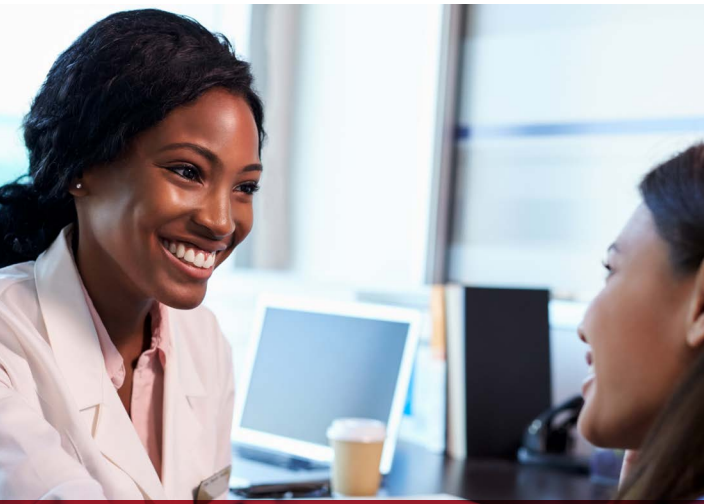
Weill Cornell Medicine
Obstetrics & Gynecology



Prenatal Genetic Counseling Program

What you should know about prenatal genetic screening

Call **212-746-3045** to Speak With a Genetic Counselor Today.

weillcornell.org

Pregnancy can be an exciting time for parents, but also a time of uncertainty—especially if there are genetic health concerns. The expert team of physicians and genetic counselors at the Weill Cornell Medicine Prenatal Genetic Counseling Program work closely with each patient to evaluate the chance that a pregnancy may be affected by a genetic condition, explain the limitations and benefits of genetic testing, and assist with the interpretation of genetic testing results.

Genetic counselors are healthcare professionals with specialized education and training in genetics who provide information to guide and support decisions being made about one's genetic health. You may consider seeing a genetic counselor if you:

- Want more information regarding genetic screening or diagnostic testing in pregnancy
- Have received an abnormal screening or ultrasound result in pregnancy
- Are concerned about a genetic condition or disease in your family
- Or your partner is a carrier for a genetic condition

Genetic Screening in Pregnancy

Nuchal Translucency Measurement

- Sonogram at approximately 11–14 weeks
- Abnormal nuchal measurement can be associated with chromosome conditions

First Trimester/Nuchal Screen

- Finger prick blood test
- Screens for Down Syndrome, Trisomy 13 and Trisomy 18
- Considered less accurate than cell-free DNA

Cell free DNA/Non-Invasive Prenatal Testing (NIPT)

- Blood draw as early as 9–10 weeks gestation
- Screens for Down Syndrome, Trisomy 13 and Trisomy 18 and sometimes abnormalities of the sex chromosomes (e.g. Turner Syndrome, Klinefelter Syndrome)
- High sensitivity but does NOT replace diagnostic testing
- Tests maternal blood for free floating (cell free) placental DNA

Diagnostic Genetic Testing in Pregnancy

Chorionic Villus Samplings (CVS)

- Done at approximately 10–13 weeks gestation
- Placental sample obtained for genetic testing
- Types of testing that can be done on sample:
  - Chromosome analysis/karyotype
  - Microarray
  - Mutation specific testing (if warranted)

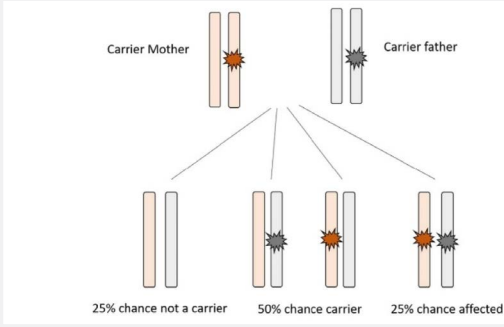
Amniocentesis

- Can be done as early as 15 weeks gestation
- Small sample of amniotic fluid obtained for testing
- Types of testing that can be done on sample:
  - Chromosome analysis/karyotype
  - Microarray
  - AFP testing
  - Mutation specific testing (if warranted)

Speak to your OB/genetic counselor about limitations and risks with CVS and amniocentesis to determine which of these options is best for you.

Carrier Screening

- Blood test on parents before/early in pregnancy
- If both parents are carriers for the same genetic condition, they are at increased risk for having a child with that disease
- Examples of common diseases tested for carrier screening include:
  - Cystic Fibrosis
  - Alpha Thalassemia
  - Spinal Muscular Atrophy
  - Tay Sachs Disease



Speak with your OB/genetic counselor about which carrier screening panel is right for you.

We encourage patients to speak with their healthcare provider about their family history and what genetic screening can help reveal.

To schedule an appointment with a genetic counselor, please call 212-746-3045

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