# *Early Detection through Amniocentesis*

The Center for Human Genetics can test for certain problems in the fetus caused by chromosomal or other genetic errors. This is usually done between the 14th and 16th week of pregnancy through a procedure called genetic amniocentesis. In amniocentesis, a thin needle is inserted into the uterus and a small amount of amniotic fluid is withdrawn for analysis.

By studying the chromosomes from the fetus, the presence or absence of chromosomal conditions such as Down syndrome can be detected. Additional testing can be performed to detect open spine defects (spina bifida), certain single gene defects, or other specific metabolic abnormalities. Ultrasound evaluation may be useful in detecting large structural abnormalities of the skeleton or internal organs.

**Not** every pregnant woman is an appropriate candidate for amniocentesis. The procedure should be considered in women who:

- Are 35 years of age or older at delivery.
- Have concerns about the results of a prenatal screening test such as maternal serum alpha-fetoprotein (AFP).
- Have a child with a chromosome abnormality.
- Are known carriers of an X-linked condition such as hemophilia or muscular dystrophy.
- Have a family history of a neural tube defect such as spina bifida or anencephaly.
- Are known to be at risk for a genetic abnormality for which there is a prenatal test.

# *Making Appointments*

Appointments for genetic counseling and other services at the Center for Human Genetics (4420 Dewey Avenue) may be made by physician referral or self referral. For more information, contact:


Hattie B. Munroe Center for Human Genetics
Meyer Rehabilitation Institute
University of Nebraska Medical Center
600 South 42nd Street
Omaha, NE 68198-5440
(402) 559-5070


*"She has her father's eyes . . . her mother's smile."*

# Genetic Services at U N M C

## *Just as we*

can inherit a parent's hair color and other readily identifiable family characteristics, certain physical and emotional disorders can be passed from generation to generation. Such inherited conditions, reproductive risks and related matters are the concern of the Hattie B. Munroe Center for Human Genetics of the Meyer Rehabilitation Institute, a unit of the University of Nebraska Medical Center. Here individuals and families may receive a variety of diagnostic services and genetic counseling.

## *Physical Examination*

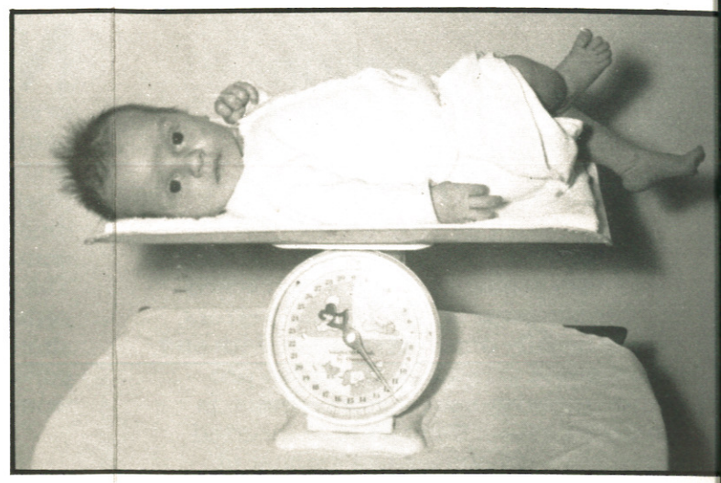
Evaluation of individuals at risk for genetic conditions is provided by physician referral. In a typical session, medical family histories are reviewed, a physical examination may be conducted and, on occasion, further testing may be recommended to gather additional information. In conjunction with other medical specialties, medical evaluation and/or laboratory testing are performed for diagnostic purposes.

## *Genetic Counseling*

Genetic counseling deals with issues surrounding the occurrence or the chance of occurrence of a birth defect or an inherited condition in a family. Through this process, individuals and families are helped to understand the diagnosis, cause, treatment and management of a given disorder. Alternatives for dealing with the chance of recurrence in future children may be discussed. The intent is to assist both the family and the primary physician in making informed choices about medical management and family life. Common reasons for genetic counseling include family members with:

- Birth defects (e.g., cleft lip and palate, spina bifida)
- Mental retardation
- Known or suspected chromosome abnormality (e.g., Down syndrome)
- Known or suspected genetic disorder (e.g., cystic fibrosis, hemophilia)
- Metabolic disorder (e.g., PKU)
- Delayed onset of puberty
- Sensory defect (e.g., blindness, deafness)
- Progressive neurologic disorder (e.g., Huntington's)
- Neuromuscular disorder (e.g., muscular dystrophy)
- Affective disorder (e.g., schizophrenia, manic depression)
- Congenital heart disease
- Familial cancer
- Diabetes

DADDY WATCHING US AND OUR CATS



Genetic counseling is also appropriate in cases of:

- Consanguinity (e.g., close relative marriage)
- Miscarriage, stillbirth or infertility
- Environmental exposure (e.g., drugs, alcohol, smoking, X-rays, maternal infection)
- An ethnic background known to have a high incidence of specific genetic conditions. For example, Blacks are at risk for sickle cell anemia and Jews for Tay-Sachs disease.