Prenatal Genetic Screening Information

The following screening tests can assess if there is an increased risk for your baby to have certain genetic conditions. No screening test is 100% accurate and no test can guarantee a healthy pregnancy. Test results may not be available for up to two weeks. If a test result reports an increased risk for these conditions, there will be an opportunity to discuss the results and consider further diagnostic testing options.

Most of the tests offered are considered standard of care and appropriate for all pregnancies. If you have questions, please contact your insurance company.

What should I consider when deciding whether to have prenatal genetic testing?

- Prenatal genetic screening is optional. It is your choice whether to have prenatal testing. Your personal beliefs and values are important factors in that decision.
- It can be helpful to consider how you would use the results of the prenatal screening tests in your pregnancy care. These screening tests will tell you if there is a higher risk of having a baby with a specific condition. A diagnostic test should be done if you want to know a more certain result.
- Some parents want to know if their baby will be born with a genetic condition before birth to learn more about the condition and to plan for medical care.
- There is no right or wrong decision.

General facts about genetics and chromosome disorders

- A normal human cell has 46 chromosomes, divided into 23 pairs. Chromosomes are the structures inside each cell which hold a person's DNA and genetic information.
- Missing or extra chromosomes, or a piece of a chromosome, can interfere with normal development.
- Many chromosomal abnormalities can lead to serious physical birth defects, intellectual and developmental disabilities, or both.
- Most cases are not inherited. These conditions can happen to any pregnancy, even without a family history.
- The most common chromosome abnormalities are Down Syndrome, Edward Syndrome and Patau Syndrome.

What are some of the genetic and chromosome disorders?

Down Syndrome (Trisomy 21)

- Caused by an extra #21 chromosome
- Occurs in about 1 out of 700 newborns
- Abnormal features of the face and body, medical problems such as heart defects, and learning problems/intellectual disabilities

Edward Syndrome (Trisomy 18)

- Caused by an extra #18 chromosome
- Occurs in 1 out of 5,000 newborns
- Serious birth defects and severe learning/intellectual disabilities
- Often pass away before or soon after birth

Patau Syndrome (Trisomy 13)

- Caused by an extra #13 chromosome
- Occurs in 1 out of 10,000 newborns
- Serious birth defects and severe learning/intellectual disabilities
- Often pass away before or soon after birth

Sex Chromosome Aneuploidy (SCA)

- Caused by extra or missing X and/or Y chromosomes rather than the typical female (XX) and male (XY) chromosomes
- Examples include Turner Syndrome (XO) and Klinefelter syndrome (XXY)
- Impact varies with some people not having any health or learning problems while others may have severe heart defects and other health challenges

Carrier Testing (Cystic Fibrosis, Spinal Muscular Atrophy)

- Detects whether you or your partner carry a mutation in a gene
- Can be done any time, ideally before pregnancy

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Types of Genetic Screening Tests

Non-Invasive Prenatal Screening (NIPS or Cell-free DNA Screening)*
NIPS can be performed after 10 weeks gestation. It is a blood test to screen for the most common autosomal fetal abnormalities: Down syndrome, Edwards syndrome, and Patau syndrome. It can also detect abnormalities of the sex chromosomes such as Turner syndrome (X0) and Klinefelter syndrome (XXY). NIPS can help determine fetal sex. However, if you do not wish to know the fetal sex, your healthcare provider can opt out of having the fetal sex reported. This test detects >99% of pregnancies with Down syndrome and approximately 95% of pregnancies with trisomy 18 and trisomy 13. The detection rate for sex chromosome differences is 90%. Results about whether a baby is a boy or a girl is about 99% accurate. If the screen yields a positive result, a follow-up diagnostic procedure may be recommended for definitive confirmation.

Sequential Screen
A nuchal Translucency ultrasound and blood work are used to determine your baby's chance of having Down Syndrome, Edwards Syndrome and Open Neural Tube Defects (Spina Bifida). Part 1 is completed around 11-13 weeks of pregnancy with blood drawn on same day as the ultrasound. Part 2 is completed around 16-18 weeks with blood work only. When parts 1 and 2 are combined, this test detects approximately 90% of cases of Down syndrome and Edwards syndrome. It detects about 80% of open neural tube defects. If the Part I result is abnormal, your provider will follow-up with you. Otherwise, you will receive the results after Part 2 is completed.

First Trimester Screen*
A nuchal Translucency ultrasound and blood work are used to determine your baby's chance of having Down Syndrome and Edwards Syndrome. Blood work is drawn around 11-13 weeks of pregnancy on same day as the ultrasound. This test detects about 83% of Down syndrome cases and 80% of Edwards syndrome cases.

Quad Screen/Second Trimester Screen
Blood work is used to determine your baby's chance of having Down Syndrome, Edward Syndrome and Open Neural Tube Defects (Spina Bifida). Blood work is drawn around 16-18 weeks of pregnancy. This test detects about 80% of cases of Down syndrome, Edwards syndrome and open neural tube defects.

**If you choose NIPS or the first trimester screen, a separate blood test to screen for open neural tube defects (spina bifida) is recommended in the second trimester.*

Cystic Fibrosis
Blood work is used to determine if alterations in the gene that causes cystic fibrosis (CF) are present. If both parents have an altered gene, their child might inherit CF. This test can be done at any time during pregnancy.

Spinal Muscular Atrophy
Blood work is used to determine if alterations in the gene that causes muscular atrophy (SMA) are present. If both parents have an altered gene, their child might inherit SMA. This test can be done at any time during pregnancy.

Anatomy Ultrasound
An anatomy ultrasound will be performed to screen for some birth defects.

CPT CODES: Call your insurance to check if these test are covered.	
Cystic Fibrosis Carrier Screen (CFVantage)	81220
Spinal Muscle Atrophy (SMA) Carrier Screen	81329, 81401
Hemoglobinopathy	83020, 85014, 85018, 85041
Sequential Integrated Screen, Part 1	84163, 84702
Sequential Integrated Screen, Part 2	82105, 82677, 84702, 86336
Qnatal (NIPT or cell-free DNA)	81420
Quad Screen/Second Trimester Screen	81511
Maternal Serum Alpha-Fetoprotein (AFP)	82105
Nuchal Translucency Ultrasound	76813

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