

Genetic Testing Options

Screening for risk of Downs 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:

- First trimester screen (US and maternal blood test)
 - Detection rate of 85% with a 5% false positive rate
- Cell-free DNA (maternal blood test)
 - Detection rate of 99% with a less 1% 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 at the same time we are currently offering Prequel. If Prequel 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 Prequel.

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.

For more information about these tests go to: https://myriadwomenshealth.com

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

Codes:

1. First trimester screen:

84163

84704

82105

36415 76813 (ultrasound portion - single baby)

76814 (ultrasound portion - twins)

2. Preguel cell-free DNA

81507

76813

3. Foresight 81162

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