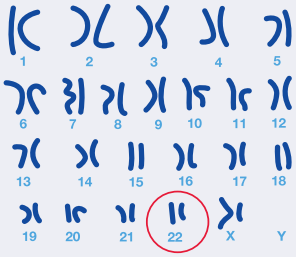
22q11.2 deletion syndrome

What is 22q11.2 deletion syndrome?



A person typically has **46 chromosomes** grouped into 23 pairs. 22q11.2 deletion syndrome is caused by a **missing piece of chromosome number 22,** which affects the development of various organs and systems in the body. This genetic change can cause an array of health issues including **congenital heart defects, immune deficiencies, hypocalcemia, palatal abnormalities, developmental delays, and behavioral and learning differences.**

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.**
- **Identify palate abnormalities to help breathing, feeding, and speech.**
- **Consider delivery at tertiary care center and plan surgeries for heart defects.**
- **Customize care strategies for immune deficiencies, developmental delays, and learning difficulties.**

Scan to hear a patient's personal experience

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 person.

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, further diagnostic testing (such as amniocentesis) may be recommended by your healthcare provider to confirm if the baby has the condition.

It is important to work closely with a healthcare provider to develop a personalized treatment plan. Genetic counselling may also be recommended to understand the condition and its inheritance patterns.

Certified Lab

natera™ | Panorama™ Next-generation NIPT

LifeLabs GENETICS®