

Desert
Perinatal
ASSOCIATES



A
helpful
guide
to
Carrier Testing

for Common

Genetic Diseases

Hemoglobinopathies

Cystic Fibrosis

Spinal Muscular Atrophy

Fragile X



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Please keep this brochure as a reference.

In many different ethnic groups there are common inherited diseases, such as hemoglobinopathies, cystic fibrosis, spinal muscular atrophy, and fragile X syndrome (additional information on back). Carrier screening can identify couples in the general population at risk of having a child with any of these severe genetic disorders.

Who should consider carrier testing?

Everyone has some chance of being a carrier of one of these common genetic conditions. Carriers of abnormal genes generally have no symptoms of the disease. A carrier may not have a family history of the disorder and could already have healthy children. However, if there is a family history of these disorders, the chance of being a carrier is increased. In these cases, information on the family history should be discussed with a physician or genetic counselor at Desert Perinatal Associates.

Carrier screening is optional; each patient or couple can decide which screening tests are most appropriate based on their ethnic background and/or family history.



How is testing done?

Carrier screening involves a blood draw from one or both parents.

What does a positive carrier screen mean?

If one person is identified as a carrier it is important that their partner completes testing. Both parents must be carriers of the same disorder in order to have an affected child. When one person is a carrier, but their partner has a negative test result and no family history, the chance that their child will be affected is significantly decreased. It is also important to share this information with other family members so they can consider testing.

What does a negative carrier screen mean?

It is important to understand that screening does not detect all carriers. A negative screen result significantly lowers the risk of being a carrier and having an affected child, however the risk cannot be completely eliminated.

When should carrier testing be done?

Testing can be completed at any time. Ideally, couples should be tested before becoming pregnant or early in their pregnancy. If carrier couples are identified during a pregnancy they would be eligible for genetic counseling and could consider prenatal diagnostic studies by CVS or amniocentesis. Preconception carrier screening allows carrier couples to consider the fullest range of reproductive options. A person's carrier screen results will not change, so this testing does not need to be repeated in future pregnancies.

Is it covered by insurance?

Most insurance companies cover carrier screening for pregnant couples or those considering pregnancy. Cost and insurance coverage for carrier screening varies depending upon the laboratory and insurance policy.

The genetic counselors at Desert Perinatal Associates can provide additional information about carrier testing and the common genetic diseases.

Hemoglobinopathies*

Clinical Features

Caused by an abnormal amount or misshapen hemoglobin molecule in red blood cells; red blood cells carry oxygen

Sickle cell disease causes severe anemia, pain episodes, and a weakened immune system

Thalassemia major is characterized by liver/spleen damage and chronic anemia that ranges in severity; regular blood transfusions may be required to treat the severest forms

Other hemoglobinopathies are less common and may be associated with milder anemia

Inheritance

Autosomal recessive

Carrier Frequency

Sickle cell trait: 1 in 12

Alpha thalassemia minor: 1 in 20

Beta thalassemia minor: 1 in 20-30

Disease Incidence

1 in 200

High Risk Groups

Southeast Asian
Chinese
East Indian
African
Hispanic
Middle Eastern
Mediterranean (Greek, Italian)

Testing

Hemoglobin electrophoresis and CBC can detect abnormal red blood cells

Detection rates vary by condition and ethnic group; high detection rates overall

Cystic Fibrosis*

Clinical Features

Leads to a build-up of thick mucus in the lungs and digestive system; causes lung infections, digestive problems, and poor growth

Some milder forms may be associated with pancreatitis and male infertility

Intelligence is normal

Lifespan is shortened; current life expectancy is 37 years

Inheritance

Autosomal recessive

Carrier Frequency

1 in 30 average in US

Varies by ethnic group; highest in Caucasians

Disease Incidence

1 in 3,500

High Risk Groups

Northern European
Jewish
Hispanic

Testing

Genetic testing can detect approximately 80-90% of carriers; differs by ethnic group

Screens for the most common genetic mutations; a negative result reduces risk but cannot eliminate risk

Spinal Muscular Atrophy*

Clinical Features

Progressive weakness of lower motor nerves; variable severity and age of onset

Type I is characterized by severe muscle weakness beginning at birth; death typically results from respiratory failure by two years of age

Type II has significant muscle weakness (able to sit, cannot stand or walk without assistance); may survive beyond 4 years of age

Type III is milder; children can learn to walk unaided

Intelligence is normal

Inheritance

Autosomal recessive

Carrier Frequency

1 in 41

Disease Incidence

1 in 6,000 - 10,000

High Risk Groups

All ethnicities

Testing

Genetic testing can detect 94% of carriers

Screens for the most common genetic mutations; a negative result reduces risk but cannot eliminate risk

Fragile X Syndrome

Clinical Features

Males with a full mutation have mental retardation, ranges from learning disability to severe mental retardation

Autism and behavioral problems (hyperactivity)

Females with a full mutation (approximately 50%) have some degree of learning disability, generally mild

Family members with a premutation (carrier) may have premature ovarian failure or FXTAS (tremor ataxia)

Lifespan is normal

Inheritance

X-linked recessive

Carrier Frequency

1 in 260 females

Disease Incidence

1 in 4,000 males
1 in 8,000 females

High Risk Groups

All ethnicities

Individuals with a family history of mental retardation, autism, premature ovarian failure, or tremor ataxia

Testing

Genetic testing detects 99% of carriers

**Carrier screening is recommended by the American College of Obstetrics and Gynecology or the American College of Medical Genetics.*



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If you are interested in learning more about carrier testing for these disorders please indicate your choice below. Please detach after signing for review by a physician or genetic counselor:

Hemoglobinopathies

- YES**, I would like to have more information on screening.
- NO**, I am not interested in screening or in receiving more information.

Cystic Fibrosis

- YES**, I would like to have more information on screening.
- NO**, I am not interested in screening or in receiving more information.

Spinal Muscular Atrophy

- YES**, I would like to have more information on screening.
- NO**, I am not interested in screening or in receiving more information.

Fragile X Syndrome

- YES**, I would like to have more information on screening.
- NO**, I am not interested in screening or in receiving more information.

Signature _____ Date _____

Reviewed by _____ Date _____

Name: _____

Address: _____

City/State/Zip: _____

Phone: _____

E-mail: _____

