



Houston IVF

Leaders in Fertility Research and Care

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PRECONCEPTION GENETIC SCREENING INFORMATION

What is genetic testing? Genetic testing is the examination of an individual's chromosomes, genes, proteins and/or other analytes for alterations associated with genetic disorders or conditions. The specific type of genetic testing recommended is based on your family history, self-reported ancestry, and/or your personal medical history.

What is genetic testing used for? Genetic testing in this context is to determine whether you are at increased risk for having a child with a particular genetic disorder (carrier testing).

What are the limitations or risks regarding genetic testing? The genetic testing will not identify all genetic abnormalities. It is specific for only the particular disorder that is being tested. Additionally, as with all testing, it does not provide 100% accuracy. The physical risks associated with genetic testing are typically small, especially as the tests we order typically require only a blood sample or a cheek swab. In some cases, genetic tests can also reveal previously unknown information about family relationships such as non-paternity or undisclosed adoption. Some patients have concerns about financial consequences of genetic testing such as the potential for discrimination or loss of employment or insurance. In May of 2008, the Genetic Information Nondiscrimination Act was signed into federal law. This offers protection against genetic discrimination in health insurance and employment.

What if I have further questions? The following genetic counselors are available to provide you with more information regarding appropriate genetic screening: Sandra Darilek with Baylor College of Medicine 713-799-1930 or Jennifer Hoscovek with University of Texas 713-500-6383 or 832-325-7133 option 2.

Which screening tests are recommended for me?

- Cystic Fibrosis** - Cystic fibrosis is a disorder characterized by pulmonary and gastrointestinal manifestations of varying severity. Although there is a wide spectrum of clinical expression most individuals who have CF experience substantial morbidity and reduced life expectancy. The carrier frequency is 1:29 among Caucasians, 1:46 among Hispanics, 1:62 among African-Americans, and 1:90 among Asian-Americans.
- Spinal Muscular Atrophy**- This is a group of inherited diseases that cause progressive muscle degeneration and weakness, eventually leading to death. One in fifty people carry this gene.
- Fragile X Syndrome**- Fragile X syndrome is a genetic condition involving changes in part of the X chromosome. It is the most common form of inherited intellectual disability (mental retardation) in boys. Screening is recommended to those with a family history of: fragile X, undiagnosed mental retardation, developmental delay or autism. Screening is also recommended for women that have been diagnosed with ovarian insufficiency, and some groups recommend universal screening.
- Ashkenazi Jewish Panel**-this is a panel including multiple disorders such as Canavan disease, Gaucher disease, Tay-sachs, Fanconi anemia, etc. These disorders are most prevalent in individuals of Ashkenazi Jewish descent.
- Hemoglobinopathy**- this is a broad group of disorders involving abnormalities of hemoglobin, the oxygen-carrying protein of the red blood cells. This testing is recommended for individuals of African American, Asian, Mediterranean, Middle Eastern and/ or Hispanic descent.
- Sickle Cell Disease**- Sickle cell disease refers to a group of disorders involving abnormal hemoglobin. It occurs most commonly in people of African origin. Approximately 1:12 African Americans has sickle cell trait. This screening includes a hemoglobin electrophoresis.

- ❑ **Karyotype**- A karyotype is the characterization of chromosomes based on size, shape and number. This is typically recommended in couples that have experienced multiple miscarriages, repeated IVF failures, or a woman diagnosed with premature ovarian failure. It is also recommended in men with significantly diminished sperm counts or azo-spermia (no sperm in the ejaculate).
- ❑ **Y Chromosome Microdeletion**- A Y- chromosome microdeletion screen is a test a test to determine if there is a portion of the Y chromosome that is missing. The incidence of Y chromosome microdeletions among men with significant sperm abnormalities is between 4-18%. A male with a Y-chromosome microdeletion is typically asymptomatic, although will often have sperm abnormalities. If a male with a Y- chromosome microdeletion conceives a son, his son will likely also demonstrate sperm abnormalities.

Optional Testing

- ❑ **Counsyl Testing**- Based in California, Counsyl is a company that offers genetic testing for over 100 of the most common autosomal recessive genetic abnormalities.

