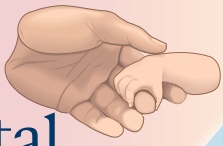
Desert Perinatal ASSOCIATES



A helpful guide to Carrier Testing

for Common Genetic Diseases

Hemoglobinopathies
Cystic Fibrosis
Spinal Muscular Atrophy
Fragile X

Desert Perinatal ASSOCIATES

THREE LOCATIONS:

5761 S. Fort Apache Road
Las Vegas, NV 89148
(Corner of Russell Rd & Fort Apache)

10105 Banburry Cross Dr., Suite 430
Las Vegas, NV 89144
(Next to Summerlin Hospital)

3001 W. Horizon Ridge Pkwy.
Henderson, NV 89052
(Next to St. Rose Siena Hospital)

TELEPHONE **(702) 341-6610**
FAX **(702) 341-6961**
www.DesertPerinatalAssociates.com

Please keep this brochure as a reference.

Introduction

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.

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 Chorionic Villus Sampling (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.

How is testing done?

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

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.

(Please keep this portion of the brochure as a reference)

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*

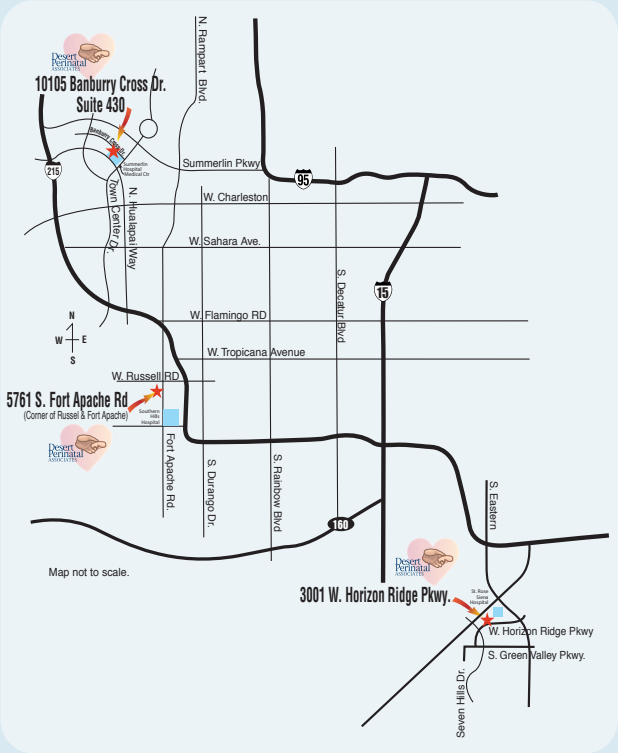
Clinical Features	Leads to a build-up of thick mucus in the lungs and digestive system; causes lung infections, digestive problems, and poor growth Some milder forms may be associated with pancreatitis and male infertility Intelligence in normal Life span is shortened; current life expectancy is 37 years
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 detects 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 detects 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) Life span 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.*



Desert Perinatal ASSOCIATES

THREE LOCATIONS:

5761 S. Fort Apache Road
Las Vegas, NV 89148
(Corner of Russell Rd & Fort Apache)

10105 Banburry Cross Dr., Suite 430
Las Vegas, NV 89144
(Next to Summerlin Hospital)

3001 W. Horizon Ridge Pkwy.
Henderson, NV 89052
(Next to St. Rose Siena Hospital)

TELEPHONE **(702) 341-6610**
FAX **(702) 341-6961**
www.DesertPerinatalAssociates.com

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: _____

Desert Perinatal ASSOCIATES


Desert
Perinatal
ASSOCIATES