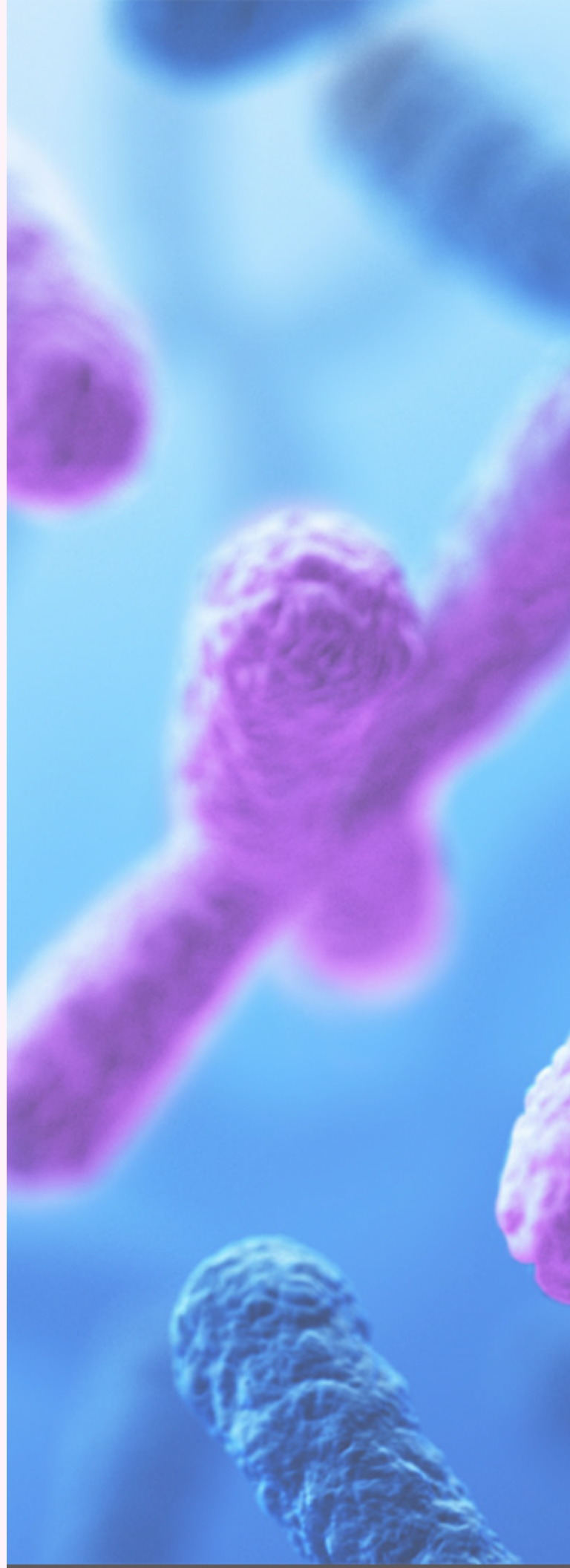


# Understanding Genetic Screening and NIPS



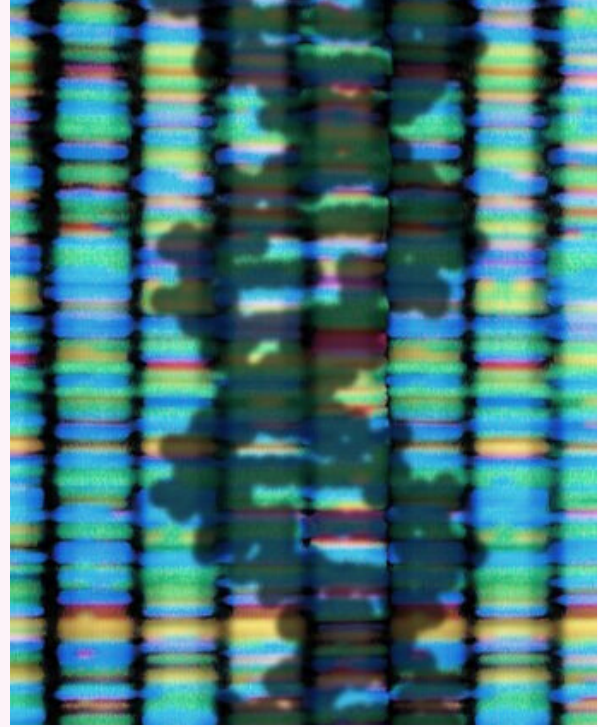
Each cell in the human body contains your genetic information, coded in strands of deoxyribonucleic acid (DNA). A complete set of DNA contains 46 **chromosomes**, combined from your biological parents – 23 from your mother and 23 from your father.

Variations or changes that have occurred in parent sperm and egg cells can sometimes result in clinically significant **chromosomal abnormalities**, such as an extra chromosome or a missing portion of a chromosome. These changes can affect the **fetus** (baby growing in the womb) during development or after birth. Occasionally, chromosomal changes can also happen after conception, when cells are rapidly dividing during fetal development.



