Proposal: Clinical Biochemical Genetics Fellowship certified by the Canadian College of Medical Geneticists

Name of Institution: McGill University Health Centre
Location: Montreal, QC
Number of positions: One every two years
Length: 2 years (maximum) (Length of training to be assessed by the Canadian College of Medical Geneticists (CCMG), which certifies the training

Program Information: See appendices (General Guidelines, CCMG Training and Specialty Requirements in Clinical Biochemical Genetics, Logbook-Clinical Biochemical Genetuics, Logbook of Educational activities, ITER-Clinical Biochemical Genetics, FITER-Clinical, CCMG Rotation Evaluation, CCMG Faculty Evaluation, Research Summary)
Number of fellowship positions requested: One every two years
Academic affiliation: McGill University Health Centre (MUHC)

McGill University Health Centre (MUHC) – 92% (minimum)
Clinical Biochemical Genetics, Cytogenetics and Molecular Laboratory, Clinical Genetics And Biochemical Genetics Laboratory

Specific rotations in laboratories of non-affiliated hospitals:
- CHU Ste-Justine -1%
- CHU Sherbrooke- 1%
- Children's Hospital of Eastern Ontario- 1%
- CHU Quebec -1%

Electives and Research – 4%

Background:
McGill University has long been a training site for clinical genetics and laboratory genetics fellowships that are certified by the CCMG. We would like for these fellowships to be recognized by the Postgraduate Medical Education Office.

The Clinical Biochemical Genetics Fellowship is **2-year** training program that is certified by the Canadian College of Medical Geneticists (CCMG).Trainees are expected to participate fully in all aspects of clinical medicine as it relates to biochemical genetics, multi-disciplinary case discussion, rounds, seminars and meetings related to clinical and biochemical genetics. There is increasing responsibility over the training period to include more independence in patient encounters, counseling and provision of patient/family support, treatment and management, the interpretation of laboratory results and other competencies as outlined in the Training Objectives below.

Research: The trainee will participate in a research project that will be identified in the second year of training. Lysosomal, mitochondrial, neuropsychiatric- are just a few of clinical fields where clinical research could be completed. Laboratory research expertise in the Department of Medical Genetics includes Peroxisomal Disorders (Dr. Nancy Braverman), B12 and folate disorders (Dr. David Rosenblatt). There are other biochemical genetics related research opportunities in other departments.

Mission: To produce clinical specialists who are competent to effectively diagnose and manage individuals of all ages with inherited metabolic disease.

Outline: This fellowship will enhance the residency training of the fellow by providing training in provision of having a thorough grounding in the diagnosis, investigation and management of a broad spectrum of disorders typically encountered in the biochemical genetics setting.

Name of the Fellowship Program Director: Dr. Daniela Buhas

Names of the Teaching Faculty

- D. Buhas, N. Braverman, Y. Trakadis, F. Parente, B. Gilfix, D. Rosenblatt: biochemical laboratory test methods and interpretation
- J. Lavoie and M. Blumenkrantz : Cytogenetics laboratory training and result interpretation
- A. Ruchon and I. DeBie: Molecular Genetics laboratory training and result interpretation
- D. Buhas, N. Braverman, J. Mitchell ,Y. Trakadis, I. DeBie, L. Russell, D. D'Agostino: clinical and biochemical genetics
- CHU Sherbrooke--P. Waters and C. Aurais-Blais: biochemical laboratory test methods and interpretation
- CHU Ste-Justine--P. Allard and C. Brunel-Guitton: biochemical laboratory test methods and interpretation
- Chu Quebec--Y. Giguere and M-T. Berthier: biochemical laboratory test methods and interpretation
- CHEO--N. Lepage and O. Al-Dirbashi: biochemical laboratory test methods and interpretation

The teaching faculty has a broad range of expertise in a number of different types of laboratory biochemical genetics, molecular genetics and cytogenetics. A number of the teaching faculty also have extensive experience in pediatric and adult clinical and biochemical genetics. The clinical and laboratory faculty collaborate closely with respect to patient care and training of residents and fellows.

Academic Facilities

The MUHC Biochemical Clinical Genetics (BCG) team includes physicians, nurse and metabolic dietician. The pediatric BCG clinics, scheduled twice a week, serve patients with known or suspected inborn errors of metabolism. Once a month, there is a BCG Adult clinic, where known metabolic patients or under investigations are evaluated. The inpatient service is actively involved in the diagnosis and the management of patients with biochemical genetic phenotypes.
The care of these patients is discussed weekly at a case review conference.

The MUHC offers a variety of biochemical genetic laboratory investigations. The clinical investigations include plasma, CSF and urine amino acids, acylcarnitine profile, transferrin isoelectric focusing, vitamin B12 and folate disorders, homocysteine and disorders of porphyrin metabolism. As well, there is research laboratory for peroxisomal disorders. These laboratories maintain a collection of teaching cases, consisting of patient clinical presentation and laboratory abnormalities.
The MUHC also has laboratories for molecular genetics and cytogenetics. As well, the MUHC Cell Bank serves as repository for fibroblasts and other tissues obtained from patients with a variety of disorders.

