



DiGeorge syndrome, also called 22q11.2 deletion syndrome, is associated with several health and developmental concerns. It is important to remember that each individual with DiGeorge syndrome is unique, and the symptoms can vary tremendously from person to person. Some people with DiGeorge syndrome do not have any major noticeable health effects, and in fact may never know they have it, while others may have a number of significant health and developmental concerns which in some cases can be quite serious.

DiGeorge syndrome also goes by a couple other names, including velocardiofacial syndrome, Shprintzen syndrome, and conotruncal anomaly face syndrome. There are also a couple other health conditions, called autosomal dominant Opitz G/BBB syndrome and Cayler cardiofacial syndrome, that are more common in people with DiGeorge syndrome.

What is life like for people with DiGeorge syndrome?

People with DiGeorge syndrome can have loving relationships with friends and family, go to school with their peers, and 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 DiGeorge syndrome will have children of their own. You can find stories about children and adults with DiGeorge syndrome at 22q.org.

What Causes DiGeorge syndrome?

DiGeorge syndrome is caused by a deletion on the long arm ("q") of the 22nd chromosome, which is also why it is called 22q11.2 deletion syndrome (11.2 is specific location where the deletion is). This deletion results in the loss of several genes. Most of the time this deletion is said to be 'de novo', which means that it was not passed down from a parent; it is brand new in the individual who is diagnosed with the condition. In these cases, the deletion occurs in the development of an egg or a sperm and is then copied over into every cell in the developing baby's body. About 10% of people who are diagnosed with DiGeorge syndrome have inherited the deletion from a parent who also has it. In many of these cases, the parent did not know they had DiGeorge until their child was diagnosed.

What are the health and developmental concerns associated with DiGeorge syndrome?

The health and developmental concerns associated with individuals with DiGeorge syndrome varies significantly from person to person, even within the same family. Below is a list of some of the more common concerns associated with DiGeorge syndrome. It is important to know that typically people with 22q11.2 deletion syndrome do not necessarily



experience all of these health concerns.

- Congenital (present from birth) heart defects
- Abnormalities of the palate (roof of the mouth), such as cleft palate
- Developmental delays, such as walking or talking later than normal
- Learning difficulties
- Mild intellectual disability
- Immune deficiency/autoimmune disorders
- Hypocalcemia (not enough calcium in the body)
- Difficulties with feeding and swallowing
- Gastrointestinal anomalies
- Hearing loss
- Growth hormone deficiency
- Seizures
- Ophthalmologic (eye) abnormalities
- Autism or autistic spectrum disorder
- Psychiatric illness in adults

If I have a baby with DiGeorge syndrome, what is the chance I will have another baby with this condition?

The chance to have another baby with DiGeorge syndrome depends on whether or not one of the parents carries the deletion. If one of the parents of an individual with DiGeorge syndrome is found to carry the deletion, the chance for each pregnancy to be affected is 50%. If both parents undergo genetic testing for the deletion and neither are found to carry it, then the risk to for future pregnancies is low in most cases.

How do parents of children with DiGeorge syndrome feel about raising a child with it?

A study that surveyed the parents of 76 school-aged children with DiGeorge syndrome reported that the majority of parents felt that their child had brought joy and happiness to their lives. They said they enjoyed their child's sense of humor, and felt that having their child increased their patience and tolerance. Many parents indicated challenges regarding parenting a child with DiGeorge syndrome related to their learning, communication, and medical difficulties. Many parents expressed some concern about the future for their child related to their child's ability to live independently, find meaningful employment, and concerns related to future mental and physical health.



What is the treatment for for DiGeorge syndrome?

Typically, the chromosome deletion that causes DiGeorge is copied over into every cell in the person's body, and the missing genes and proteins cannot be replaced. There is no "cure" for DiGeorge syndrome. Screening for and treatment of various health concerns can be important to reduce complications associated with this condition. Individuals with DiGeorge syndrome may require regular follow-up and treatment from various health specialties depending on their specific needs. In many cases, early intervention programs for individuals with DiGeorge syndrome can help them reach their full potential. Intervention programs may include speech, physical, occupational, and/or educational therapy.

What are the long term outcomes for individuals with DiGeorge syndrome?

The health and developmental effects of DiGeorge syndrome vary tremendously, so the long term prognosis is very individual. Some people with DiGeorge syndrome have no intellectual delays at all, while others may have more significant developmental and intellectual delays and may need more help. Some individuals with DiGeorge syndrome will develop mental health concerns, others will not. Physical health issues related to the heart, immune system, and endocrine system be ongoing issues throughout life, or they could not be diagnosed until adulthood. The information available on life expectancy is limited, however some studies indicate that individuals with DiGeorge syndrome may often have a shorter than average expectancy.

How common is DiGeorge syndrome?

Most studies indicate that the DiGeorge syndrome occurs in about 1 in 4,000 births. However, many think it may be more common given that a high percentage of people with DiGeorge syndrome may never be diagnosed.

Click [here](#) to learn more about scheduling a genetic counseling appointment for pregnancy-related questions.

Click [here](#) to learn more about scheduling a genetic counseling appointment for infertility or preconception questions.

Click [here](#) to learn more about scheduling a genetic counseling appointment for questions about pediatric or adult genetic conditions.



Other Resources

[The International 22q11.2 Foundation, Inc.](#)

[The 22q Family Foundation References](#)