Pediatric Genetics Conditions Evaluated and Treated

Medical Genetics is committed to providing you, your newborn, children, or adolescents with excellent care in diagnosis, management and treatment of all forms of hereditary disorders and birth defects. Our staff includes many specialists from clinical geneticists, counselors to neurologists that work as a team to develop your child’s treatment plan.

**Pediatric Genetic Problems Evaluated and Treated:**

- Metabolic Clinic
- Lysosomal storage disorders clinic
- Fetal Alcohol Syndrome
- Newborn Screening Counseling & Education
- Pediatric General Evaluation • Disorders of growth
- Neurocognitive impairment including autism
- Birth defects
- Distinctive facial features
- Hearing impairment (in conjunction with Otolaryngology)

**Specific Expertise in:**

- Neurofibromatosis (NF-1 & NF-2)
- Prader-Willi Syndrome
- Marfan Syndrome
- Down Syndrome
- Chromosome 22q11
- Deletion Syndrome
- Turner Syndrome
- Chromosome Abnormalities

**Types of Genetic Changes**

The human body has 20,000 to 25,000 different genes. Genes are located on chromosomes, which are stick-shaped structures in the middle of each cell in the body. Each cell usually has 46 chromosomes grouped in 23 pairs. Each gene has a specific function. And when a gene or chromosome is abnormal, it may cause health problems in the body.

**There are 2 main types of genetic changes:**

Information gathered from [http://healthcare.utah.edu/pediatrics/services/genetics.php#tabs-accordion1](http://healthcare.utah.edu/pediatrics/services/genetics.php#tabs-accordion1). Last updated 06/13/16.
• Chromosome abnormalities
• Single-gene defects

What are chromosomal abnormalities?

Chromosomal abnormalities in the baby may be inherited from the parent or may occur with no family history. These are the most common:

Aneuploidy: More or fewer chromosomes than the normal number. Examples include:
• Down syndrome (trisomy 21): Cells contain 3 copies of the 21st chromosome.
• Turner syndrome: One of the two sex chromosomes is not transferred, leaving a single X chromosome for 45 total chromosomes instead of 46.

Deletion: This is when part of a chromosome is missing, or part of the DNA code is missing.

Inversion: This is when a chromosome breaks and the piece of it turns around and reattaches itself. Inversions can be passed down in families, but they may or may not cause birth defects.

Ring: A ring chromosome is one where the ends are attached to itself to form a ring. Rings can be passed down in families. They may or may not cause health problems.

Translocation: This is when a chromosome segment rearranges from one location to another. It can happen either within the same chromosome or move to another chromosome. There are two types:
• Balanced translocation: This is when the DNA is equally exchanged between chromosomes. No DNA is lost or added. A parent with a balanced translocation is healthy, but he or she may be at risk for passing on unbalanced chromosomes to a child.
• Robertsonian translocation: This is a balanced translocation in which one chromosome joins the end of another.

Mosaicism: This is when a person has 2 or more sets of chromosomes in his or her cells with different genetic material.

What are single-gene disorders?

A change in a single gene causes a defect or abnormality. Single-gene changes usually have a higher risk of being passed on to children. Single gene changes can be:

• Dominant: This means the abnormality occurs when only one of the genes from one parent is abnormal. If the parent has the disorder, the baby has a 1 in 2 chance of inheriting it. Examples include:
  o Achondroplasia: This is a bone development disorder that causes dwarfism.
  o Marfan syndrome: This is a connective tissue disorder that causes long limbs and heart defects.
• Recessive: This means the abnormality only occurs when both parents have abnormal genes. If both parents are carriers, a baby has a 1 in 4 chance of having the disorder. Examples include:

- **Cystic fibrosis**: This is a disorder of glands that causes excess mucus in the lungs. It also causes problems with how the pancreas works and with how food is absorbed.
- **Sickle cell disease**: This condition causes abnormal red blood cells that don’t carry oxygen normally.
- **Tay-Sachs disease**: This is an inherited autosomal recessive condition that causes the central nervous system to decline. The condition is fatal, usually by age 5.

**X-linked**: The disorder is determined by genes on the X chromosome. Males are mainly affected and have the disorder. Daughters of men with the disorder are carriers of the trait and have a one in two chance of passing it to their children. Sons of women who are carriers each have a one in two chance of having the disorder. Examples include:

- **Duchenne muscular dystrophy**: This is a disease that causes muscle wasting.
- **Hemophilia**: This is a bleeding disorder caused by low levels or lack of a blood protein that is needed for clotting.

**How genetic changes are passed along in a family**

The way a gene is inherited can help determine the risk of it in a current or future pregnancy. The risk of having a baby with a birth defect from a genetic change increases if:

- The parents have another child with a genetic disorder.
- There is a family history of a genetic disorder.
- One parent has a chromosome abnormality.
- The baby in the womb has abnormalities seen on an ultrasound.

Families at risk for genetic diseases may want to talk with a certified genetic counselor. Making a chart of members of the family and their health histories can help show risk for certain problems. Genetic counseling also helps parents understand the effects of a disorder and ways it may be prevented or treated.

**Getting genetic testing**

Each parent’s DNA may need to be checked. This is done to learn about some genetic inheritance patterns. Prenatal testing can also be done to check the baby in the womb for problems. Testing may include:

- **Ultrasound**: This test uses sound waves to look at how a baby in the womb is growing.
- **Chorionic villus sampling (CVS)**: This test uses a sample of tissues around the baby to look for problems.
- **Amniocentesis**: This test uses a sample of the amniotic fluid from the sac around the baby to check for problems.