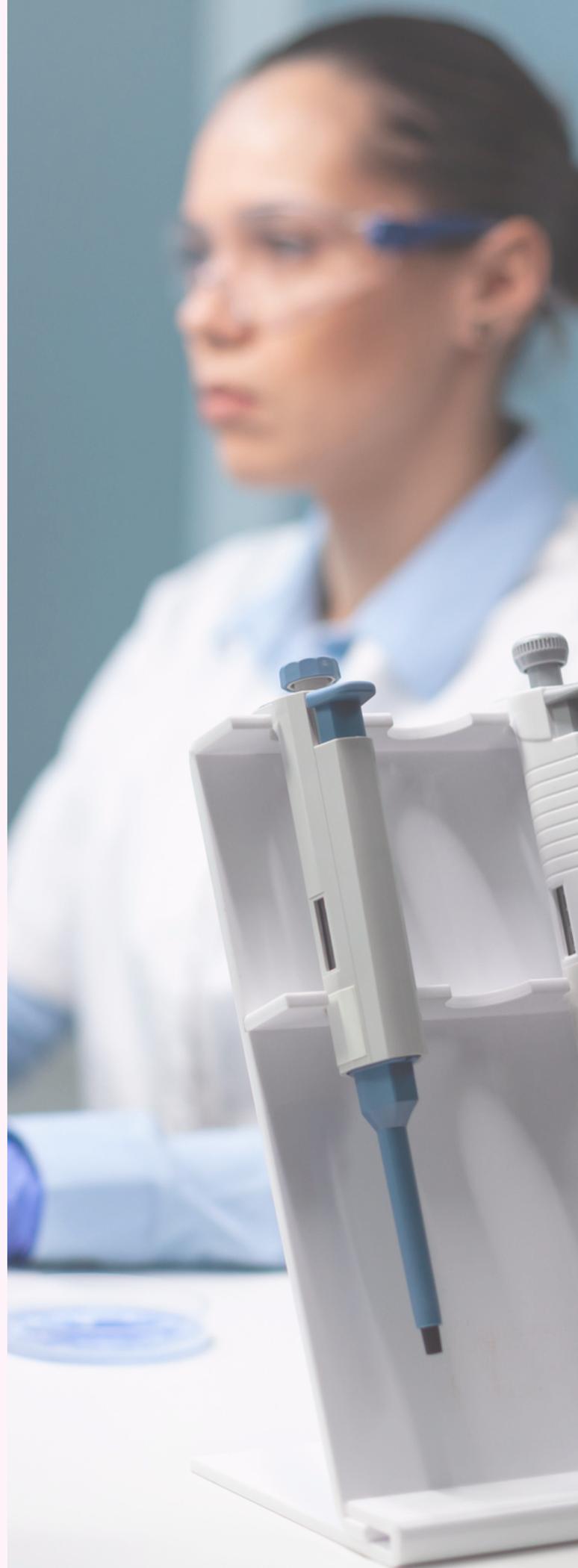


Genetic Conditions Screened by NIPS

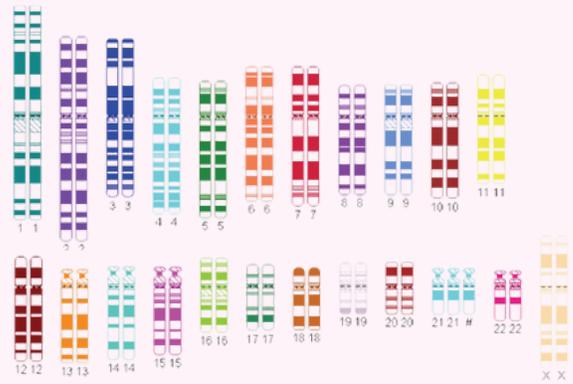


Most people have 23 pairs of **chromosomes** that, together, contain nearly all of their genetic information, or DNA. Sometimes, due to the genetic makeup of the parental sperm and egg prior to conception or through a variation that occurs during **fetal** development, a baby may have an extra or missing chromosome (**aneuploidy**). The most common type of chromosomal aneuploidy is a **trisomy** – when there are three copies of chromosome instead of the usual two.

Noninvasive prenatal screening (NIPS) can be used to assess the chance of a baby having a chromosomal aneuploidy, typically screening for the following, which are described in more detail below:

- Trisomy 21, which causes **Down syndrome**
- Trisomy 18, which causes **Edwards syndrome**
- Trisomy 13, which causes **Patau syndrome**

Other conditions that may be screened for by NIPS include changes to the number of sex chromosomes (e.g., Turner syndrome or Klinefelter syndrome) or conditions related to chromosomal **microdeletions** (when only a small piece of the chromosome is missing). However, the evidence and accuracy of NIPS to screen for these additional outcomes is limited.



I just want to know the sex.

Yes, NIPS can predict the sex of your baby before it is visible on an ultrasound, but this is NOT the purpose of NIPS. Before seeking out NIPS to determine the sex of your baby, consider ALL of the implications of undergoing screening, including what happens if you receive an abnormal result.

If you choose to undergo NIPS, talk to your health care provider and/or a **genetic counselor** about which conditions are appropriate to include in your screening. If the NIPS results indicate a high risk of an affected baby, confirmatory diagnostic testing (during pregnancy or after delivery) is highly recommended.



Down Syndrome (Trisomy 21)

Down syndrome is a condition that can result from someone having a full or partial extra copy of chromosome 21. This extra chromosome disrupts fetal development, resulting in features like reduced muscle tone, distinctive physical features, and intellectual and developmental disabilities. Down syndrome can increase the likelihood of developing hearing and vision problems or disorders of the endocrine or immune systems. However, with appropriate support and treatment, the life expectancy for someone with Down syndrome is over 60 years.

- Down syndrome occurs in about 1 in every 700 babies in the United States.²
- A prenatal ultrasound is expected to show a structural abnormality in 50% of fetuses confirmed with trisomy 21.³

Visit the [National Down Syndrome Society](#) to learn more about this condition.



Edwards Syndrome (Trisomy 18)

Edwards syndrome is a rare but serious condition that can result from having a full or partial extra copy of chromosome 18. Whether this chromosomal abnormality occurs in just some or all of the body's cells affects the severity of birth defects and intellectual disability. Pregnancies with trisomy 18 can spontaneously miscarry, and there is a high mortality rate for children with trisomy 18 immediately after birth. Between 5 and 10% of babies born with Edwards syndrome survive beyond the first year.

- Trisomy 18 occurs in 1 in 2,500 pregnancies in the United States.⁴
- A prenatal ultrasound is expected to show a developmental abnormality in the majority of fetuses confirmed with trisomy 18.⁵

Visit the [Trisomy 18 Foundation](#) to learn more about this condition.

Patau Syndrome (Trisomy 13)

Patau syndrome can result from someone having a full or partial extra copy of chromosome 13. It is the least common trisomy of the three commonly screened conditions, and it is the most severe. Neurological and heart defects and restricted growth in the womb contribute to the high mortality rate for children with trisomy 13, and long-term survival of babies born with Patau syndrome is rare.

- Trisomy 13 occurs in approximately 1 in 10,000 births.⁶
- A prenatal ultrasound is expected to show a developmental abnormality in more than 90% of fetuses confirmed with trisomy 13.⁷

Visit the [National Organization for Rare Disorders \(NORD\)](#) to learn more about this condition.



Always seek confirmation of a condition through follow-up diagnostic tests. Reproductive health care considerations and treatment decisions should never be made without receiving clinical and laboratory confirmation and, if possible, consulting with a genetics or maternal-fetal medicine health care provider.