Carrier Screening Overview

Ethnicity	CF	Fragile X	SMA	Sickle Cell	Alpha thal	Beta thal	Ashkenazi Panel	Tay-Sachs
Caucasian	√	√*	√*					
African American	√	√*	√*	√		√		
Ashkenazi Jewish	√	√*	√*				√	
Asian	√	√*	√*		√	√		
Mediterranean	√	√*	√*	√	√	√		
Hispanic	√	√*	√*	√		√		
French Canadian/Cajun	√	√*	√*					√
Native American	√	√*	√*	√				

√* **Universal screening recommended by the American College of Medical Genetics; but recommended “per indication” by the American College of OBGYN**

References

American College of Obstetricians and Gynecologists Practice Bulletin Hemoglobinopathies in Pregnancy Number 78, January 2007

Joint SOGC-CCMG Clinical Practice Guideline for Carrier Screening for Thalassemia and Hemoglobinopathies in Canada No. 218, October 2008.

American College of Obstetricians and Gynecologists Preconception and Prenatal Carrier Screening for Cystic Fibrosis: Clinical and Laboratory Guidelines October 2001

American College of Medical Genetics and Genomics <http://www.acmg.net> accessed 2012



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Informed Consent/Refusal for Genetic Carrier Screening

PATIENT NAME

DATE OF BIRTH

ETHNICITY

- Cystic Fibrosis, Spinal Muscular Atrophy (SMA), Fragile X--** Recommended for all patients

The following additional tests have been recommended by my physician based on the ethnic information I provided:

- Ashkenazi Jewish Panel (includes Tay-Sachs, Canavans, Gauchers, etc.)**
- CBC (Complete Blood Count) and Hemoglobin electrophoresis. This will also test for Sickle Cell, Alpha Thalassemia and Beta Thalassemia.**
- Tay-Sachs only**

The purpose of my DNA testing is to determine whether I have any mutations known to be associated with genetic diseases listed above. Because genetic mutations vary among differing populations, I understand that the laboratory needs accurate information about my family history and ethnic background for the most accurate interpretation of the test results. When DNA testing demonstrates a genetic mutation, the person is either a carrier or is affected with the specific condition or disease. Consulting a doctor or genetic counselor is recommended to learn the full implication of the results.

When DNA testing does not demonstrate a known genetic mutation, the chance that the person is a carrier or is affected is reduced. There is still a risk to be a carrier or to be affected because the current testing cannot identify all the possible changes within a gene.

In some cases, DNA testing may discover other previously unknown information about family relationships, such as non-paternity (someone who is not the biological father) or adoption.

Additional considerations of consent/refuse applicable to the above testing:

The decision to consent or to refuse any suggested testing is entirely my own. No test(s) will be performed and reported on my sample other than the one(s) authorized by my doctor, and any unused portion of my original sample will be destroyed within 2 months of receipt of the sample by the laboratory. Test results will be disclosed ONLY to the doctor named below, or to his/her agent, unless otherwise authorized by the patient or required by law.

My signature below indicates that I have read, or had read to me, the above information and that I understand it. I have had the opportunity to discuss this information, including the purpose and possible risks, with my doctor or someone my doctor has designated. **I know that I may obtain professional genetic counseling, if I wish, before signing this consent. I have all the information that I desire, and all my questions have been answered.**

- Yes: I REQUEST** the following genetic screens be performed. I understand and accept the significance of this decision.

TEST(S) REQUESTED

- No: I DECLINE** to have the following genetic screening be performed. I understand and accept the significance of this decision.

TEST(S) REQUESTED

PATIENT SIGNATURE

DATE

WITNESS

Genetic Counseling: We understand that we may request a consultation with a genetic counselor so that we may fully review our family histories and genetic carrier screening recommendations in order to sufficiently understand the potential genetic risks to our offspring. Information regarding genetic counseling was given to me. My signature below indicated that I either accept or waive genetic counseling.

- YES: I REQUEST** genetic counseling. I understand and accept the consequences of this decision.

- NO: I DECLINE** genetic counseling. I understand and accept the consequences of this decision.

PATIENT SIGNATURE

DATE

WITNESS