

# Result Navigator

## Your screening result is positive for 47, XXY (Klinefelter 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 47, XXY?

Every year, up to 4,000 babies in the United States are born with 47, XXY (an extra chromosome X). People with 47, XXY may be taller than average and have delayed or incomplete puberty. They are at a higher risk for certain health and reproductive problems and may have some problems with learning.

**47, XXY happens by chance.** There is nothing that a parent can do to cause or prevent 47, XXY, and it doesn't usually run in families.

### ▶ What does “positive” mean?

A positive screening result does not diagnose your baby with 47, XXY. It means your baby's risk to have 47, XXY 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 47, XXY. 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?
<b>Prenatal Diagnostic Test</b>	It tells you if your baby definitely has 47, XXY.	During pregnancy (At certain times)	Chorionic villus sample (CVS) or an amniotic fluid sample	Yes. Discuss risks with your provider.	1-2 weeks
<b>Prenatal Ultrasound Screen</b>	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
<b>Postnatal Diagnostic Test</b>	It tells you if your baby definitely has 47, XXY.	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](http://ghr.nlm.nih.gov)

**Genetic Support Foundation**  
[geneticsupportfoundation.org](http://geneticsupportfoundation.org)