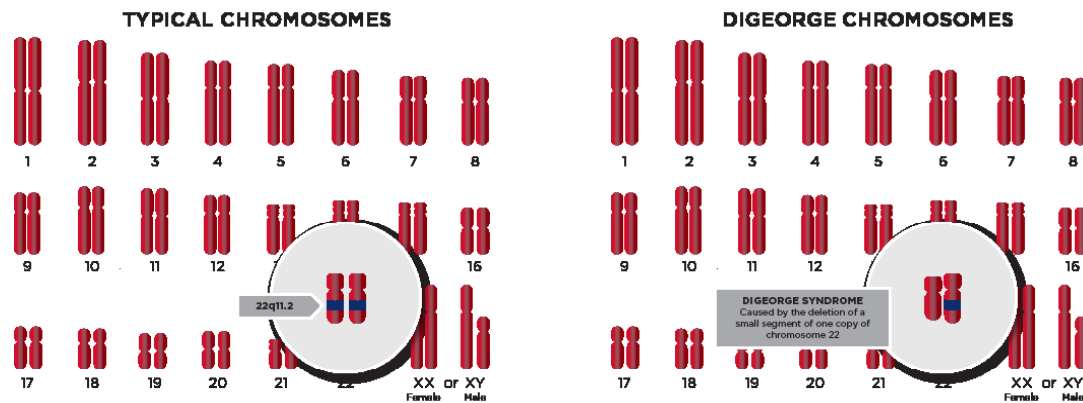


22q11.2 Deletion Syndrome

What is 22q11.2 Deletion Syndrome?

At least 1 in 2,000 to 1 in 4,000 people have 22q11.2 deletion syndrome (22q11.2 DS), which is also known as DiGeorge syndrome or velocardiofacial (VCF) syndrome. 22q11.2 DS is a genetic condition in which a small portion of chromosome 22 is deleted or missing.



In a small percentage of cases, the condition is inherited, but most often, it appears as a random occurrence in a family. Some affected individuals have signs and symptoms identified before birth, some have signs apparent shortly after birth, and still others may go undetected for years or decades.

22q11.2 DS has a wide array of symptoms that can affect different organ systems. If your child is diagnosed with this condition, he or she will need treatment from specialists in a variety of fields.

Medical challenges commonly associated with 22q11.2 DS include:

- » Congenital heart defects
- » Frequent infections
- » Developmental delays or learning difficulties
- » Underdeveloped or absent thymus
- » Immune deficiencies
- » Gastrointestinal disorders
- » Cleft palate and other facial abnormalities
- » Low levels of calcium in the blood which can cause seizures if untreated
- » Increased risk of mental health conditions, such as schizophrenia, ADHD, and autism spectrum disorders

A Cardinal Glennon St. Louis Fetal Care Institute nurse is available 24 hours a day, seven days a week to discuss referrals with physicians and potential families by calling 314-268-4037, option 2.

Phone 314-268-4037, option 2
Toll-free 1-877-SSM-FETL (776-3385)
Web stlouisfetalcare.com
Email fetalcare@ssmhealth.com
Facebook facebook.com/fetaldocs

22q11.2 Deletion Syndrome

What can I expect if I have been told my unborn baby may have 22q11.2 DS?

When you visit the Cardinal Glennon St. Louis Fetal Care Institute with a presumed or confirmed diagnosis of 22q11.2 DS in your baby, you have the opportunity to meet with a team of experts who can provide information about your baby's diagnosis. This team will involve:

- » Maternal-fetal medicine specialists
- » Genetic counselors
- » Clinical geneticists
- » Social workers
- » Footprints/palliative care nurses
- » Neonatologists

Additionally, based on your unique pregnancy, you may have follow-up visits with other pediatric specialists.

How is 22q11.2 DS diagnosed?

A probable 22q11.2 DS case can be discovered prenatally through:

- » Amniocentesis
- » Genetic screening
- » Prenatal ultrasound

An amniocentesis can confirm a diagnosis before birth. Diagnosis can also be made after birth, typically through blood testing. Genetic screening such as noninvasive prenatal testing (NIPT) identifies pregnancies at increased probability of having the chromosome 22q11.2 deletion, but does not confirm the diagnosis. Signs of a congenital heart defect, cleft palate, or other defect on a prenatal ultrasound also increase the possibility that a baby has this condition. All of these tests are available through the Cardinal Glennon St. Louis Fetal Care Institute.

How will 22q11.2 DS affect my baby after delivery?

At the Cardinal Glennon St. Louis Fetal Care Institute your little one will receive specialized care in a state-of-the-art hospital that has the equipment, team, and experience to meet his or her needs. Because of the complex nature of 22q11.2 DS, babies may face medical challenges during delivery and in the newborn period, which is why it is recommended that they be delivered at a hospital with the resources to handle their unique needs.

Every baby's situation is different, and the Cardinal Glennon St. Louis Fetal Care Institute works with each family to prepare for their baby's delivery and to create a care plan to follow after birth.

How will 22q11.2 DS affect my baby into childhood?

There is no cure for 22q11.2 DS. The prognosis for newborns with the condition varies and depends on the severity of their symptoms and the organs affected. Most people with 22q11.2 DS who receive ongoing healthcare and treatment from specialists have a normal life expectancy and achieve some degree of independence. A smaller percentage of affected babies have significant complications related to their hearts or immune systems

that may be life-threatening.

What are the treatment options for babies with 22q11.2 DS?

Babies with 22q11.2 DS may struggle with feeding and growth problems, hearing or vision loss, and other medical conditions. Fortunately, surgery to correct medical problems associated with 22q11.2 DS, such as heart defects and cleft palate, often provide effective treatment for many of these babies.

Children with the condition will benefit from the care of a multidisciplinary medical team, which may include:

- » Pediatricians
- » Medical geneticists
- » Surgeons
- » Cardiologists
- » Endocrinologists
- » Speech pathologists
- » Otolaryngologists
- » Ophthalmologists
- » Psychologists
- » Occupational therapists
- » Other healthcare professionals.

For children under the care of five or more specialists, the Complex Medical Care program at SSM Health Cardinal Glennon Children's Hospital can help by bringing together all of your child's care providers to help ensure continuity of care. This team provides:

- » Individualized care plans that we make accessible in different health care environments
- » Coordination to ease the child's transition between hospital, home, and community outpatient facilities
- » Safe transportation between facilities
- » Education to help optimally deliver a child's care once he or she gets home
- » Ongoing communication with family, primary care providers, and specialists

Preparing for the birth of a baby with 22q11.2 DS can be overwhelming and scary. That's why we're available to help 24 hours a day, 7 days a week. For more information or to schedule an appointment, call us at 314-268-4037, opt. 2 or toll free at 877-SSM-FETL (877-776-3385).
