renal abnormalities

6. How many symptoms are related to 22q?

The count seems to change all the time, but at present there are approximately 189.

7. Will my child have all 189 symptoms?

No, it is varied in each person. Some will have more, some will have less and none will be affected in the same way. This is one of the reasons why it is so hard for physicians to see that what they are observing all goes together, and is in fact a syndrome.

8. What are the medical needs if diagnosed with 22q Deletion Syndrome?

Medical needs are varied depending in the affected body systems. The mortality rate in children with the 22q11.2 deletion is very low (~ 4%) and those children who do succumb to problems associated with the deletion often pass away at a very young age (~ 4 months). However, many children and adults have numerous medical problems across their lifetime that requires specialized care.

9. What treatments are available for 22q?

Treatment for 22q11.2 deletion syndrome varies depending on your child's symptoms. Because your child's genetic makeup can't be changed, doctors can't "cure" the chromosome 22q11.2 deletion. Once your child is diagnosed, doctors will usually recommend that your child undergo evaluation in the areas we know this chromosomal difference affects. Once your child's medical and developmental problems are pinpointed, doctors can help develop plans that will manage them and help your child live as healthy a life as possible.

10. How do families cope?

Most children and adults with the 22q11.2 deletion do quite well both medically and as members of their families and communities at large. As with anything that is unexpected, coming to terms with the diagnosis is often difficult at first but becomes easier as more information becomes available and as families have an opportunity to meet other children and adults with the 22q11.2 deletion and/ or to converse with them through diagnosis specific internet sites. In addition, attendance at family meetings/picnics; contacting support networks; and sending children to camps specifically designed for those individuals with a 22q11.2 deletion such as Dragonfly Forest (www.dragonflyforest.org) is often helpful as families realize they are not alone.

11. Can a child with 22q attend regular education classes?

As with any child, educational decisions need to be decided by the individual family. Many children with 22q are in regular education classes, however some do need extra supports and accommodations. Many individuals with 22q have graduated from college or advanced technical programs.

12. What do teachers need to know about 22q?

Most children (90%) with 22q deletion experience some degree of developmental disability with delayed speech and language development. In standardized testing, most school aged children have an IQ of borderline intellectual disability.

13. What other names have been used for 22q?

DiGeorge syndrome
Velocardiofacial syndrome (VCFS)
Shprintzen syndrome
Conotruncal anomaly face syndrome (CTAF)
Sedlackova syndrome

14. Can someone with 22q drive?

This depends on your child's abilities at the time. Many 22q young adults do drive but many do not have the ability to do so. This can only be determined as the child gets closer to the age of learning to drive. You will know by their level of common sense, motor skills, and sense of direction.

15. Can someone with 22q live independently?

This is another area where you will need to know your child's abilities. These abilities are things you'll need to determine as they get into puberty and close to adulthood. If not, then these are areas where you need to try and teach them to prepare for the best outcome!

16. Can someone with 22q marry and have children?

Like anything else with this syndrome, the answer can be YES and it can be NO. Only time and the child's individual abilities will determine this outcome.

17. How can I help promote 22q awareness?

Our goals are to have 22q become a household name so that no one says, "What is that, I've never heard of it!" AND to have OB/GYNs request that newborns be screened for it at birth. To accomplish these goals, we each need to carry literature that explains the subject. We also need to be prepared to talk about it when we have the chance. The key is, networking, talking, and sharing. Our efforts will result in recognition and appropriate treatment for this disorder.

18. What does 22q11.2 Deletion Syndrome mean?

22q11.2DS is pronounced "twenty-two Q, one, one, point two deletion syndrome. Just like how we describe the location of a house by using its street name and number, 22q11.2 indicates a location within our DNA: chromosome 22, section 11.2. People with 22q11.2DS are missing a small section of DNA from the 22nd chromosome. This small section is "deleted".

19. Who is more likely to have 22q11.2DS?

Both boys and girls have the same chance of having this deletion. 22q11.2DS is found world-wide. It is found in every culture and in every ethnic group. No one ethnicity has more people with 22q11.2DS than another. When a child is born with 22q11.2DS, it is just a random genetic occurrence.

20. What does it mean to be "missing a small piece of DNA"?

Every cell in the human body is missing a small piece of genetic material, As a result, various health problems may occur. Every person is unique from one another. Some people with 22q11.2DS may have few features while others may have many. This variation in 22q11.2DS features can even occur when a parent passes the deletion to his/her child. In other words, the features of a parent with 22q11.2DS may be very different from those of his/her affected child.

21. Can doctors repair my DNA so I don't have the deletion anymore?

No, but there are many ways to manage the needs of someone with 22q through genetic and medical testing.

22. What is life like for people with 22q?

People with 22q can have loving relationships with friends and family, go to school with their peers, are interested in learning about a variety of topics. As adults, many individuals work in jobs they find meaningful and are often able to live independently or semi-independently. Many people with 22q will marry and have children of their own.