When should you consider referring a patient to a genetic counselor?

**General Adult Genetics or Pediatric Genetics**
- A chromosomal disorder: Down syndrome, Edwards syndrome
- A monogenic disorder: Fragile X syndrome, Rett syndrome
- Visual loss or a congenital eye defect: retinitis pigmentosa, microphthalmia, early-onset macular degeneration, cataracts
- Significant hearing impairment not secondary to recurrent otitis media
- A progressive neurologic condition: peripheral neuropathy, unexplained myopathy, progressive ataxia, early onset dementia, facial movement disorder
- A progressive muscle weakness: muscular dystrophy, spinal muscular atrophy, myotonic dystrophy
- Skeletal dysplasia or dwarf stature: neurosensory imperfections, achondroplasia
- An unexplained intellectual disability, global developmental disability or autism spectrum disorder
- An inherited bleeding disorder: hemophilia, von Willebrand disease
- An abnormal newborn screening test result
- One or more birth defects: heart defect, cleft lip & palate etc.

**Hereditary Cancers**
- A cancer known to be associated with specific genes or mutations: breast, ovarian, colorectal
- A positive germline mutation revealed by family genetic testing or tumor profiling testing
- A compelling family history of cancer: young age at onset, bilateral lesions, familial clustering of related tumors

**Preconception Genetics or Prenatal Genetics**
- An abnormal prenatal ultrasound examination: neural tube defects, down syndrome, trisomy 18
- Fetal anemia following exposure to medications, or defect(s) in fetal organs, agents: drugs, chemicals, radiation, infection
- A positive carrier screening test result: cystic fibrosis, thalassemia, sickle cell anemia, Tay-Sachs etc.
- Mother is a known/presumed carrier of an X-linked disorder: Duchenne Muscular Dystrophy (DMD), Hemophilia
- Recurrent pregnancy loss (2 or more) or multiple IUFDs (Intrauterine Fetal Decease)
- Infertility where either parent is suspected of having a chromosome abnormality or other genetic factors

**Cardiovascular Genetics**
- A cardiovascular disorder: cardiomyopathy, long QT, congenital heart defect
- A vascular disorder including arterial aneurysms/dissections and connective tissue disorders: Marfan & Ehlers Danlos syndrome
- A high level of lipids (fats, cholesterol, and triglycerides) circulating in the blood: hyperlipidemias, familial hypercholesterolemia

*This list indicates only some of the common indications for referral to a genetic counselor. It is not an exhaustive list.

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Genetics disorders in the United Arab Emirates (UAE)

The UAE has a high incidence of genetic disorders—approximately 360 are reported to be present according to the Center of Arab Genomic Studies database. The most common are hemoglobinopathies such as β-thalassemia, α-thalassemia and sickle cell disease. The abnormal gene frequency for β-thalassemia (including βS for sickle cell) is as high as 8.5%, and the positive screen rate for α-thalassemia among Emiratis is approximately 50%. Other common genetic disorders in the country include: α-thalassemia; cystic fibrosis; congenital abnormalities; hereditary cancer syndromes; chromosomal syndromes and cytis disorders.

Approximately 65% of all genetic disorders in the UAE are autosomal recessive, meaning a child has to inherit one copy of a defective recessive gene from each parent for the development of the disorder. Contributing factors include: consanguinity (up to ~ 50%); the founder effect: a group of patients in a certain geographical area having a common ancestor; selective environmental elements that give a survival advantage to carriers; gene pool homogeneity; and the founder effect: a group of patients in a certain geographical area having a common ancestor.

Given the high incidence of genetic disorders in the UAE, it is important that patients and their families have access to a licensed certified genetic counselor.

The benefit of working collaboratively with a licensed genetic counselor

Genetic counselors are healthcare providers, who are skilled in communicating complex genetics-related information to patients in an easy-to-understand and digestible manner. They help patients adapt to the medical, psychological and familial implications of their genetic disorders, and facilitate informed decisions in a personalized manner. They ensure each patient receives: useful and understandable information regarding the cause, symptoms, inheritance, recurrence risk and complexities of genetic disorders relevant to them and their family; advice on available genetic testing options which may be relevant to their circumstances; help in understanding the results of genetic testing undertaken; support in planning their next steps (for example, reproductive preservation options); and referrals to other medical specialties, advocacy and support groups to help them effectively deal with their diagnosis.

The Unified Healthcare Professional Qualification Requirements9—issued by the UAE Ministry of Health and Prevention, the Department of Health Abu Dhabi, and the Dubai Health Authority—stipulate that in order to obtain a license to practice in the UAE, genetic counselors must hold a master’s degree in genetic counseling.

About genetic counselors & how they support patients

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