

# Congratulations On Your Pregnancy!

This is a time for careful planning. For the health and well-being of your family, genetic screening options are available to help you make informed family planning choices. **We encourage you to discuss these options with your healthcare provider to see if they might be right for you and your pregnancy.**

## Your genetic testing options include:

- Genetic Carrier Screening
- Non-Invasive Prenatal Screening
- Hereditary Cancer Testing

Depending on your family history, your provider may discuss additional genetic testing.

## Carrier Screening

**The American College of Obstetricians and Gynecologists recommends that "Information about genetic carrier screening should be provided to every pregnant woman."<sup>1</sup>**

- Carrier screening identifies if you and your reproductive partner are carriers of a recessive genetic condition (carrier couple) that you could pass onto your child.
- Generally, carriers of recessive genetic conditions are healthy, but if you and your partner are a "carrier couple" for the same genetic condition, your baby may be affected.
- Using a comprehensive carrier screen that can detect most carriers, regardless of ethnicity or family history, is highly recommended.

## Genetic Condition Fast Facts

- Most babies (80%) that are born with a genetic condition have no known family history of it.<sup>2</sup>
- 1 in 300 pregnancies will be affected by a recessive genetic condition.<sup>3</sup>
- Genetic conditions are becoming increasingly treatable when identified early.

**If you and your partner are a carrier couple, options you may consider include:**

### Pregnancy Considerations

Prenatal diagnosis

Improved pregnancy and postnatal planning

Earlier diagnosis and timely treatment

**NxGen MDx's Super Panel screens for 145 common genetic conditions**, providing you with actionable information to improve health outcomes for your child.

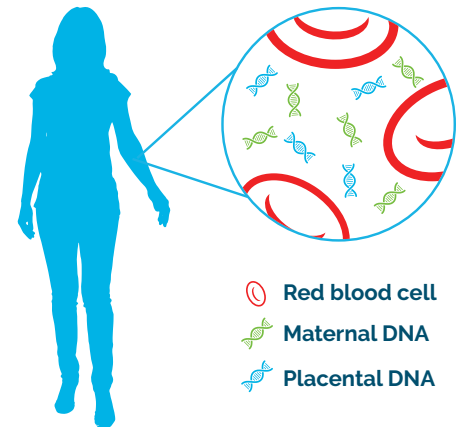
If you and your reproductive partner are identified as a carrier couple for the same condition, a genetic counselor can help you identify your options and how best to proceed.



# Non-Invasive Prenatal Screening (NIPS)

The American College of Obstetricians and Gynecologists states that  
"Cell-free DNA is the most sensitive and specific screening test for the common fetal aneuploidies."<sup>4</sup>

- Non-invasive prenatal screening (NIPS) is a genetic test that analyzes cell-free DNA that comes from the placenta and is found in the mother's blood.
- It can tell you if your baby has too few or too many chromosomes (aneuploidy) which can lead to health problems.
- An extra or missing chromosome can lead to several developmental and health issues.
- NIPS is non-invasive and does not jeopardize the health of your unborn baby.
- NxGen MDx's NIPS is available at or after 10 weeks into your pregnancy.
- NIPS helps identify pregnancies that may be at increased risk of certain chromosomal disorders.
- NIPS is not diagnostic, and if you receive a positive result, it should be confirmed through diagnostic testing.



## What is an Aneuploidy?

- An aneuploidy is when you have too many or too few chromosomes
- Typically, individuals have 2 copies of each chromosome (23 pairs, 46 chromosomes total)

### Possible Chromosome Arrangements



Typical Chromosome Arrangement



Monosomy (missing chromosome)



Trisomy (extra chromosome)

## Aneuploidies Screened for by NxGen MDx NIPS:

### Common Trisomies:

- 21 (Down Syndrome)
- 18 (Edwards Syndrome)
- 13 (Patau Syndrome)

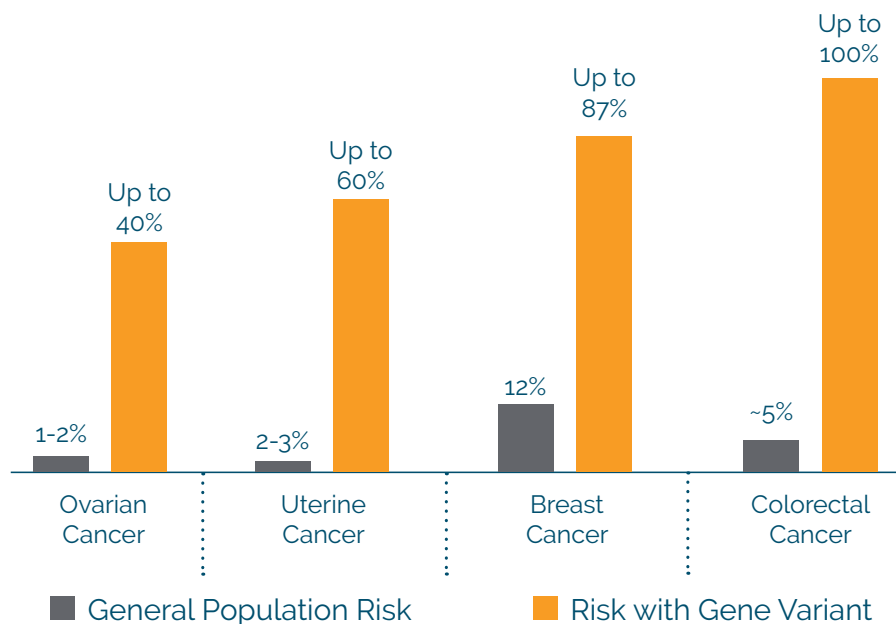
### Sex Chromosome Aneuploidies:

- Monosomy X (Turner Syndrome)
- Trisomy X (Triple X Syndrome)
- XXY Syndrome (Klinefelter Syndrome)
- XYY Syndrome (Jacobs Syndrome)

# Hereditary Cancer Testing

- While most cancers occur by chance, **approximately 10% of cancers are hereditary.**
- These hereditary cancers can be passed down from generation to generation.
- If you have a family history of multiple young or rare cancers, cancer genetic testing may be appropriate for you.
- Hereditary cancer testing will help you determine your personal risk of developing a hereditary cancer and can help you understand the likelihood of passing it on to the next generation.
- Your hereditary cancer test results can help you form a personalized cancer management plan to reduce your risk of developing a hereditary cancer.

## Lifetime Risks for Common Cancers<sup>5</sup>



[NxGen MDx's Hereditary Cancer Panel](#) tests for your genetic risk in 8 different types of cancers including breast, ovarian, uterine, gastric, colorectal, pancreatic, prostate, and melanoma.

[Click here to fill out the questionnaire and talk to your healthcare provider to see if Hereditary Cancer Testing might be appropriate for you.](#)

### Sources

1. Committee Opinion No. 691: Carrier Screening for Genetic Conditions. Obstet Gynecol. 2017 Mar;129(3):e41-55.
2. Blythe SA, Farrell PM. Advances in the diagnosis and management of cystic fibrosis. Clin Biochem. 1984 Oct;17(5):277-83.
3. Johansen Taber KA, Beauchamp KA, Lazarin GA, Muzzey D, Arjunan A, Goldberg JD. Clinical utility of expanded carrier screening: results-guided actionability and outcomes. Genet Med. 2019 May;21(5):1041-8.
4. ACOG Practice Bulletin 226
5. Adapted from NCCN data.