

Result Navigator

Your screening result is positive for trisomy 21 (Down syndrome).

Expectant parents often have questions about what a positive result means and what to do next. This information answers some common questions and helps guide you on this journey.

What is trisomy 21?

Every year, about 6,000 babies in the United States are born with trisomy 21 (an extra chromosome 21). Children with trisomy 21 have distinct facial features. They require special care due to below average intelligence, difficulty achieving life skills, and a higher risk for certain health problems.

Trisomy 21 happens by chance. There is nothing that a parent can do to cause or prevent trisomy 21, and it doesn't usually run in families.

▶ What does “positive” mean?

A positive screening result does not diagnose your baby with trisomy 21. It means your baby's risk to have trisomy 21 is higher than most other babies.

Information about you and your baby was used to calculate the chance that this result correctly predicts that your baby has trisomy 21. This is called a positive predictive value (PPV) on your report. Other information may change this value. It is important to discuss PPV with your provider.

●●● What other tests can tell me more?

Other Tests	What will it tell me?	When can I do it?	How is it done?	Is there added risk to my baby?	How long do results take?
Prenatal Diagnostic Test	It tells you if your baby definitely has trisomy 21.	During pregnancy (At certain times)	Chorionic villus sample (CVS) or an amniotic fluid sample	Yes. Discuss risks with your provider.	1-2 weeks
Prenatal Ultrasound Screen	It tells you if your baby's risk might be higher or lower.	During pregnancy (At certain times)	Non-invasive imaging	No. There are no added risks.	Immediate
Postnatal Diagnostic Test	It tells you if your baby definitely has trisomy 21.	After birth (At any time)	Blood sample from baby	No. There are no added risks.	About 1 week

What should I do next?

Take time to process this information. Write down additional questions. Then, talk with your provider about ways you can learn more about your pregnancy.

▶ Where do I find more information?

Genetics Home Reference
ghr.nlm.nih.gov

March of Dimes
marchofdimes.org