In addition to these laboratory facilities, the clinical teams (physician, genetic counsellors, nurse and metabolic dietician) of the MUHC Department of Medical Genetics hold outpatient clinics a minimum of four days a week. A weekly Academic Half-Day includes Basic Science seminars, Clinical Case Presentation, Journal Club and clinically oriented Resident Teaching.

The fellows and other trainees have full time electronic access to the McGill University libraries. As well, the Department of Medical Genetics maintains its own small library of books relevant to many aspects of medical genetics. Finally, the Department offers full time access to London Database, a database of physical traits and syndromes.

Fellow Duties and Responsibilities

- Call responsibilities to cover service: *the fellow will be involved in the evaluation of patients with known/suspected metabolic condition in an "availability "mode (ER presentation, in-patients, NBS or Medical Day evaluations), thus being involved in that cases that would most contribute to his learning*
- The fellow is the senior supervisor of residents: *Only during the fellow's second year of training. In the last clinical BCG rotation the fellow will be acting as junior staff (with minimal supervision)*
- Outline whether there are fixed rotations at the various institutions: *Yes*
- Outpatient clinic responsibilities need to be outlined: *The fellow is expected to gain experience in all aspects of diagnosis and management of metabolic disorders in both children and adults*
- Outline the role of the fellow towards residents on service: *The fellow will actively participate as a team member in the evaluation and management of metabolic patients. In the second year of training, his supervision role will increase gradually.*
- Teaching responsibilities towards residents: *The fellow will apply teaching skills with residents on service, under the supervision of staff in Biochemical Genetics. The fellow will deliver effective lectures and presentations on the concepts of inborn errors of metabolism.*
- Outline participation in academic activities involving the residents: seminars, outcome assessment (morbidity and mortality rounds etc). *The fellow will attend the weekly Academic Half Day and will give Journal Club at least once per year. He will also attend the weekly case discussion of patients with inborn errors of metabolism; as part of his participation in this meeting, he will develop and present one or more topics relative to his interests. At the end of the fellowship, he will present a seminar that summarizes his fellowship project.*
- Describe any support staff available to the fellow: program coordinator, nurse clinician, secretarial *The fellow has support from the nurse, metabolic dietician and technicians in the various laboratories, including the Biochemical Genetics laboratory. Once the fellowship is approved, the fellow will have the support of a program coordinator.*
- Proposed meetings to be attended by the fellow *The fellow will attend the weekly case discussion of patients with inborn errors of metabolism and the weekly Academic Half Day. He will also attend weekly discussions of specific metabolic topics or abnormal biochemical laboratory test results that are pertinent to his training. The fellow will also attend an annual genetics meeting of his choice, such as the Garrod Association or the Society for Inborn Metabolic Disorders.*
- Research productivity and publications expected by the Fellow. *The fellow will develop a research project under the supervision of his training committee. The fellow will be encouraged to present this research project at a scientific meeting or to prepare a manuscript that is suitable for publication.*

Curriculum

- Intended case load
  A logbook documenting at least 150 cases for whom the fellow has assumed consultant level of responsibility. This should reflect a broad range of metabolic disorders including inpatient and outpatient consultation in both the pediatric and adult setting.
  The fellow is expected to gain experience in interpreting results and communicating to others, with a logbook recording involvement in 50 cases.
  At least 120 of the 200 (combined clinical and laboratory) cases should represent known or newly diagnosed IEM. The remainder may include clinical cases where a diagnosis of an IEM may reasonably be considered part of the differential diagnosis. Cases may include telehealth and/or telephone consultations but the fellow involvement should clearly indicate discussion of history, exam and initial investigation, formulation of a differential diagnosis in that particular case as well as advising on diagnostic workup and plan for management.

- Intended Percentage of varieties of cases. *Not specified but exposure should be broad.*

  Regular reading materials provided (if any) :

1. *Techniques in diagnostic human biochemical genetics A Laboratory Manual, edited by Frits A. Hommes, Wiley-Liss New York 1991.*
2. *Physician's guide to the Laboratory diagnosis of metabolic diseases, edited by N Blau*
3. *Inborn metabolic diseases - diagnosis and treatment, edited by J Fernandes, J-M Saudubray and Van den Berghe*
4. *A clinical guide to inherited metabolic diseases, edited by JTR Clarke (second edition)*
5. *Inherited Metabolic Diseases, edited by GF Hofmann (a copy will be provided)*
6. *Scriver's Online Metabolic and Molecular Bases of Inherited Disease,edited by Valle, Beaudet,Vogelstein, Kinzler, Antonarakis,and Ballabio(online access provided)*

- Conference weekly schedules *The fellow will attend the weekly case discussion of patients with inborn errors of metabolism and the weekly Academic Half Day. He will also attend weekly discussions of specific metabolic topics or abnormal biochemical laboratory test results that are pertinent to his training.*
- Role of the fellow in attending, presenting, supervising, organization *See above under Fellow Duties and Responsibilities*

Canadian College *of* Medical Geneticists
Collège canadien *de* généticiens médicaux

CCMG Clinical Biochemical Genetics Training Guidelines and Specialty Requirements

Preamble

The aim of the Clinical Biochemical Genetics Training Program is to produce clinical specialists who are competent to effectively diagnose and manage individuals of all ages with inherited metabolic disease. Competence implies the individual has the knowledge, skills and attitudes to:

1) Recognize patients with metabolic disease and request the appropriate investigations for confirmation of diagnosis;
2) Interpret results from laboratory investigations of inborn errors of metabolism;
3) Manage patients (both acute and long term care) and their families with metabolic disorders;
4) Assume the day-to-day responsibilities for the operation and standards of a Metabolics clinic.

