



AMERICAN SOCIETY FOR REPRODUCTIVE MEDICINE

Formerly The American Fertility Society

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PATIENT'S FACT SHEET

Genetic Screening for Birth Defects

Birth defects, which occur in nearly one in 20 pregnancies, range in severity from minor anatomic abnormalities to extensive genetic disorders or mental retardation. Some couples have a greater than average risk of having a child with a birth defect. Genetic screening may help identify couples at risk for certain genetic disorders but not all birth defects. 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. There is no single test that will detect the risk of any genetic disease in a couple's offspring. In addition, birth defects may occur that are not genetically based (e.g., environmental and toxic exposure, or random and unexplained) and may not be detected with genetic screening. Preimplantation genetic diagnosis (PGD) is a technique that can be used during in vitro fertilization (IVF) procedures to test embryos for genetic disorders prior to their transfer to the uterus. PGD makes it possible for couples or individuals with serious inherited disorders to decrease the risk of passing the disorder to their child. This technique is controversial and raises issues of sex selection and genetic engineering. At present, PGD is only offered in a few centers, usually under the supervision of an institutional ethics review board, but its use may be more widespread in the near future. For more information on PGD, refer to the ASRM Fact Sheet titled *Preimplantation Genetic Diagnosis*.

INDICATIONS FOR GENETIC SCREENING

ADVANCED MATERNAL AGE

Women over 35 have a higher risk of chromosomal problems and miscarriage. Prior to attempting pregnancy, women in this age group may wish to talk with their physician or a genetic counselor about their chances of having a child with a chromosomal problem, such as Down's syndrome, and the choices for prenatal genetic testing if pregnancy is achieved. Chorionic villus sampling and amniocentesis are two methods of prenatal testing. Many parents want to know this information so they can make informed decisions about the pregnancy.

RACIAL OR ETHNIC ASSOCIATIONS TO SPECIFIC DISEASES

- **Sickle Cell Disease:** Anyone with African-American ancestry should be screened via hemoglobin electrophoresis for carrier status of this disease, as one in 10 may be a carrier.
- **Thalassemia:** People of Mediterranean or Asian descent experience a high incidence of this disease. It is recommended that patients have a CBC with MCV to rule out the possibility of Thalassemia status. An MCV of <80 should be further evaluated by hemoglobin electrophoresis. About 3% of the world's population carry the β Thalassemia gene.
- **Tay Sachs:** This disease has a high incidence in Eastern European Jews and French Canadians.

FAMILIAL ASSOCIATIONS TO SPECIFIC DISEASES

A family history of any of the following genetic disorders should prompt genetic counseling and/or possible screening for carrier status:

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| • Down's syndrome | • Muscular Dystrophy | • Tay Sachs |
| • Other chromosomal abnormalities | • Neurofibromatosis | • Sickle Cell |
| • Unexplained stillbirths or neonatal deaths | • Cystic Fibrosis | • Seizures |
| • Huntington's disease | • Mental retardation | • Maternal exposure |
| • Hemophilia or other bleeding disorders | • Neural tube defects | • Multiple miscarriages |

MEDICAL HISTORY/CONDITIONS ASSOCIATED WITH GENETIC CONDITIONS

- About 7% of stillbirths and neonatal deaths have chromosomal abnormalities, compared with 0.5% of all newborns.
- Recurrent miscarriages may be due to genetic abnormalities.
- Congenital absence of vas deferens (absence of the two muscular tubes that carry sperm from the epididymis to the urethra) is associated with Cystic Fibrosis. Carrier status should be determined.
- Azoospermia/Oligospermia (absence of sperm in semen) has been associated with sex chromosome abnormalities and deletions within the Y chromosome. A karyotype may be helpful in selected cases.