**1 in 150** live births have chromosomal abnormalities.<sup>1</sup>

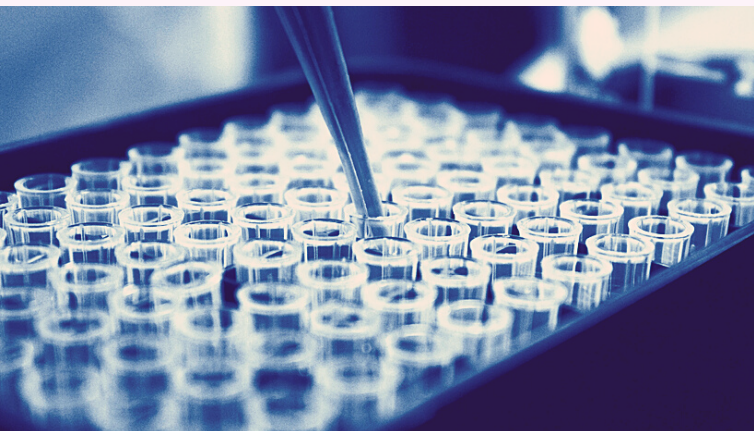
The occurrence of certain types of fetal chromosomal abnormalities increases as a woman gets older and is not related to race or ethnicity. Depending on the type of change in the chromosome, they can significantly influence the baby's health and well-being, particularly if an entire chromosome is added or missing.



## Genetic Screening

Screening for genetic disorders can be done using a number of different methods and varies depending on the type of disorder being screened. **Genetic screening** for chromosomal abnormalities often looks for specific sections of a person's DNA sequence to identify variations that are associated with certain genetic conditions. This screening is usually conducted with a blood sample or cheek swab, and can be done at different time points in a person's life, depending on what they are looking to assess. However, genetic screening is often associated with pregnancy and childbirth.

**Prenatal genetic screening** can offer expecting parents and those considering parenthood information about the chance of passing on certain conditions to their child, allowing women and their partners to make informed decisions about planning their family, pregnancy management, and future child-rearing. Screening can also be used to help those who are pregnant assess the risk of their baby having a chromosomal abnormality.



Different genetic screenings are available before, during, and after pregnancy – each offering unique insights:

- **Carrier screening** is used to determine the chance of **one or both parents** passing on certain inherited genetic conditions to a child. Ideally, this screening should be conducted before conception, but it can also be done during pregnancy. **Expanded carrier screening** includes a wider range of conditions than those included in carrier screening.
- **Noninvasive prenatal screening (NIPS)** is used to assess the risk of chromosomal **aneuploidy** – an irregular number of chromosomes – in the fetus. NIPS can be performed as early as 10 weeks into a pregnancy.
- **Newborn screening** is a national public health program that assesses all newborns for specific serious, but treatable conditions. Screening is typically performed 1-2 days after birth using blood taken by a small prick of a newborn's foot to test for metabolic, hormone, and hemoglobin (blood) disorders. Hearing and heart tests are also done to promote early detection, diagnosis, and treatment of certain defects. Newborn screening program requirements, the conditions screened, and screening methods may vary from state to state.

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**Genetic screening can also be used outside of pregnancy to assess risk for individuals with a personal or family history of certain cancers that have a hereditary component.**  
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## Noninvasive Prenatal Screening (NIPS)

Through the natural process of fetal growth and development during pregnancy, some of the tissue from the **placenta** (which contains similar DNA to fetal DNA) degrades, releasing that DNA into the mother's bloodstream. This **cell-free DNA (cfDNA)** can be examined by taking a blood sample from the mother and screening it for certain chromosomal changes. NIPS (also referred to as cfDNA screening) is considered noninvasive because the required blood draw poses minimal to no risk to mother or baby. Even though the blood draw is simple, NIPS should only be done after a woman is provided education and information about the process, risks, and benefits of NIPS, and gives her informed consent to proceed.





NIPS is not a required risk assessment, but experts agree that everyone should have access to this screening because of its accuracy in detecting pregnancies at increased risk for common chromosomal abnormalities in the fetus. As a result, NIPS may be considered more strongly under the following circumstances:

- If the mother is over the age of 35 at delivery
- A positive first or second trimester standard biomarker screening from initial blood work
- A fetal ultrasound that indicates an increased risk of aneuploidy
- A prior pregnancy with a **trisomy**
- If either parent has a chromosomal abnormality where there is an increased risk of fetal trisomy 13 (**Patau syndrome**) or trisomy 21 (**Down syndrome**)

## Screening vs. Diagnostic Testing

Genetic screening, including NIPS, only provides information about the risk of certain genetic conditions like Down syndrome. Screening results should never be interpreted as a concrete determination of your or your baby's health; however, results can be used to inform conversations with your provider, make decisions about managing maternal and infant health, and determine whether to undergo diagnostic testing.

**Screening is not the same as diagnostic testing.**

**Genetic screening results cannot provide a definitive diagnosis. NIPS results can provide information about the *likelihood* of your baby having a certain condition. Follow-up diagnostic tests help rule out **false-positive** or **false-negative** screening results.**

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It is important to talk to your health care provider about the purpose, potential risks, and benefits of genetic screening options before starting the process. A consultation with a genetic counselor, a specialist in genetics with advanced training in medical genetics and counseling, may also be very helpful before screening. For tips on how to have these conversations, visit the **Talking to Your Health Care Provider** section of the SWHR Noninvasive Prenatal Screening Resource Guide for Women.