The Clinical Biochemical Geneticist will have a thorough grounding in the diagnosis, investigation and management of a broad spectrum of disorders representing all modes of inheritance and indications typically encountered in the biochemical genetic setting.

Trainees are expected to participate fully in all aspects of clinical medicine as it relates to biochemical genetics, multi-disciplinary case discussion, rounds, seminars and meetings related to clinical and biochemical genetics. There is increasing responsibility over the training period to include more independence in patient encounters, counseling and provision of patient/family support, treatment and management, the interpretation of laboratory results and other competencies as outlined here in the Training Guideline.

The CCMG training guidelines are modeled after the CanMEDS framework[1]. This framework includes the competencies required of specialists and the role of the specialist beyond that of the specialty medical expert. The other roles of the specialist are that of communicator, collaborator, manager, health advocate, scholar, and professional. The detailed objectives describe minimal standards and in no way exclude the necessity for mastery of additional knowledge, skills or attitudes necessary for the clinical practice of biochemical genetics.

Required Background

Trainees must have a MD (or equivalent e.g. MB BS) degree.
A candidate's MD specialty must be in Genetics, Pediatrics, Internal Medicine or Medical Biochemistry (FRCPC qualified or Board eligible). Consideration will be given to alternate qualifications (e.g. ABMG, etc) on an individual basis. Trainees must have the appropriate licensing requirements to complete their clinical training.

Administrative Aspects

1. **Supervisory committee:**
    a. Each Clinical Biochemical Genetics trainee's program will be supervised by a committee, headed by:
        - a Fellow of the CCMG in Biochemical Genetics with 2 years post Fellowship experience; or
        - a Medical Geneticist with 2 years experience in the practice of inborn errors of metabolism; or
        - a physician specialist with 5 years experience in the practice of inborn errors of metabolism; or
        - co-chaired by a CCMG Laboratory Biochemical Geneticist and an experienced Metabolic Specialist.

        A Specialist who is not a Fellow of the CCMG in Biochemical Genetics must be accepted by the Metabolics committee and approved by the Credentials committee of the CCMG and be required to be an Affiliate member of the CCMG.
    b. The committee will consist of the supervisor (above) and a minimum of two additional members. One member of the committee must be a Fellow of the CCMG. Other members may consist of clinical specialists with experience in care of patients with Inborn Errors of Metabolism, laboratory-based biochemical geneticists, laboratory physicians or clinical chemists. The structure of the committee can vary depending on the background of the trainee.
    c. The program director or supervisor on behalf of the committee ensures the trainee is registered with the CCMG Credentials Committee by submitting a registration form to the CCMG Secretariat by August 1 of the first year of training.
    d. The committee takes responsibility for ensuring that the training program is meeting the needs of the trainee and is in keeping with CCMG guidelines, including graduation of responsibility in the clinical setting. The committee must submit an outline of completed and planned training with the trainee's application for credentialing.
    e. The committee meets every six months with the candidate and ensures that all in-training evaluation forms (ITER; one for each rotation and one for every 6 months for longer rotations) are completed and discussed with the trainee. If remedial work is needed by the trainee, the committee must ensure that this is provided.
    f. The committee completes and submits the Final In-Training Evaluation Report (FITER) to the CCMG secretariat by August 1 of the final year of training. Please note that the FITER is additional to the ITER covering the last 6 months of training.

2. **Location of training**
    a. Clinical Biochemical Genetics training must take place in a CCMG-accredited training center. Note the centre does not need to be accredited for training in Clinical Biochemical Genetics.
    b. Elective training may be done at non-accredited centres at the discretion of the supervisory committee.

c. In the event that accreditation of a center is terminated during the candidate's training, the trainee will be allowed a maximum of six months to move to an accredited centre.

3. **Training in foreign centers**
   a. Training in an American center accredited by the American Board of Medical Genetics (ABMG) is recognized by the CCMG.
   b. As the ABMG and CCMG have different training and credentialing requirements, it is the responsibility of the trainee to ensure completion of all requirements of the CCMG.

4. **Part-time training**
   a. Part-time training is recognized by the CCMG, provided it conforms to all requirements in this document and the trainee spends a minimum of 50% of time in the training program.
   b. The total amount of time must equal the required period of training.

5. **Second specialty training**

   CCMG fellows (holding a MD with the appropriate background; see "required background" above) currently certified in a laboratory specialty who want to certify in Clinical Biochemical Genetics must complete a minimum of 12 months full time training in a CCMG-accredited facility that provides Clinical Biochemical Genetics/Metabolic services. All training as outlined below in Mandatory training item 1 must be completed. The Supervisory Committee may recommend additional training depending upon previous training and experience.

