Genetic Counseling

Genetic counseling helps people look at the risks of genetic problems in pregnancy. Genetic problems are problems that may be passed from the mother and father to their child.
Genetic counseling is done:

- To see if a couple is at risk of passing on a problem.
- To explain the cause of a disorder if present.
- To understand the odds passing it on to the baby.
- To determine what can be done medically.
- To use in family planning to prevent or avoid a problem.

Genetic counseling can be done by:

- A geneticist.
- A physician with special training in genetics.
- Genetic counselors.

The following are reasons to seek a referral for genetic counseling and/or genetic evaluation with a genetic physician:

FAMILY HISTORY FACTORS

- Mental retardation.
- Neural tube defects (such as *spina bifida*).
- Chromosome abnormalities (such as Down syndrome, Fragile X syndrome).
- Cleft lip/cleft palate.
- Heart defects.
- Short stature.
- Single gene defects (such as cystic fibrosis or PKU).
- Hearing or visual impairments.
- Learning disabilities.
- Psychiatric disorders.
- Cancers (breast, uterus, ovary and intestine).
- Multiple pregnancy losses (miscarriages, stillbirths, or infant deaths).
- Other disorders which could be considered genetic.
- Either parent with a dominant disorder or any disorder seen in several generations including:
  - High blood pressure.
  - High cholesterol.
  - Muscular dystrophy.
  - Huntington's chorea.
  - Polycystic kidney.

BOTH PARENTS ARE CARRIERS FOR:

- Depression.
- Seizures (*convulsions*).
- Alcoholism.
- Diabetes.
- Thyroid disorder.
- Others in which birth defects may be associated either with the disease process or with common medications prescribed for the disease.
- Fetal or parental exposure to potentially harmful agents. These include:
  - Drugs.
  - Chemicals.
  - Infection.
  - Radiation.
- Infertility cases where either parent is suspected of having a chromosome abnormality.

- Couples requiring assisted reproductive techniques to achieve a pregnancy, or individuals donating eggs or sperm for those purposes.

OTHER FACTORS

- Persons in specific ethnic groups or geographic areas with a higher incidence of certain disorders. These include Tay Sachs disease, sickle cell disease, or thalassemia.
- Extreme parental concern or fear of having a child with a birth defect.
- Parents are blood relatives (*consanguinity*) or incest in a pregnancy is involved.
- Premarital or preconception counseling in couples at high risk for genetic disorders based on family or personal medical history.
- The birth of an affected child or by carrier screening.
- Mother, known, or presumed carrier of an X-linked disorder such as hemophilia.
- Either parent is a known carrier of a balanced chromosome abnormality.
- A previous baby with a genetic disorder.
- After the baby is born, you find out the baby has a genetic disorder.

PREGNANCY FACTORS

- The mother is over 35 years old a the time of delivery.
- Maternal serum screening indicating an increased risk for neural tube defects, Down Syndrome, or trisomy 18.
- Abnormal prenatal diagnostic test results or abnormal prenatal ultrasound examination.
- Maternal factors such as:
  - Schizophrenia.
  - Depression.
  - Seizures.
  - Alcoholism.
  - Diabetes.
  - Thyroid disorder.
  - Other factors in which birth defects may be associated either with the disease process or with common medications prescribed for the disease.
- Fetal or parental exposure to potentially harmful agents such as:
  - Drugs.
  - Chemicals.
  - Radiation.
  - Infection.
- The father is over the age of 55 years at the time of conception.
- Infertility cases where either parent is suspected of having a chromosome abnormality.
- Couples requiring assisted reproductive techniques to achieve a pregnancy, or individuals donating eggs or sperm for those purposes.
- Recurrent pregnancy loss.
- Stillborn baby.

FINDING A GENETIC COUNSELOR

Most caregivers know and have a list of genetic counselors. Your caregiver can refer you to one of them. You can also contact the National Society of Genetic Counselors for their locations and for more information.

AFTER RECEIVING GENETIC COUNSELING

Genetic counselors can help you understand what options you have. They can help you understand what to do next. They can share the experiences of other couples who had babies with genetic problems. If you are at high risk for having a baby with a severe or fatal genetic disorder, you have several options:

- Have in vitro fertilization with healthy eggs that were fertilized in vitro.
- Use donor sperm or eggs.
- Adopt a baby.

Genetic counselors can also help if you find out when you are pregnant that the baby has a severe or fatal

genetic disorder. Some options are:

- Help you prepare for a particular disorder by speaking to:
  - Specialists in caring for Down syndrome.
  - Surgeon for heart defects.
  - Social workers.
  - Support groups.
- Fetal surgery is sometimes possible and helpful to treat some genetic defects.
- Ending the pregnancy.

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