Partners
Obstetrics & Gynecology, PA
specialty care for women
a division of Premier OBGYN of Minnesota

Testing for Birth Defects in Pregnancy

There are various tests that could be employed to test for birth defects in pregnancy. These tests can vary in method, in accuracy, and even in the risks associated with just doing the test itself. Each test is most accurate at specific points in the pregnancy and may or may not be covered by your insurance. We urge you to check on insurance coverage beforehand.

The most common abnormalities that can be tests for are Down syndrome (Trisomy 21), Trisomy 18 (Edwards syndrome), Trisomy 13 (Patau syndrome), open neural tube defects (open spina bifida and anencephaly), and cystic fibrosis.

The trisomy's are caused by genetic abnormalities in the number of chromosomes. Chromosomes are structures that are inside every cell of the body. They hold our genes that control how the body grows and develops. The genes are inherited from both the mother and the father. Most people have 23 pairs of chromosomes, all of which carry many genes. The first 22 pairs are the same in males and females. The last pair is the sex chromosomes, X and Y. Females have 2 X's normally and males have one X and one Y. Some people are born with extra or missing chromosomes. Having three copies of a chromosome instead of two is called a trisomy. People can also be born with an extra or missing sex chromosome.

Chromosomal abnormalities increase with maternal age, but women of all ages can have a baby with this type of abnormality, and it can occur in all races. Most pregnancies that have a chromosomal abnormality will be miscarried in the first trimester, but some will progress into later pregnancy and give birth.

Down syndrome (Trisomy 21) is the most commonly occurring chromosome abnormality and is found in one in every 700 babies. Down syndrome results in developmental problems and a higher risk of conditions including heart defects, mental retardation, breathing and hearing problems, and childhood leukemia. The severity of these conditions varies widely from individual to individual.

Trisomy 18 (Edward syndrome) is the second most common trisomy and affects 1 in every 3,000 babies. Babies with this condition often have multiple birth defects and many don't survive the first few months of life. Those who do survive have serious health problems from deformities of the heart, intestines, esophagus, hands, feet and kidneys and typically do not talk or walk. There is usually profound mental retardation.

Trisomy 13 (Patau syndrome) occurs in 1 in every 16,000 babies, (far less common than Down's and Edwards syndromes). These babies also often don't survive the first few months of life as they are profoundly intellectually and physically impaired and have multiple birth defects.

Open neural tube defects result from improper development of the brain and spinal cord which causes an opening to remain along the spine or head after the baby is born. The problems for the baby can be minor or major depending on the size and location of the opening. Walking and muscle tone may be affected as well as bladder and bowel function.

Cystic fibrosis (CF) is a genetically inherited disease that affects sweat and mucous producing glands in the body. In CF, mucous can be very thick and sticky and collect in the lungs, pancreas, intestines, liver and sinuses, impairing breathing and digestion. It also affects the ability to keep salt in our bodies when a person sweats, causing problems with hydration, heart rate, blood pressure, and more. It is a life-threatening condition and occurs in about 1 in 3,300 Caucasians in the U.S. The average lifespan of a person with CF is 37 years.

The testing options are generally broken down into two categories: Diagnostic Tests and Screening Tests. Diagnostic tests are nearly 100% accurate, whereas screenings give the risk of having certain birth defects. Screening tests will also give some results that are either falsely positive (meaning the test shows presence of the abnormality when in fact there is none) or falsely negative (test shows no abnormality when in fact there is one).

Mother's A	Risk of Down Syndrome	Risk of Any Chromosomal Disorder
20	1/1,667	1/526
25	1/1,250	1/476
30	1/952	1/385
35	1/378	1/192
36	1/289	1/156
37	1/224	1/127
38	1/173	1/102
39	1/136	1/83
40	1/106	1/66
41	1/82	1/53
42	1/63	1/42
43	1/49	1/33
44	1/38	1/26
45	1/30	1/21