6. **Credentialing**
   a. Candidates are advised to review the Credentialing requirements on the website early in their training to facilitate Credentials submission and review.

Content of Training

Trainees with Medical Genetics training, one year program (exemption is given for Clinical Genetics, Molecular/Cytogenetic Laboratory, Research and Elective rotations); completion of mandatory training item 1 is required.

All other trainees, two year program.

Mandatory Training:

1. **Minimum of 12 months in a CCMG-accredited facility providing Biochemical Genetics (Metabolics) service**. *Training sites must ensure compliance with their clinical accreditation programs. The Supervisory Committee must ensure that the service load does not interfere with the educational goals of the program.*

   It is recognized that not all training centres will be able to provide a fully-comprehensive training program. Those responsible for training at a given center must identify any 'gaps'

and encourage trainees to obtain the appropriate training at other CCMG- or ABMG-accredited centres to meet the overall training objectives of the program.

a. Clinical skills: Trainees are expected to gain experience in all aspects of diagnosis and management of metabolic disorders in both children and adults. A logbook documenting at least **150 cases** for whom the trainee has assumed consultant level of responsibility. This should reflect a broad range of metabolic disorders including inpatient and outpatient consultation in both the pediatric and adult setting.

b. Laboratory skills: Trainees are expected to gain experience in interpreting results and communicating to others, with a logbook recording involvement in **50 cases**. Laboratory cases should be case based (1 case per patient, although the same patient can be listed under clinical log if the trainee is involved in both aspects of care). Laboratory cases can also be archival. All lab cases to be reviewed by laboratory supervisor. It is the sole responsibility of the local fellowship committee to determine the total number of cases to be reviewed by the candidate to ensure a high level of competence.

Case distribution: At least 120 of the 200 (combined clinical and laboratory) cases should represent known or newly diagnosed IEM. The remainder may include clinical cases where a diagnosis of an IEM may reasonably be considered part of the differential diagnosis. Cases may include telehealth and/or telephone consultations but the trainee involvement should clearly indicate discussion of history, exam and initial investigation, formulation of a differential diagnosis in that particular case as well as advising on diagnostic workup and plan for management.

c. Management skills: Experience in management of a Metabolic Clinic.

2. Rotations in other medical genetics specialties, including

a. **Clinical genetics training**: a minimum of **two months** on a clinical genetics service supervised by an expert in clinical genetics whose credentials are acceptable to the CCMG. During this time the fellow should gain experience in multiple aspects of clinical genetics including pediatric and adult genetics and prenatal diagnosis. The trainee must document **25 cases** in their logbook demonstrating experience with a variety of cases.

b. **Biochemical Genetics Laboratory**: a minimum of **three months** to be spent in a CCMG-accredited laboratory providing biochemical genetics laboratory services.

c. **Molecular and Cytogenetics laboratory training**: a minimum of **one month** to be spent in a CCMG-accredited laboratory or laboratories providing molecular and cytogenetics diagnostic services.

3. Courses/conferences

a. Documented participation in educational events and courses prescribed by the trainee's supervisory committee.

b. Documented attendance at one local (e.g. departmental annual research day, a research institute research day), national or international genetics meeting during the training period.

4. Elective or research training

The trainee must complete 6 months full time elective clinical training relevant to the specialty and/or research training. The 6 months can be devoted to one elective or divided amongst two or more electives. This time is intended to allow a trainee to further training in a clinical field relevant to Biochemical Genetics (including additional training in one of the above rotations) or research. All electives require approval by the candidate's supervisory committee.

Research training

a. The trainee may participate in clinical or laboratory-based research for a period up to the equivalent of 6 months full time research; this may be accomplished through a dedicated block of time or distributed throughout the training period. Training may be obtained at the training centre or another hospital or university centre in Canada or abroad as approved by the candidate's supervisory committee and Program Director. The research can be applied or translational in nature such as development of a new test, test validation, test improvement, quality improvement, case follow up and cost-benefit analysis. The aim of the Research rotation is to acquire basic competencies ie formulating a question, preparing a proposal, understanding the REB and funding processes, managing a study and disseminating results through presentation and /or publication. Candidates may seek credits for this rotation, particularly if research was a designated part of their RCPC residency and/or they have prior research experience (eg MSc, PhD, publication record).
b. A research supervisor must be identified and the proposed research objectives and methodology are to be submitted to the supervisory committee for review and approval. The research supervisor is responsible for completing an evaluation (ITER) of the trainee. The trainee is responsible for a written summary of completed research to be submitted to Program director and dissemination to the appropriate audience (laboratory staff, clinicians, publication).

5. Logbooks

CCMG Logbook templates available on the CCMG website **must** be used. Patient confidentiality must be guarded. Therefore, before submitting to the CCMG, cases must have all identifiers removed so as not to be traceable. Each Logbook is in an Excel format with tabs for documentation of:

a. Metabolics service experience (tab: Clinical Biochem Log)
b. Biochemical Genetics laboratory experience (tab: Lab Biochem Log)
c. Clinical Genetics experience (tab: Clinical Log)
d. Education experience (tab: Educational Activities)
e. Research experience (tab: Research Log)
f. Outline of training program (tab: Training Outline)

A logbook should not only be viewed as a mechanism for tracking the number of cases/experiences accumulated but as a means for documenting learning, illustrative cases and approaches undertaken that can be reflected upon and recalled as you study for the CCMG exam and as you start your career as a laboratory geneticist. The logbook should be reviewed regularly and discussed by the supervisor and trainee to ensure they represent the breadth of cases required and acquisition of competencies.

