Genetic Conditions for Which Screening is Available

The following list shows some genetic conditions for which screening is available. Screening for these conditions may not be covered by your insurance.

Condition Name	Condition Description	At Risk Group	Approximate Population Risk
Down Syndrome	Down Syndrome (DS) is a chromosome disorder that presents with a combination of birth defects including mental retardation (which can range from mild to moderate to severe) and characteristic facial features. Approximately 30 to 50 percent of babies with DS have heart defects and other health issues. Individuals with DS can live into adulthood. The risk of Down Syndrome increases with maternal age.	All Populations	Age Dependent
Trisomy 18	Trisomy 18 is a chromosome disorder with a spectrum of severe birth defects including heart defects and severe mental retardation. The majority of babies with this disorder are stillborn or die shortly after birth. The risk of Trisomy 18 increases with maternal age.	All Populations	Age Dependent
Neural Tube Defects	A neural tube defect is an opening that remains along part of the baby's spine (spina bifida) or head (anencephaly) if the neural tube does not completely close during early development of the embryo. Symptoms of spina bifida can include paralysis of the legs, lack of bowel or bladder control and hydrocephalus. Fetuses with anencephaly are often stillborn or die shortly after birth. Surgery can repair the opening in babies with spina bifida, however the nerve damage cannot be repaired.	All Populations	1:1,000
Cystic Fibrosis	Cystic Fibrosis (CF) causes the body to produce large amounts of thick mucus usually affecting the lungs and digestive tract that can lead to chronic lung problems and poor growth. There is no cure for CF currently, however life expectancy has improved with many people living into their 20's or 30's and beyond.	Caucasian	1:2,500
Tay-Sachs	Tay-Sachs (TS) is a progressive neurological disorder. A baby with TS develops normally for the first few months, but then both mental and physical abilities deteriorate. There is no treatment and most children with TS die by age 5. Additional testing may be indicated for families in which both parents are of Jewish ancestry.	Eastern European Jewish, French-Canadian	1:3,600
Alpha Thalassemia	Alpha Thalassemia is an inherited blood disorder. It can cause severe problems for babies before they are born and is associated with stillbirth. It causes anemia, liver and spleen enlargement and bone abnormalities. There is no cure for Alpha Thalassemia at this time.	Southeast Asian	1:2,500
Beta Thalassemia	Beta Thalassemia is an inherited blood disorder. Children with this disorder may appear normal at birth, but during the first or second year may develop poor appetite, frequent infections and may develop problems with their liver, spleen and other parts of their body. There is no cure for Beta Thalassemia at this time.	Mediterranean, Asian	1:3,600

Sickle-Cell Anemia	Sickle-Cell Anemia is a hereditary chronic form of anemia that can cause pain or damage to vital organs, risk of serious infection and can lead to early death. There is no drug or therapy that can correct the disease, however therapies for reducing the severity and frequency of pain are available.	African-American, Hispanic	1:600
Fragile X Syndrome	Fragile X syndrome is a disorder that causes mental retardation and autism. It primarily affects males. Women who are carriers are at risk to have a child with this condition.	All Populations	1:26 (carrier females) 1:2,000 (affected males)
Spinal Muscular Atrophy (SMA)	SMA destroys nerve cells that affect voluntary movement. Infants with SMA have difficulty breathing, swallowing, head and neck control, crawling and walking. The most common form is generally lethal and affects infants in the first months of life. Less commonly, the disease starts later and individuals can survive into adulthood. SMA does not affect intelligence. There is no cure at this time.	All Populations	1:6,400