MEDICAL COUNCIL OF CANADA | LE CONSEIL MÉDICAL DU CANADA

Genetic concerns

(January 2017)

Rationale

An individual's genetic make-up has an impact on their development, as well as their predisposition to disease. Genetic variation and mutation may cause disease directly, or interact with various experiential and environmental factors to influence development and medical conditions.

Causal Conditions

(list not exhaustive)

- Chromosomal (e.g., aneuploidy, rearrangements)
- Single-gene
    a. Mendelian (e.g., autosomal dominant)
    b. Non-Mendelian (e.g., mitochondrial, epigenetic)
- Prenatal Exposure
    a. Drugs or toxins (e.g., fetal alcohol spectrum disorder)
    b. Infectious (e.g., congenital rubella)
    c. Maternal disease (e.g., maternal diabetes)
- Multifactorial (e.g., neural tube defects)

Key Objectives

The candidate will recognize situations where a person or a population is at risk of a genetic or epigenetic condition. Given a patient with evidence of, or a family history consistent with, a genetic or congenital condition, the candidate will diagnose the cause, severity and complications, and will initiate an appropriate management plan.

Enabling Objectives

Recognize where disease in an individual might reflect the existence of risk factors inherent to a given population (e.g., Tay-Sachs disease).

Given a patient presenting with clinical findings suggestive of a genetic etiology, the candidate will

- list and interpret relevant clinical findings, including
    a. obstetrical, medical and family history, as well as ethnic or geographic origin and social determinants of health;
    b. results of a physical examination of the patient and of selected family members, if need be;
- list and interpret relevant laboratory and diagnostic imaging;
- construct an effective initial management plan, including, if required:
    a. genetic counselling;
    b. examination of reproductive options;
    c. a referral for specialized evaluation, genetic testing, community resources, social and psychological support services.