Clinical Biochemical Genetics Specialty Requirements

Key and Enabling Competency Statements

Note:
The 7 *Roles* are the thematic groups of competencies that organize the CanMEDS format (Medical Expert, Communicator, Collaborator, Manager, Health Advocate, Scholar, Professional). The training and specialty requirements were developed according to the CANMED 2005 Guidelines (1)

The *Key Competencies* are the overall culminating objectives of training. They are meant to be summative and cumulative, while also being observable and measurable.

The *Enabling Competencies* are the skills that allow the *Key Competencies* to be achieved. The *Enabling Competencies* break-down the *Key Competencies* into observable and measurable statements

Medical Expert / Clinical Decision-Maker

Key Competencies

By the end of training, the Clinical Biochemical Genetics Trainees will be able to:

1. Explain general concepts in cell biology, human biochemistry and human genetics
2. Explain normal physiology including prematurity, changes during childhood, ageing and pregnancy.
3. Demonstrate knowledge and skills in Inborn errors of metabolism (IEM)
4. Demonstrate knowledge of Clinical Phenotypes commonly seen in Metabolic disorders
5. Demonstrate knowledge in Clinical Genetics
6. Demonstrate the ability to examine, interpret, and understand laboratory data and results relevant to the investigation of IEM, and understand their clinical significance

Enabling Competencies

1. **Explain general concepts in cell biology, human biochemistry and human genetics**
   To achieve this, the Clinical Biochemical Genetics Trainee will be able to:
   1.1 Describe and discuss general concepts of human biochemistry and molecular biology, including:
   - Structure/function relationships of intracellular components: nucleus, Golgi, endoplasmic reticulum, mitochondria, lysosomes, peroxisomes

- Enzymes/proteins: structure/function relationships, cellular distribution, mechanisms of action, control of enzyme activity, principles of measurement, enzyme kinetics
- Regulation of intermediary metabolism including biochemical and hormonal regulation, and tissue compartmentalization.

1.2 Discuss the general concepts of medical genetics outlined in the CCMG General Knowledge Guidelines

2. **Explain normal physiology including prematurity, changes during childhood, ageing and pregnancy.**
   To achieve this, the Clinical Biochemical Genetics trainee will be able to:
   2.1 Describe and discuss general concepts of human physiology and biochemistry including:
   - Fluid and electrolyte balance, acid-base regulation, intermediary metabolism and metabolic response to fasting
   - The function, and functional anatomy, of major organ systems

   2.2 Understand principles of normal nutrition and consequences of under-nutrition and specific nutritional deficiencies

3. **Demonstrate knowledge and skills in** Inborn errors of metabolism (IEM)
   To achieve this, the Clinical Biochemical Genetics trainee will:
   3.1 Demonstrate an understanding of the pathological and biochemical changes, clinical symptoms, investigations and management in metabolic disorders of the following pathways:
   - Disorders of amino acid metabolism
   - Disorders of organic acid metabolism
   - Hyperammonaemia and urea cycle disorders
   - Disorders of carbohydrate metabolism: glycogen storage disease, galactosemia, etc.
   - Disorders of fatty acid oxidation and carnitine metabolism
   - Disorders of ketone body metabolism
   - Lysosomal storage disorders
   - Peroxisomal disorders
   - Disorders of purine and pyrimidine metabolism
   - Respiratory chain disorders and disorders of pyruvate metabolism
   - Porphyrias
   - Disorders of cholesterol, sterol and bile acid metabolism
   - Disorders of lipid metabolism
   - Disorders of metal s metabolism
   - Disorders of vitamin metabolism (e.g., cobalamin)
   - Disorders of creatine metabolism
   - Defects of membrane transport: cystinuria, lysinuric protein intolerance, etc.
   - Disorders of glycosylation
   - Disorders of neurotransmitters

   3.2 Understand indications for metabolic autopsy
   3.3 Understand the principles of treatment related to inborn errors of metabolism

- Demonstrate knowledge of the general principles of dietary management in inborn errors of metabolism
- Demonstrate knowledge of the appropriate indications for emergency /crisis management of metabolic disorders
- Demonstrate knowledge of the underlying principles, and skill in the application, of various dietary or pharmacological treatment strategies employed for metabolic disorders:
    - Substrate reduction
    - Correcting co-factor or product deficiency
    - Providing alternative substrates/promoting alternative pathways
    - Blocking effects of toxic metabolites
    - Stimulating residual enzyme activity
    - Enzyme replacement
    - Organ transplantation
    - Gene therapy

4. Demonstrate knowledge of clinical phenotypes commonly seen in metabolic disorders
To achieve this, the Clinical Biochemical Genetics trainee will:

