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

You are encouraged to refer your patient to a genetic counselor if they:

1. inform you of a family history of one of the below indications*
2. have been diagnosed with one of the below indications
3. are suspected to have one of the below indications
4. present an abnormal genetic test result which they need support in understanding
5. need support in making a decision regarding reproductive prevention options
6. are seeking premarital or preconception counseling

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, familial movement disorder
- A progressive muscle weakness: muscular dystrophy, spinal muscular atrophy, myotonic dystrophy
- Skeletal dysplasia or short stature: osteogenesis imperfect, achondroplasia
- An unexplained intellectual disability, global developmental disorder or autism
- Abnormal sexual maturation or delayed puberty
- An inherited bleeding disorder: hemophilia, thrombophilia
- An immune deficiency: Severe Combined Immunodeficiency (SCID)
- A kidney disorder: polycystic kidney disease
- A child with a metabolic disorder
- An abnormal newborn screening test result
- One or more birth defects: heart defect, cleft lip & palate etc.

Preconception Genetics or Prenatal Genetics

- An abnormal prenatal diagnostic test result, abnormal NIPT or abnormal prenatal ultrasound examination: neural tube defects, down syndrome, trisomy 18
- Fetal or parental exposure to potentially teratogenic, mutagenic, or carcinogenic 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 Demise)
- Infertility where either parent is suspected of having a chromosome abnormality or other genetic factors

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

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) or lipoproteins 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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Document Code: NRL-MKT-CLT-013

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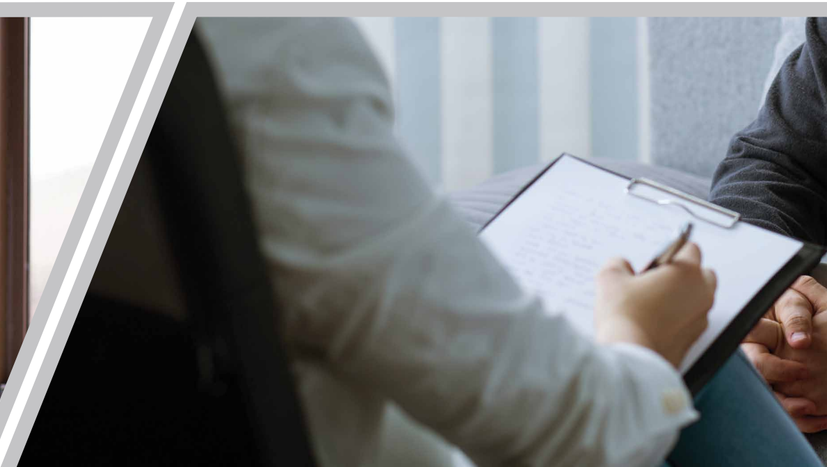
Genetic Counseling Services

Physician Guide



Evolution of the field of medical genetics

Genetic disorders are found in all medical specialties and can affect all patients: young and old, male and female, those unaware of underlying illnesses and those diagnosed with life-limiting conditions. Thankfully over the past decade, remarkable progress has been made in the field of medical genetics. The development and integration of new testing technologies—whole exome sequencing (WES) and whole genome sequencing (WGS)—has accelerated the discovery of genetic conditions and massively improved diagnosis and management for patients.

With the significant increase in the number of genetic tests now available and requested, there has been a parallel increase in the complexity of test ordering. Consequently, a need has arisen to assist non-genetic providers with results interpretation. As a result, genetic counseling has become an integral part of today's healthcare system.

Genetic 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.[1] The most common are hemoglobinopathies such as β-thalassemia, α-thalassemia and sickle cell disease. The abnormal gene frequency for β-thalassemia (including β^S for sickle cell) is as high as 8.5%,[2] and the positive screen rate for α-thalassemia among Emiratis is approximately 50%.[3] Other common genetic disorders in the country include:

- G6PD (incidence ranges from 11-15% in males)[4]
- metabolic disorders (prevalence ~ 1 in 1,329 Emiratis)[5]
- hearing impairments
- hereditary cancer syndromes
- congenital abnormalities
- intellectual disabilities and developmental delays
- chromosomal syndromes
- cystic fibrosis

Approximately 60% of all genetic disorders in the UAE are autosomal recessive,[6] 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%)[7]
- gene pool homogeneity
- the founder effect: a group of patients in a certain geographical area having a particular genetic disorder, as a result of a common mutation inherited from a common ancestor
- selective environmental elements that give a survival advantage to carriers: carriers of beta-thalassemia or sickle cell disease are protected against malaria[8]

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.

About genetic counselors & how they support patients

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 preventive options)
- referrals to other medical specialties, advocacy and support groups to help them effectively deal with their diagnosis

The Unified Healthcare Professional Qualification Requirements[9]—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.

The benefit of working collaboratively with a licensed genetic counselor

- ✓ Through the provision of a comprehensive genetic counseling service, your patient's overall satisfaction and treatment outcomes will be enhanced
- ✓ You receive support in determining the optimum genetic testing strategy for each of your patients and their family members, thereby increasing efficiency and decreasing liability
- ✓ There is knowledge sharing on specimen collection, specimen storage and test sensitivity
- ✓ You have access to specialized expertise in results interpretation and recommendations

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