homebirth & women's health

WomanWise

Informed Choice: Genetic Screening in Pregnancy

Informed Choice for Genetic Screening in Pregnancy Please read the following information and do your own research before considering whether or not you wish to have genetic screening done during this pregnancy. Please fill in the following so we can, together, determine any factors that may influence your decision to do genetic screening for this pregnancy.

Yes	No	Include client, baby's father, or anyone in either family with:
		Client aged ≥ 35 years by estimated due time
		Thalassemia (Italian, Greek, Mediterranean, or Asian descent) MCV <8012
		Neural tube defect (meningomyelocele, spina bifida, or anencephaly)
		Congenital heart defect
		Down syndrome
		Tay-Sachs (Jewish, Cajun, or French Canadian descent)
		Canavan disease
		Familial dysautonomia (Ashkenazi Jewish)
		Sickle cell disease (African descent)
		Hemophilia or other blood disorders
		Muscular dystrophy
		Cystic fibrosis
		Huntington's chorea
		Mental retardation / autism
		Other inherited chromosomal or genetic disorder
		Maternal metabolic disorder (i.e. Type I diabetes)
		Previous child with a birth defect
		Recurrent pregnancy loss or stillbirth
		Use of contraindicated supplements, herbs, OTC drugs and/or recreational drugs since last meses

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What is genetic screening?

Screenings assess the degree of risk, or chance, that the baby may potentially have certain common birth defects. As a risk ratio these screenings are not diagnostic and cannot tell with certainty if the baby actually has any problems. In the event of a positive test result, further testing can be arranged if you so choose.

Something to consider: *It is important to remember that each of these tests carries both a risk of false negatives (the test comes back "negative" but the baby does in fact have a genetic condition) and of false positives (the test comes back "positive" but the baby is in fact completely healthy.) There is also the possibility that your baby may have another genetic condition that is not screened for in any of these tests.*

What conditions are screened for and how?

- **First trimester screening** looks for Down syndrome and trisomy 13 and 18. One in every 691 babies born in the U.S. will have Down syndrome, making it the most common genetic condition. Trisomy 13 effects 1 in every 5,000 babies, and trisomy 18 affects 1 in every 6,000 babies.

Each of these ratios are based on a variety of factors including family history and maternal age, your chances of having a baby with one of these genetic conditions may be either higher or lower.

- **The first trimester screen** is a maternal blood draw done between 11 and 13 weeks of pregnancy. Mom's blood is sent to a lab where specific proteins are analyzed and those results are considered with the results of a specialized ultrasound to look for the presence of certain genetic markers. After this screening parents are given a risk ratio (i.e. 1:250 chance) that their baby may have a genetic disorder.

● **Second trimester screening** (also known as the quad screen) is another maternal blood draw and is done between 15 and 20 weeks of pregnancy. It screens for Down syndrome, trisomy 18, and neural tube defects such as spina bifida. One in every 1,000 babies are born with an open neural tube defect.

Something to consider*: Taking a supplement of 400 mcg of folate daily before becoming pregnant and during your first trimester can greatly reduce the risk of your baby having an open neural tube defect. Folate is naturally occurring in beans, citrus fruits, egg yolks and dark green vegetables.*

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- **Sequential Integrated Screening** is the most accurate at detecting the likelihood of Down syndrome and trisomy 18, because it calculates the chance of a genetic condition based on the findings of both the first and second trimester screenings.

- **Non-invasive prenatal screening** looks for markers indicating a higher possibility for genetic conditions including Down syndrome, trisomy 13, and trisomy 18, as well as inherited diseases such as cystic fibrosis, hemophilia, and other conditions. It is a different kind of blood test, which is far more sensitive than the blood tests performed in the 1st or 2nd Trimester Screening because it looks at baby's DNA rather than specific proteins found in maternal blood. *It is important to note that this test is relatively new and not currently covered by insurance companies for women who aren't considered at elevated risk.*

What are the risks & benefits of screening?
These tests are all considered safe and pose no risk of miscarriage or other pregnancy complications. Benefits of having the testing done include either putting the parent's minds at rest with a "negative" result; or providing the time to make the best decisions for your family regarding how to proceed and as needed, what kind of post-birth care plans (such as therapies and/or surgeries) to put in place should the test come back "positive."

What happens if my screen is positive?
If you choose to have genetic screening done and the results come back positive, you can choose to meet with a genetic counselor and/or have further diagnostic testing (amniocentesis or chorionic villus sampling.) You can also choose to not do any further testing, regardless of the initial screening results.

Amniocentesis & Chorionic Villus Sampling
Both of these diagnostic tests will give you far more specific information on exactly what, if anything, is going on with your baby. With that information, you can work to make the best decisions for yourself and your family regarding the continuation of the pregnancy and/or plans for birth location. You can also begin researching how best to care for a child with special needs, as well as begin finding appropriate specialists, therapists, and other resources.

CVS is usually done between 10 and 12 weeks of pregnancy and involves the removal of a small piece of the placenta through your cervix (via a thin tube and guided by ultrasound) which is then tested. The risk of miscarriage after CVS is 1.9%.

Amniocentesis is usually done between weeks 15 and 20 of pregnancy and involves taking a sample of amniotic fluid via a long needle placed with the guidance of ultrasound through your abdomen. The risk of miscarriage after amniocentesis is 1.4%.

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Please sign and date below to indicate your decisions regarding Genetic Screening during your pregnancy.

I have read WomanWise's informed choice document regarding genetic testing during pregnancy and have had an opportunity to ask questions and have had them answered to my satisfaction.

____I wish to do the following Genetic Screening:_____

____I wish to decline all Genetic Screening at this time

Client's signature _____Date _____

Client's name _____

Partner's signature _____

Midwife's signature_____ Date _____