4.1 Perform a complete evaluation of physiological and pathological states relevant to inborn errors of metabolism. In order to do this the trainee must be able to:

4.1.1. Elicit a comprehensive medical history and an appropriate family history, and to construct and interpret a standardized pedigree, and calculate risks

4.1.2. Carry out a comprehensive physical examination

4.1.3 Appreciate clinical variation or phenotypic spectrum within genetic disorders;

4.1.4 Formulate an appropriate differential diagnosis, and plan an appropriate course of investigation with respect to genetic disease

4.2 Formulate an appropriate metabolic differential diagnosis, and plan an appropriate course of investigation with respect to the following:

- Acute encephalopathy
- Chronic encephalopathy, including leukodystrophies
- Newborn screening results
- Liver disease including acute liver failure, intrahepatic cholestasis
- Organomegaly
- Cardiomyopathy
- Eye disease: corneal clouding, cataract, retinal changes, optic neuropathy
- Specific dysmorphic syndromes associated with IEM
- Skeletal abnormalities of metabolic disorders (e.g., dysostosis multiplex, rhizomelic chondrodysplasia punctata, frequent fractures)
- Hair and skin manifestations of metabolic disorders (e.g., ichthyosis, angiokeratoma, lipomatosis)
- Malodour (e.g., trimethylaminuria)
- Renaldisorders: Fanconi syndrome, récurrent rénal calculi
- Muscle disease: myopathy, exercise induced rhabdomyolysis
- Sudden Unexpected death

- Hypoglycemia
- Metabolic acidosis
- Hyperammonemia
- Lactic acidemia

5. Demonstrate knowledge of clinical genetics

To achieve this, the Clinical Biochemical Genetics trainee will:

5.1 Appreciate clinical variation or phenotypic spectrum within genetic disorders
5.2 Apply concepts of human heredity (dominant vs. recessive, autosomal vs. sex linked, nuclear vs. mitochondrial, single gene vs. multi-factorial) to clinical situations
5.3 Advise on prenatal diagnostic options and understand the risks and limitations
5.4 Perform pedigree analysis and risk calculation
5.5 Recommend appropriate enzymatic, biochemical or molecular carrier tests for metabolic disorders given the clinical context (family based, population based)
5.6 Advise on appropriate testing for metabolic disorders that present with dysmorphic features (e.g., CDG, storage and peroxisomal disease)
5.7 Understand the deleterious effects of toxic metabolites on the fetus (e.g., maternal PKU)

6. Demonstrate the ability to examine, interpret, and understand laboratory data and results relevant to the investigation of IEM, and understand their clinical significance

To achieve this, the Biochemical Genetics trainee will:

6.1 Advise on appropriate tests and testing algorithms in the investigation of inborn errors of metabolism
6.2 Demonstrate an understanding of the role of screening tests vs. diagnostic tests
6.3 Advise on normal and abnormal biochemical phenotypic variation including secondary causes of abnormal biochemical phenotypes (e.g., secondary causes of lactic acidemia)
6.4 Advise on the influence of clinical context and pre-analytical variables on test results: fasting, nutritional status, medications, age, gender, pregnancy, sampling procedure, etc
6.5 Appropriately use laboratory testing for long-term monitoring of metabolic disorders
6.6 Understand principles of newborn screening and demonstrate the ability to interpret results and select appropriate follow-up testing algorithms
6.7 Demonstrate proficiency in the appropriate indications for and limitations of testing, and demonstrate a familiarity and broad understanding of the methodologies as they relate to result interpretation of:
    - Amino acids
    - Organic acids
    - Intermediary metabolites (glucose, ammonia, lactate, pyruvate, free fatty acids, ketones)
    - Carnitine/acylcarnitines
    - Enzymes studies: specific and non-specific assays (flux studies), respiratory chain enzymes
    - Glycosaminoglycans and oligosaccharides
    - Transferrin isoforms, glycans and glycoprotein

- Peroxisomal metabolites (VLCFA, phytanic, plasmalogens, bile acids, pipecolic acid)
- Neurotransmitter and biogenic amine metabolites in CSF and other fluids
- Purines and pyrimidines
- Mutation analysis, including mtDNA mutation analysis

6.8 Advise on indications, patient preparation, and result interpretation of loading, fasting, and other challenge tests for disorders of intermediary metabolism metabolic autopsy

6.9 Advise on appropriate selection of reference labs for testing not offered on-site and demonstrate an understanding of the interpretation of results from reference labs

6.10 Advise on appropriate indications and interpretation of molecular genetic analyses in the context of inborn errors of metabolism

Communicator

Key Competencies

By the end of training, the Biochemical Genetics Trainees will demonstrate the ability to:

1. Effectively elicit and understand health and other information communicated by patients, their families
2. Effectively communicate health information (e.g. diagnosis, prognosis, treatment decisions, end of life decisions, genetic issues, etc.) to patients and their families
3. Consult with the Metabolics team and other relevant healthcare providers to provide optimal patient care

Enabling Competencies

1. Effectively elicit and understand health and other information communicated by patients, their families.

To achieve this, the Clinical Biochemical Genetics Trainee will be able to:

1.1. Obtain and interpret the medical, family and social history

1.2 Recognize one's own biases, including ethno-cultural differences, and their impact on communication and patient care

1.3. Understand how cultural background, age, gender, socioeconomic background and spiritual values influence communication

1.4. Use appropriate non-verbal communication

1.5 Gather information not only about the disease but also about the patient's beliefs, concerns and expectations about the disorder, while considering the influence of factors such as the patient's age, gender, ethnic, cultural, and socioeconomic background, and spiritual values

2. Effectively communicate health information (e.g. diagnosis, prognosis, treatment decisions, end of life decisions, genetic issues, etc.) to patients and their families.

