McMaster University

Genetics
Rotation Specific Objectives
Department of Pediatrics
McMaster University

MEDICAL EXPERT

The learner should gain a basic understanding of:

1. The mendelian modes of inheritance (autosomal and X-linked, dominant and recessive).
2. The “non-mendelian” patterns such as mitochondrial inheritance, imprinting, anticipation, uniparental disomy, etc.
3. Multifactorial inheritance.
4. How chromosomes segregate in meiosis and mitosis from both normal and abnormal parental contributions.
5. The concepts of trisomy rescue and confided placental mosaicism.
6. Types of tests currently available for prenatal diagnosis (e.g., CVS, amnio, ultrasound, MSS).
7. Principles of screening.
8. The genetic screening programs used in Ontario (e.g., newborn, MSS, LMA).
9. The approach to the dysmorphic and/or developmentally delayed child.
10. The difference between malformations, deformations, disruptions and sequences.
11. The concept of minor anomalies.
12. The consequences of exposure to various agents during fetal development, especially alcohol, diagnostic radiation and anticonvulsants.
13. The effect of maternal disease on the fetus, especially maternal diabetes and maternal PKU.
14. Elicit an appropriate genetic family history using standardized notation, with an awareness of time constraints.
15. Elicit a pertinent pregnancy, developmental, medical and social history.
16. Apply the family history information to the clinical situation (e.g., interpretation, risk assessment and calculation).
17. Assess which type of testing is appropriate to order in the investigation of genetics patients (e.g., metabolic, chromosome or DNA tests).
18. Assess which type of prenatal testing could be offered in a particular clinical situation.
19. Recognize a dysmorphic child and appreciate how minor congenital anomalies contribute to the whole picture.
20. Comprehend basic cytogenetic and molecular lab reports.

COMMUNICATOR

1. The ability to convey genetic, medical and technical information in a clear way to families.

2. The ability to access lay materials written for patients, as well as support group information.

COLLABORATOR

1. Demonstrate the ability and willingness to collaborate with the entire health care team.

MANAGER

1. Ability to prioritize and manage multiple simultaneous clinical demands.
2. To show an awareness of cost-benefit considerations in patient care.

HEALTH ADVOCATE

1. The ability to recognize the advocacy needs of children with genetic or metabolic disorders.

SCHOLAR

1. The ability to develop and implement an effective learning strategy.

PROFESSIONAL

1. To demonstrate integrity, honesty and compassion.
2. To demonstrate an awareness of the psychosocial impact of genetic conditions on families.
3. To understand the issues surrounding genetic testing of adolescents and children.

ADDITIONAL INFORMATION

Each resident will be exposed to a counseling session for *at least one* case of each of the following types of inheritance possible:

- Autosomal recessive
- Autosomal dominant
- X-linked recessive
- Multifactorial
- Cytogenetic
- DNA-based diagnostics
- Predictive testing
- Mitochondrial

Most of these encounters will be under the supervision of one of the four genetic counselors.

Modified: August 10, 2007