



Genetic Testing Options

Screening for risk of Down syndrome (Trisomy 21), Trisomy 18, and Trisomy 13

Invasive tests: amniocentesis and CVS; these are the most accurate tests and considered diagnostic.

Non-invasive screening tests:

- Cell-free DNA (maternal blood test)
 - Detection rate of 99% with a less 1% false positive rate
- First trimester screen (US and maternal blood test)
 - Detection rate of 85% with a 5% false positive rate
- If there is a positive screen, further testing may be recommended, such as invasive tests, Level 2 US, or MFM referral. This screening is recommended during each pregnancy.

May –Grant Recommends these screening tests:

- 1. Ultrasound at 12-13.6 weeks gestation**
- 2. Cell-free DNA testing (NIPS) at the same time. If the testing is cost prohibitive, blood work for the 1st trimester screen can be done instead. If you are pregnant with twins, we may recommend the first trimester screen blood work instead of the NIPS.**

Genetic Carrier Screening:

A genetic carrier is an individual who usually shows no signs of a genetic disease but may pass those genes on to a child. If that child is conceived with a partner who is also a carrier for the same genetic disorder, the child may be at high risk to have the disease. The likelihood of having a positive test result increases as more conditions are tested for, however, the chances of conceiving a child with the disease are still low.

This screening is generally a one-time screening which can be done anytime (ideally before pregnancy). If new mutations are identified in the future, new testing could be recommended. If there is a positive result, a referral to genetic counseling will be made.

Insurance coverage may vary, so please discuss anticipated coverage and out-of-pocket costs with your insurance company.

Codes:

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|---|--|
| 1. Non-Invasive prenatal screen (NIPS) /
cell-free DNA
81420
81422 | 3. First trimester screen:
84163
84704
82105
36415
76813 (ultrasound portion - single baby)
76814 (ultrasound portion - twins) |
| 2. Carrier Screening
81220 | |

MAY-GRANT IS A LEADING OBGYN PROVIDER IN LANCASTER AND LEBANON COUNTY
AND A PROUD MEMBER OF