To achieve this, the Clinical Biochemical Genetics Trainee will be able to:

2.1. Integrate the history with other physical and investigational findings and communicate with the patient and/or family regarding appropriate metabolic differential diagnoses, diagnostic investigations, management and prognosis
2.1.1. Display empathy and compassion, especially in delivering bad news
2.1.2. Remain objective and impartial
2.1.3. Provide psychological support either personally or through referral;
2.1.4. Employ active listening skills
2.1.5. Deliver information to the patient and family in a manner that is understandable;
2.1.6. Encourage discussion
2.1.7. Promote patient and family participation in decision-making
2.1.8. Assess with the patient and/or family as to what is their understanding of the communication
2.2. Help the individual and family choose an appropriate course of action for themselves
2.3. Advise patients and families about support agencies

3. Consult with the Metabolics team and other relevant healthcare providers to provide optimal patient care.
To achieve this, the Clinical Biochemical Genetics Trainee will be able to:
3.1 Recognize the importance of clinical or other laboratory information for cases referred for metabolic testing
3.2 Utilize clinical and other laboratory information to make decisions regarding appropriate metabolic testing to be performed
3.3 Correlate results with clinical and/or other laboratory information
3.4 Communicate biochemical genetic results and interpretation in both oral and written forms
3.5 Provide consultative services regarding the appropriate molecular tests and other additional investigations

Collaborator

Key Competencies

By the end of training, the Clinical Biochemical Genetics Trainees will demonstrate the ability to:
1. Participate effectively as a team member with relevant health care providers in collaborative decision making for metabolic cases
2. Contribute effectively to other interdisciplinary team activities

Enabling Competencies

1. Participate effectively as a team member with relevant health care providers in collaborative decision making for metabolic cases.
To achieve this, the Clinical Biochemical Genetics Trainee will be able to:

1.1. Describe the role and responsibilities of a clinical biochemical genetics professional to other health care providers
1.2. Develop rapport, trust and ethical relationships with the health care team
1.3. Participate effectively as a team member in activities related to clinical biochemical genetic, including education, research and clinical care
1.4. Demonstrate respect for other health care professionals and their role in health care teams
1.5. Network with other clinical biochemical genetics services

2. Contribute effectively to other interdisciplinary team activities
To achieve this, the Clinical Biochemical Genetics Trainee will be able to:
2.1 Participate in an interdisciplinary team meeting, and demonstrate the ability to consider and respect the opinions of other team members, while contributing biochemical genetics-specific expertise him/herself
2.2 Communicate effectively with the members of an interdisciplinary team in the resolution of conflicts, provision of feedback, and where appropriate, be able to assume a leadership role.

Manager

Key Competencies:

By the end of training, the Clinical Biochemical Genetics Trainees will demonstrate the ability to:
1. Work effectively and efficiently in a health care organization
2. Understand and apply the essential elements of Quality Management system within the clinic and/or laboratory
3. Utilize health care resources effectively;
4. Manage time effectively and prioritize required activities

Enabling Competencies

1. Work effectively and efficiently in a health care organization.
To achieve this, the Clinical Biochemical Genetics Trainee will be able to:
1.1 Demonstrate an understanding of clinical needs of the patient and family and ensure availability of relevant supports, testing and treatment options
1.2. Demonstrate understanding of the role of the various members of the "Biochemical Genetics Team" including dietician, nurse, Genetic counselor, pharmacist, laboratorian specialists
1.3 Demonstrate an understanding of the process of staff recruitment and interview skills
1.4 Demonstrate the ability to effectively prioritize the work of Metabolics staff as appropriate
1.5 Demonstrate an understanding of safety procedures

1.6 Demonstrate an understanding of the accreditation process and understand the process for responding to reviewers recommendations

2. Understand and apply the essential elements of a Quality Management system within the clinic and/or laboratory

To achieve this, the Clinical Biochemical Genetics Trainee will be able to:

2.1 Understand the basic principles of continuous quality improvement, patient safety, and quality control

2.2 Demonstrate understanding of Laboratory standards and guidelines such as external proficiency programs, turnaround times etc

2.3 Ability to respond effectively to clinical and laboratory-related complaints from an expert clinician perspective

3. Utilize health care resources effectively;

To achieve this, the Clinical Biochemical Genetics Trainee will be able to:

3.1 Demonstrate knowledge of planning, evaluation, and assessment of outcome of a health care program

3.2 Acquire the management skills required for development and use of resources in the clinic including budget control, contracting, strategic planning, writing a business plan and evaluating outcomes

