



Prenatal Screening for Chromosomal Abnormalities

Chromosomes are the structures that contain our genetic information. Chromosomal abnormalities are missing, extra or irregular portions of chromosomal DNA. Genetic disorders may be inherited or they may occur without a family history. Some genetic disorders are more likely if you have a certain ethnic background or if you have a family history of a disorder.

The chromosomal abnormalities most commonly screened for are Trisomy 21 (Down syndrome), Trisomy 13 and Trisomy 18.

Mercy offers all women screening or diagnostic testing for fetal genetic disorders with each pregnancy, regardless of maternal age. However, age and family history increase the risk of chromosomal abnormality. For instance, a woman who is age 20 at term has a 1 in 1,480 chance of having a baby with Down syndrome, but a woman age 40 at term has a 1 in 85 chance.

Although birth defects can occur with chromosomal abnormalities, not all birth defects are related to chromosomal abnormalities. Most birth defects are the result of many factors, such as genetics, environment, medical conditions and behaviors. Common factors that can increase the risk of birth defects include smoking, drinking alcohol, taking “street” drugs, certain medical conditions (i.e. uncontrolled diabetes, obesity) and some medications.

Not all birth defects can be prevented, but there are things that can be done before and during pregnancy to increase the likelihood of a healthy pregnancy and baby:

- Start prenatal care as soon as you think you’re pregnant and be sure to see your health care provider regularly.
- Take a prenatal vitamin that has at least 400 micrograms (mcg) of folic acid every day starting at least a month prior to conceiving.
- Avoid alcohol, smoking and illegal drugs.
- Talk to your health care provider about medications you are currently taking and how they may affect a fetus.
- Talk to your health care provider about managing current medical conditions prior to conceiving.

Risk Factors for Genetic Abnormalities

- Older maternal age
- Older paternal age
- Parental carrier of chromosome rearrangement
- Parental carrier of genetic disorder
- Previous fetus or child with chromosomal abnormality
- Structural irregularities on ultrasound

If you would like to have a prenatal genetic screening, please let your Mercy OB provider know.



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Prenatal Genetic Screenings and Tests

Test	When Performed	Purpose	Detection Rates
First Trimester Screening	10 - 14 weeks	Blood test and ultrasound to screen for Down syndrome and Trisomy 18	82 - 87%
Cell-free DNA (NIPS)	10 - 36 weeks	Blood test to screen for Down syndrome, Trisomy 13 and 18	99% Down syndrome and Trisomy 13 96% Trisomy 18
Maternal Serum Alpha-Feto-protein Screening (MSAFP)	15 - 21 weeks	Screen for open tube defects (e.g. spina bifida)	80% open neural tube defect 5% false positive rate
Comprehensive Ultrasound	18 - 22 weeks	Assess major anatomical structures	
Chorionic Villus Sampling (CVS)	10 - 13 weeks	Small sample of tissue from placenta to evaluate baby's chromosomes	
Amniocentesis	15 - 24 weeks	Sample of amniotic fluid to evaluate baby's chromosomes and measure Alpha-fetoprotein (AFP) to evaluate for open neural tube defect	

Carrier Screening

Carrier screening allows you to find out if you are a carrier of certain genetic disorders. A carrier usually has no symptoms of a disorder but either parent can pass on the gene for that disorder to a child. Unlike prenatal screening tests, carrier screening only needs to be done once.

The American College of Obstetrics and Gynecology (ACOG) recommends offering prenatal screening for the following genetic conditions:

Spinal Muscular Atrophy (SMA): A genetic disorder that affects the nervous system that controls skeletal muscles and leads to muscle weakness. Incidence is 1 in 6,000 to 1 in 10,000 live births, and is reported to be the leading genetic cause of infant mortality. Carrier frequency may be as high as 1 in 40. Transmission is autosomal recessive, meaning that the condition requires genes inherited from both parents. As of 2019, SMA screening is now part of Missouri's newborn screening, but it's not available on newborn screening in all states.

Cystic Fibrosis (CF): A genetic disorder that affects mainly the lungs, but also the digestive tract and reproduction. Incidence is 1 in 2,500 and carrier frequency may be as high as 1 in 25. Transmission is autosomal recessive. All babies born in the state of Missouri and Illinois are screened for CF.

Hemoglobinopathies: Genetic disorders that affect the shape of hemoglobin with subsequent anemia. These include sickle cell, alpha-thalassemia and beta-thalassemia. Your doctor will discuss your medical history and perform a complete blood count during pregnancy to assess risk.

Fragile X Syndrome: This is the most commonly inherited form of intellectual disability and a known cause of autism. Screening is recommended for women with a family history of Fragile X, suggestive of Fragile X, or unexplained ovarian insufficiency.

Other potential screenings:

Extended Screening in Ashkenazi Jewish population: Recommended screening includes Tay-Sachs, Familial Dysautonomia, Cystic Fibrosis and Canavan.



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