Reproductive Facts

Patient fact sheet developed by the American Society for Reproductive Medicine

Genetic Screening for Birth Defects

Birth defects, which occur in nearly one in 20 pregnancies, range in severity from minor abnormalities to extensive genetic disorders or mental retardation. Some people have a greater than average risk of having a child with a birth defect.

Genetic screening refers to the use of specific tests to determine which members of a population are at increased risk for an inherited condition.

Genetic testing is the use of specific tests to learn more about the genetic status of an individual who is suspected to be at increased risk for an inherited disease.

These terms are frequently used interchangeably. Genetic screening before pregnancy may help identify couples who have an increased risk of age-related or familial genetic disorders and birth defects. No single test, however, can accurately predict the risk of all defects in a child, and many birth defects, such as those related to environmental and toxic exposures and those that are random and unexplained, are not genetically based and may not be detected with genetic screening.

Screening for genetic diseases that may affect offspring depends upon:

- the racial or ethnic background of the couple,
- their family and medical history,
- and associated conditions.

Various racial and ethnic groups demonstrate an increased prevalence of specific diseases, and couples of these backgrounds may have their carrier status screened accordingly, as described below. Most couples have their carrier status checked if there is a family history of the disorder or if they belong to an at-risk racial or ethnic group.

In addition to screening for carrier status for genetic conditions, pregnant people may also undergo prenatal testing to determine if their child is affected by a serious chromosomal abnormality, such as Down syndrome, or an abnormality of the spinal cord such as spinal bifida. Any woman may elect to have her risk for these disorders estimated by noninvasive testing, such as measurement of alpha-fetoprotein in the mother's blood as a marker for risk of spina bifida and other neural tube defects. If the screening test identifies that she is at increased risk, specific diagnostic tests may be performed, such as ultrasound examinations and amniocentesis for spinal bifida.

"One of the most common reasons to have genetic screening performed is age..."

One of the most common reasons to have genetic screening performed is age, since the risk of having a child with a chromosomal abnormality increases with age. Additionally, if both parents are silent carriers for a genetic condition, prenatal testing can determine if the unborn child is affected with the disease.

Preimplantation genetic testing for monogentic defects (PGT-M) is a technique used in conjunction with in vitro fertilization (IVF) to test embryos for specific genetic disorders prior to transfer to the uterus. PGT-M makes it possible for couples or individuals who have or who carry serious inherited disorders to decrease the risk of passing the disorder on to their child.

Indications for Genetic Screening- Advanced Maternal Age

Age may increase the risk of chromosomal problems and miscarriage. Consider discussing this risk with your physician or a genetic counselor before pregnancy and your risk reducing treatment options.

Non-invasive prenatal testing (NIPT) is a blood test that may be performed during the end of the first trimester or early second trimester.

A normal screening test means that the risk of a chromosomal abnormality is reduced, but not zero. Chorionic villus sampling and amniocentesis are two methods of confirming the diagnosis of a chromosomal abnormality when the screening test is positive.

Both of of these tests carry a small risk of miscarriage. Many parents want to know this information so they can make informed decisions about their pregnancy.

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Racial or Ethnic Associations to Specific Diseases

- **Sickle Cell Disease:**

Anyone with African American ancestry should be offered screening with a hemoglobin electrophoresis (a simple blood test) to determine carrier status of this disease, as one in 10 may be a carrier.

- **Cystic fibrosis (CF):**

It is estimated that 3% to 10% of Caucasians carry a defective CF gene but do not have symptoms because a person must inherit two defective CF genes, one from each parent, to develop the disease. CF is the most common, serious, inherited disease in Caucasians, and is more common in those of northern or central European background and Ashkenazi Jewish background.

- **Thalassemia:**

People of Greek, Italian, Mediterranean, or southern Asian descent experience a high incidence of this disease. Patients can have a complete blood count (CBC) with mean corpuscular volume (MCV) to rule out the possibility of thalassemia. An MCV of <80 should be evaluated further by hemoglobin electrophoresis. About 3% of the world's population carries a gene for thalassemia.

- **Tay Sachs:**

This disease has a high incidence in Eastern European Jews and French Canadians.

- **Familial Associations with Specific Diseases**

A family history of any of the following disorders should prompt genetic counseling, and when relevant, screening or testing for associated gene abnormalities or carrier status:

- Down syndrome
- Muscular Dystrophy
- Tay Sachs
- Chromosomal syndromes
- Neurofibromatosis
- Sickle Cell
- Seizures
- Unexplained stillbirths or neonatal deaths
- Recurrent miscarriage
- Huntington's disease
- Cystic Fibrosis
- Hemophilia or other bleeding disorders
- Mental handicap
- Neural tube defects
- Familial dysautonomia

Medical History/Conditions Associated with Genetic Conditions

• **Recurrent miscarriages** may be due to chromosomal or genetic abnormalities. Women who experience two or more consecutive miscarriages are frequently screened to see if they carry abnormal chromosomes that predispose to miscarriage.

• **Congenital bilateral absence of vas deferens** (absence of the two muscular tubes that carry sperm from the epididymis to the urethra, CBAVD) is associated with CF. The majority of men with CBAVD have CF. For couples where the man has CBAVD, testing for CF carrier status of the female partner should be offered.

• **Azoospermia** (absence of sperm in semen) and severe oligospermia (very low sperm counts) have been associated with chromosomal and genetic abnormalities. Blood tests, including a karyotype (which looks at the number and structure of the chromosomes) and Y chromosome micro-deletion analysis (which looks for missing pieces of the Y chromosome), may be helpful in selected cases. If abnormalities are identified, genetic counseling is indicated.

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