MEDICAL COUNCIL OF CANADA | LE CONSEIL MÉDICAL DU CANADA

Congenital anomalies, dysmorphic features

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Rationale

Congenital anomalies and dysmorphic features can be associated with long-term disability, making early detection and identification vital. Although early involvement of pediatric or genetic specialists is appropriate, primary care physicians are often required to contribute immediate care and assist with long-term management.

Causal Conditions

(list not exhaustive)

- Teratogenic disorders (e.g., fetal alcohol spectrum disorder, congenital cytomegalovirus infections)
- Genetic disorders (e.g., trisomy 21, fragile X syndrome)
- Mechanical forces (e.g., constriction band syndrome)

Key Objectives

Given a patient with congenital anomalies or dysmorphic features, the candidate will investigate the cause, determine the severity of the immediate presentation, and initiate an appropriate management plan. Particular attention should be paid to the identification of patients requiring early referral for specialized care and to the provision of supportive counselling for parents.

Enabling Objectives

Given a patient with congenital anomalies or dysmorphic features, the candidate will

- list and interpret critical clinical findings, including those derived from
    a. a complete history, with particular attention to any potential teratogenic exposures, and a detailed family history; and

b. an appropriate physical examination, with particular attention to signs of severe anomalies (e.g., cardiovascular malformations, ambiguous genitalia) as well as to recognizable phenotypic patterns (e.g., trisomy 21 syndrome);

- list and interpret appropriate investigations (e.g., microarray; karyotyping; screening for toxoplasmosis, other agents, rubella, cytomegalovirus, herpes simplex [TORCH]); and
- construct an effective initial management plan, including
    a. stabilization and immediate referral in case of respiratory and/or hemodynamic instability;
    b. referral for specialized pediatric or genetic care if necessary;
    c. referral for therapeutic services, counselling, and family support groups, if indicated; and
    d. provision of family support and counselling regarding recurrence risk, including discussion of prenatal strategies for the prevention of recurrence, indications for antenatal screening and diagnostic prenatal testing, and referral for genetic counselling, if indicated.