





S/he is informally assessed throughout the rotation by direct supervision and is given feedback. At the end of the rotation, the supervisor, in consultation with other staff members, completes the evaluation form and discusses the evaluation with the student.

### **Rotation Specific Educational Objectives**

Upon completion of a rotation in genetics, the student is able to:

#### **Medical Expert**

- Gather medical and family history; construct a three-generation pedigree
- Recognize patterns of inheritance and the recurrence risks for each
  - Mendelian: autosomal recessive, autosomal dominant, X-linked recessive, X-linked dominant
  - Non-Mendelian: multifactorial, imprinting, uniparental disomy, mitochondrial
- Carry out and properly document a comprehensive physical examination with beginning expertise in features of surface anatomy and anthropometric measurement.
- Follow a logical approach in syndrome identification including the use of diagnostic aids (eg. computer assisted diagnosis, literature searches), especially in the context of the child with multiple anomalies.
- Recognize common dysmorphic syndromes (including Trisomy 21, 18, 13, Fragile X, Williams, DiGeorge, Prader-Willi and Angelman syndromes), be familiar with the approach to evaluate patients with these syndromes and to refer them to the genetic specialist for diagnosis.

#### *For the Child with an Inborn Error of Metabolism:*

- Recognize the clinical features that are suggestive of an inborn error of metabolism
- Understand the initial evaluation of patients with a suspected inborn error of metabolism.
- Understand the basis of management of the child presenting with a metabolic crisis (with back-up from the Genetics consultant).

#### *For Genetic Counselling:*

- Draw and read a pedigree.
- Understand the principles of Mendelian and non-mendelian inheritance in order to understand recurrence risk for future children (AR, AD, X linked, Mitochondrial, Imprinting, Penetrance, Expressivity, Microdeletion syndromes).

#### *For Cytogenetics and Genetic Testing:*

- Understand the different cytogenetic tests (karyotype, high resolution karyotype, FISH analysis, microarray) and the common indications for ordering them.
- Understand the use and limitations of molecular genetic and biochemical genetic diagnostic tests.

#### **Communicator:**

Demonstrate appropriate communication skills regarding:

- Patient communication, directly to the patient and his/her family
- Written communication and records as documented by clear notes in the medical chart and well-written, informative letters written to the referring physician

#### **Collaborator:**

- Interact appropriately with other trainees, physicians and health professionals in clinical situations.
- From role modeling, appreciate how to be respectful of each member of the health care team and emulate that approach in his/her interactions with team members.



**Scholar:**

- Articulate a clinical question, perform a literature review, and critically appraise this review in order to develop an answer to the clinical question based on best available evidence.
- Develop skills in the presentation of scholarly work to colleagues

**Manager:**

- Demonstrate effective time management in the balancing and prioritizing of clinical and academic duties, as well as those of the program
- Describe an approach to the rational use of common genetic screening tests

**Advocate:**

- Appreciate community resources needed to undertake successful discharge planning for the child with complex needs (such as children with inborn errors of metabolism)
- Describe the rationale behind genetics screening programs, understanding the need to promote screening and diagnosis of genetic diseases in at risk populations (understanding also the "down side" of such programs (stigmatization, discrimination etc.)

**Professional:**

Show:

- Respectful interactions with patients, families, colleagues, and co-workers
- A sense of ethics and responsibility
- Sensitivity to the diversity of humanity

**Prerequisites**

None. However, students may benefit from having been exposed previously to pediatrics.

**Scope of Work**

With supervision:

1. Obtain histories and perform physical examinations on patients in clinic and inpatient settings.
2. Obtain family histories and draw pedigrees.
3. Formulate differential diagnoses and plan of investigations.
4. Develop management plans.

**Recommended Reading/Preparation**

1. GeneReviews: an online reference
2. The Oxford Desk Reference: Clinical Genetics and Genomics (available through the McGill medical library)



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