

Special Health Matters

Genetic Counseling

♥ Who are genetic counselors? What do they provide?

Genetic counselors are health professionals with specialized graduate degrees and experience in the areas of medical genetics and counseling. Genetic counselors provide information and support to families who have members with birth defects or genetic disorders and to families who may be at risk for a variety of inherited conditions. They:

- **Identify families at risk**
- **Investigate the problem present in the family**
- **Interpret information about the disorder**
- **Analyze inheritance patterns and risks of recurrence**
- **Review available options with the family**
- **Provide supportive counseling to families**
- **Serve as patient advocates**
- **Refer individuals and families to community or state support services.**

There are many different subspecialties within genetic counseling, therefore, the specific genetic counselor that you refer to will depend upon the type patients that they see in their practice.

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|-------------------------|------------------------|
| ▪ Pediatric | ▪ Adult |
| ▪ Prenatal | ▪ Neuromuscular |
| ▪ Cancer | ▪ Research |
| ▪ Cardiovascular | ▪ Laboratory |

♥ When should you refer to a genetic counselor?

When you have a family or individual who is concerned about their risk of occurrence or recurrence of a specific condition. **Indications for referral:**

- Individuals who are suspected of having a genetic condition:

▪ Developmental delay/Mental retardation	▪ Dysmorphic facial features
▪ Growth delay	▪ Multiple congenital anomalies
▪ Seizures	
- Abnormal prenatal history:

▪ Ultrasound anomalies	▪ Teratogenic exposures
▪ Abnormal serum screening	
- Individuals with a family history of:
 - Birth defects - **Cleft lip/palate, congenital heart disease, limb malformations, holoprosencephaly**
 - Single gene disorders - **Cystic fibrosis, fragile X syndrome, muscular dystrophy, neurofibromatosis**
 - Chromosome abnormalities - **Down syndrome, Williams syndrome, translocations**
 - Adult-onset conditions - **Huntington disease, Early-onset Alzheimer's disease, ataxia, dementias**
 - Cancer - **Hereditary breast/ovarian cancer; colon cancer, melanoma, pancreatic**
 - Common complex conditions - **Coronary artery disease, diabetes, autoimmune disorders**
- Carrier testing:
 - Ethnic carrier screening - **Tay Sachs, CF, Sickle cell, Gaucher, thalassemias**
 - Spouse/partner carrier screening - **PKU**

♥ Resources

Genetic Counseling 101 http://www.medhomeportal.org/file.cfm?file_id=633& (source)

Pediatrics—**Division of Medical Genetics, University of Utah** 801-581-8943

Perinatal/preconception—**University of Utah** 801-581-7825 **LDS Hospital** 801-408-3897

High Risk Cancer Clinics—**Huntsman Cancer Institute** 801-585-3525

National Society of Genetic Counselors <http://www.nsgc.org>

