Natera**Academy**

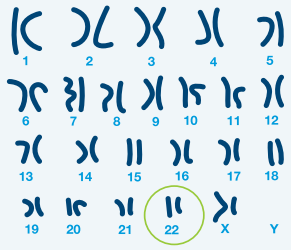
22q11.2 deletion syndrome

What is 22q11.2 deletion syndrome?

22q11.2 deletion syndrome is a **common genetic condition** that can cause a variety of **serious health problems** and **learning difficulties.**

The condition can occur in any pregnancy and is not always detected by ultrasound.

What are the causes?



A person typically has **46 chromosomes** grouped into 23 pairs. 22q11.2 deletion syndrome is caused by a **missing piece of chromosome 22, called a deletion.**

This deletion usually **happens by chance** and cannot be predicted. A small percentage of people inherit the deletion from a parent.

Why is 22q11.2 deletion syndrome important to identify during pregnancy?

Infants with the condition often have medical issues at birth, but some symptoms can be difficult to detect and the condition is often incorrectly diagnosed at first. **Identifying 22q11.2 deletion syndrome before birth allows for early care that can improve health outcomes for a child**.

Accurate diagnosis and early treatment can help a child reach their full potential by allowing providers to:

- Minimize seizures by treating low calcium levels early.
- Consider delivery at tertiary care center and plan surgeries for heart defects.
- Identify palate abnormalities to help breathing, feeding, and speech.
- Customize care strategies for immune deficiencies, developmental delays, and learning difficulties.

Scan to hear a mother's personal experience

https://natera.wistia.com/medias/qb2doh3aan

How do parents test for 22q11.2 deletion syndrome before birth?

Non-invasive prenatal testing, or **NIPT**, involves a simple blood draw from the pregnant mother

NIPT results indicate if there is an increased **chance for the baby to have a condition** and **whether additional diagnostic testing is needed.**

If NIPT results find an increased risk for 22q11.2 deletion syndrome, **diagnostic testing** with microarray analysis can tell for sure if the baby has the condition.

Diagnostic testing requires an invasive procedure, such as **amniocentesis**, which has a small chance of miscarriage and is usually not performed unless indicated by NIPT or suggestive ultrasound findings.

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