

Result Navigator

Your screening result is positive for monosomy X (Turner 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 monosomy X?

Every year, about 800 babies in the United States are born with monosomy X (a missing chromosome X). People with monosomy X have some distinct features, such as a short neck, low hairline, and a broad chest. They are at a higher risk for certain health and reproductive problems, but typically have normal intelligence. Miscarriage of a pregnancy with monosomy X is common.

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

▶ What does “positive” mean?

A positive screening result does not diagnose your baby with monosomy X. It means your baby's risk to have monosomy X 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 monosomy X. 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 monosomy X.	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 monosomy X.	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

Genetic Support Foundation
geneticsupportfoundation.org