

Genetic Counseling Referrals

When should I refer to Genetic Counseling or Genomic Medicine?

The following information is intended to provide a brief overview of some common referral indications. We have outlined the most common areas within genomic medicine, but there may be other specialty clinics available near you. Some possibilities include: neurogenetics, ophthalmologic genetics, connective tissue disorders, Down syndrome clinics, and RASopathy clinics (for conditions including Noonan, neurofibromatosis, etc). We suggest getting to know your institution, along with any genomic medicine providers there and in the specialty centers near you to best meet the needs of your patients.

Quick Reference

More specific descriptions are given below, but a quick reference is the “Rule of Two/Too”

If you find yourself describing a patient or family’s history with the words “two/too”, this may indicate the need for a referral to Genomic Medicine

- “Too” examples include: too tall/too short, too many (e.g., cafe au lait macules), too young to have this condition, experience a symptom or condition too frequently
- Two or more congenital anomalies
- Two or more family members, or two or more generations, affected with the same disease/condition

General Genetics for Adults/Pediatrics

General referral indications can include:

- Assessing developmental delay/intellectual disability, autism spectrum disorder, epilepsy
- Confirming or ruling out a genetic condition; to discuss treatment and/or screening, testing options, and inheritance
- Assessing possible syndromic associations with an apparently isolated feature, such as hearing or vision loss
- Addressing any concerns or questions arising from direct-to-consumer genetic testing, such as 23andMe or AncestryDNA

Hereditary Cancer

General referral indications can include:

- Personal or family history of cancer with unusually young age of onset
- Diagnosis of a tumor with high likelihood of heritability (e.g., male breast cancer, ovarian, metastatic prostate, Wilms tumor, neuroendocrine tumor, medullary thyroid cancer, etc.)
- Personal or family history of bilateral cancers, or multiple primary tumors in one person

- Features associated with an inherited cancer syndrome (e.g., multiple colon polyps, or rare types of GI polyps, such as hamartomatous polyps)
- Family history of clustered, related cancers (e.g., breast/ovarian, colon/uterine)
- Family history of a known inherited cancer syndrome
- Germline genetic mutation revealed by a tumor profiling test

Preconception and Prenatal Genetics

General referral indications can include:

- Interest in genetic carrier screening for themselves and/or their partner
- Interest in discussing risks for chromosomal abnormalities, such as Down syndrome
- History of a previous child with a birth defect, developmental delay, or other genetic condition
- History of multiple unexplained miscarriages or cases of unexplained infant deaths
- Consanguineous union (cousins or otherwise blood related)
- Maternal age of 35-years-old or older at time of delivery for a single pregnancy; 33-years-old or older at time of delivery for twin pregnancy
- Current pregnancy with anomalies identified by ultrasound
- Current pregnancy with an abnormal genetic screening test
- Current pregnancy with risk of or concern for maternal exposures, such as medications, radiation, drugs/alcohol, or infections

Cardiovascular Genetics

General referral indications can include:

- Personal or family history of cardiac problems, such as cardiomyopathy, arrhythmia, congenital heart defects
- Personal or family history of vascular problems, such as aortic aneurysms/dissections, other arterial aneurysms/dissections, and connective tissue disorders suggestive of Marfan and related syndromes
- Personal or family history of familial hypercholesterolemia
- Family history of cardiac arrest or sudden death

About Genetic Counselors

Genetic counselors are professionals who have specialized education in genetics and counseling to provide personalized help patients may need as they make decisions about their genetic health. Today, there are more than 4,000 certified genetic counselors.

Your patients can find additional resources on genetic diseases at www.aboutgeneticcounselors.com.

To find a Genetic Counselor who can assist you and your patients, please visit the National Society of Genetic Counselors at www.nsgc.org.