Southwest Midwives

Genetic Screening in Pregnancy

What is genetic screening?

Genetic screening are tests that can be done during your pregnancy that give you information about whether your baby has a higher risk of genetic disorders. These tests are not diagnostic meaning:

- A positive screening test result for aneuploidy means that your fetus is at higher risk of having the disorder compared with the general population. It does not mean that your baby definitely has the disorder.
- A negative result means that your fetus is at lower risk of having the disorder compared with the general population. It does not rule out the possibility that your baby has the disorder.

Why should I get genetic screening?

This is a completely individual decision. We recommend you considering the following:

- What would I do with the information?
- Am I at high risk for genetic disorders (over the age of 35)?

What are my options?

Options for genetic screening are listed below. Costs are approximate and depend on your insurance plan and risk factors. Most insurance companies will pay for the sequential screen if you are under 35. Many insurance companies will cover cell free DNA testing if you are over the age of 35. If cost is a factor, talk to your midwife or call your insurance company prior to deciding on genetic screening. If you choose to decline genetic screening, your 20 week ultrasound will check the growth and development of your baby, check for birth defects and may show markers for genetic conditions, such as Down syndrome.

Name of Test	Timing	What is tests for	Accuracy	How it's done
Sequential Screen	Part 1: 11-13 weeks	Trisomy 18 Trisomy 21	 80% 70%	Blood test and ultrasound to measure nuchal translucency
	Part 2, aka "AFP": 15-20 weeks	Trisomy 13 Trisomy 18 Trisomy 21 Neural Tube Defects	90% 80%	
Second Trimester Screen or "Quad Screen"	15-20 weeks	Trisomy 18 &21 and neural tube defects	80%	
Cell Free DNA testing ("Myiad")	Anytime after 10 weeks	Trisomy 13, 18 & 21 and sex chromosome abnormalities	99%	Blood test. Visit www.myriad.com for more information
Carrier Screening	Anytime- even prior to pregnancy	Many genetic disorders- most common are Cystic Fibrosis (CF) and Spinal Muscular Atrophy (SMA)	99.9%	Blood test. Visit www.myriad.com for more information

For more information about genetic testing: https://www.acog.org/womens-health/faqs/prenatal-genetic-screening-tests

Genetic Conditions

Name of Disease	Average Risk	Age Related Risk	Symptoms of Condition
Trisomy 21 (Down Syndrome)	1 in 7,000	20 -1 in 2,000 30 - 1 in 900 35 - 1 in 350 40 - 1 in 100 45 -1 in 30	Intellectual disability, heart conditions, intestinal defects, hearing/vision defects. Ranges in severity
Trisomy 18 (Edward's Syndrome)	1 in 5,000		Severe intellectual disability and physical deformities. Only 1/10 live past 1 year of life.
Trisomy 13 (Pateau Syndrome)	1 in 10,000		Severe intellectual disability and physical deformity. Only 1/10 live past 1 year
Open Neural Tube Defects (such as Spina Bifida)	1 in 1,500	No change in risk with age, increased risk with obesity, diabetes, fever and folate deficiency	Varies with severity of defect
Cystic Fibrosis (CF)	1 in 30 Americans are carriers. If both parents are carriers, there is 25% chance baby will be affected.	No change	Abnormal mucous production, breathing difficulties leading to permanent damage, poor digestion and malabsorption

For information on sex-linked and microdeletion conditions, see the comprehensive list at www.myriad.com