

# Trisomy 21 Down Syndrome

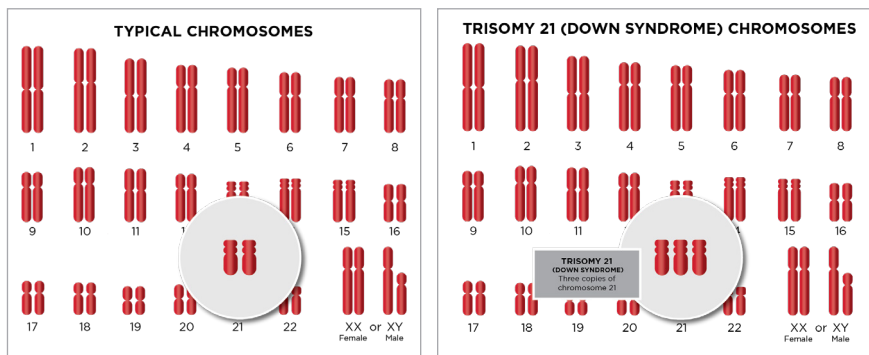
## What is Trisomy 21?

Trisomy 21, more commonly known as Down syndrome, is a genetic disorder causing developmental and intellectual delays. This disorder affects approximately 1 in every 700 babies.

Normally, a person has 23 pairs of chromosomes. Chromosomes are the packages of genetic information, made of DNA, that contain the instructions the body uses to build a person. Chromosomes come in 23 pairs, with most people having 46 total chromosomes. Trisomy 21 is caused when a person has three copies of chromosome #21 instead of the usual two, for a total of 47 chromosomes. This extra chromosome affects a baby's development, resulting in a number of medical issues which may include: a distinct facial appearance, intellectual disability, developmental delays and thyroid or heart disease.

## How is Trisomy 21 Diagnosed?

Trisomy 21 can be identified prenatally through screening tests such as non-invasive prenatal testing (NIPT) and ultrasound examinations. The diagnosis can be confirmed prenatally with better than 99% accuracy through chorionic villus sampling (CVS) or amniocentesis.



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](http://stlouisfetalcare.com)  
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Facebook [facebook.com/fetaldocs](https://facebook.com/fetaldocs)

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Both of these advanced diagnostics tests are available at the SSM Health Cardinal Glennon St. Louis Fetal Care Institute. The diagnosis can also be confirmed shortly after birth through blood testing.

Mothers over the age of 35 or with a family history of Down syndrome have a greater risk of having a baby with Trisomy 21 and may want to talk with a doctor about genetic counseling and additional screenings.

### **If I have been told my unborn baby may have Trisomy 21, what can I expect during my visits at the Cardinal Glennon St. Louis Fetal Care Institute?**

At the Cardinal Glennon St. Louis Fetal Care Institute, our team has the experience and technology to prenatally diagnosis Trisomy 21, as well as assist families in preparing for delivery and beyond.

Using a multidisciplinary approach, our team will evaluate your baby to determine its specific needs. Ultrasounds will be used to monitor your baby's growth and development, and a fetal echocardiogram may be used to check for a heart defect, which is more common in babies with Trisomy 21. You will also work with our social worker to learn about the resources available for families of individuals with Down syndrome.

The risk of miscarriage and stillbirth increases in babies with Trisomy 21, but those who are carried to full term can usually be delivered vaginally without incident unless there are other reasons to consider an alternate mode of delivery.

### **How is Trisomy 21 treated?**

Down syndrome cannot be cured, but early intervention and treatment tailored to each child's specific situation are helpful in managing this condition. Although babies born with Trisomy 21 will require some amount of care throughout their lifetimes, they can grow up to lead healthy, happy, productive lives.

### **What can I expect after my baby is born with Trisomy 21?**

Typically, patients at the Cardinal Glennon St. Louis Fetal Care Institute will schedule a visit with a pediatric geneticist at SSM Health Cardinal Glennon Children's Hospital within the first few weeks of life and then follow up at the Down Syndrome Clinic where they will be able to see multiple specialists in one location.

The specialists a particular individual with Down syndrome will see depends on their specific health issues, which can include heart defects, frequent respiratory infections, poor muscle tone, seizures, sleep problems and/or hearing, eye and dental problems. Parents will need to make sure their child has routine medical checkups and be on the lookout for early signs of these issues.

Families are encouraged to work with the Down Syndrome Association of Greater St. Louis or to seek out support from similar organizations where they live. Families who are seen at the Cardinal Glennon St. Louis Fetal Care Institute whose babies are suspected to have Down syndrome will be given resources from appropriate organizations.

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We understand that Trisomy 21 Down Syndrome can be a scary diagnosis. 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 or toll free at 877-SSM-FETL (877-776-3385).

While we can't change the diagnosis, we can provide you with expert care and support, helping your baby get the most out of treatment and life.

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