4. Manage time effectively and prioritize required activities.

To achieve this, the Clinical Biochemical Genetics Trainee will be able to:

4.1 Set, prioritize and manage time to balance required activities

4.2. Recognize critical aspects of certain activities and allocate time appropriately

Health Advocate

Key Competencies

By the end of training, the Clinical Biochemical Genetics Trainee will demonstrate the ability to:

1. Advocate for appropriate clinical services for patients with inborn errors of Metabolism including the introduction of new programs, new technologies, and new treatments
2. Recognize and respond to those issues where advocacy is appropriate

Enabling Competencies

1. Advocate for appropriate clinical services for patients with inborn errors of Metabolism including the introduction of new programs, new technologies, and new treatments

To achieve this, the Clinical Biochemical Genetics Trainee will be able to:

1.1 Understand the local/Provincial/Territorial/Federal governance for the delivery of care as it pertains to Inborn Errors of Metabolism. This includes an awareness of Health Care

Provider staffing, provision of and access to diagnostic services and the availability of affordable medical/surgical treatments.

1.2. Describe the roles of national and international organizations in the determination of guidelines affecting biochemical genetics services

1.3 Understand the decision making processes with respect to current and future delivery of care for patients with Inborn Errors of Metabolism eg the introduction of new tests or treatments. This will include needs assessment, business case development, implementation and ongoing measurement of outcomes

2. Recognize and respond to those issues where advocacy is appropriate
To achieve this, the Clinical Biochemical Genetics Trainee will be able to:

2.1. Recognize and respond to those issues where advocacy is appropriate

2.2 Become informed about community resources and related patient support groups for individuals and families affected by inborn errors of metabolism

2.3. Liaise effectively with individuals, communities and populations on issues applicable to inborn errors of metabolism

2.4. Act as a resource and information source regarding biochemical genetics for individuals, communities and populations

2.5. Demonstrate understanding of the roles of national and international agencies in the promotion of health and the detection, prevention and treatment of inborn errors of metabolism

Scholar

Key Competencies

By the end of training, the Clinical Biochemical Genetics Trainees will demonstrate the ability to:

1. Conduct ongoing learning activities to maintain and advance professional knowledge
2. Facilitate the learning of other health care professionals, students, laboratory colleagues, the public and others regarding the practice of clinical biochemical genetics
3. Contribute to the development of new knowledge

Enabling Competencies

1. **Conduct ongoing learning activities to maintain and advance professional knowledge**
To achieve this, the Clinical Biochemical Genetics Trainee will be able to:

1.1 Critically assess the literature as it relates to patient investigation and diagnosis

1.2 Attend and participate in continuing education events, including conferences, rounds, clinical and research seminars, patient conferences

1.3 Recognize limitations of current knowledge base and seek appropriate continuing educational activities

1.4 Be aware of and maintain accreditation through ongoing CME programs (eg RCPC MOC program).

2. **Facilitate the learning of other health care professionals, students, laboratory colleagues, the public and others regarding the practice of clinical biochemical genetics.**
   To achieve this, the Clinical Biochemical Genetics Trainee will be able to:
   2.1 Demonstrate the willingness and ability to enhance and apply teaching skills in the education of colleagues, undergraduate and postgraduate trainees, and other health care professionals
   2.2. Deliver effective lectures and presentations on the concepts of human genetics and inborn errors of metabolism
   2.3. Present concise and audience appropriate summaries of the diagnosis, management and treatment of patients with inborn errors of metabolism and case reports or presentations

3. **Contribute to the development of new knowledge**
   To achieve this, the Clinical Biochemical Genetics Trainee will be able to:
   3.1 Demonstrate the ability to pose a research question, and to conduct a research project including the processes of formulating the proposal, Research Ethics Board approval, budgetary planning, enrollment and consent of subjects, result analysis, and finally dissemination of the results of the research
   3.2 Summarize results in a format suitable for publication in a peer reviewed journal

Professional

Key Competencies

By the end of training, the Clinical Biochemical Genetics Trainees will demonstrate the ability to:

1 Practice medicine with integrity, honesty and care
2 Exhibit appropriate personal and interpersonal professional behaviors.

Enabling Competencies

1. **Practice medicine with integrity, honesty and care**
   To achieve this, the Clinical Biochemical Genetics Trainee will be able to:
   1.1 Demonstrate understanding of the regulatory framework governing the practice of medicine. These include the Legal system as well as local Medical Advisory Councils, or Appointment Boards, Provincial and Territorial licensing bodies and Federal guidelines
   1.2 Demonstrate understanding of the importance and limitations of confidentiality
   1.3 Recognize the sensitive nature of health information and act to minimize potential harms
   1.4 Recognize ethical issues in biochemical genetics, including but not limited to the role of personal autonomy, the needs of the patient within and with respect to the family, the effects of cultural and personal beliefs, end of life decisions, as well as issues related to the impact of molecular and biochemical testing e.g. testing of minors, effects on

extended family members, testing for late onset disorders and prenatal testing, and the use of human subjects research

1.5 Identify personal limitations and the necessity of seeking the opinions of colleagues or other professionals when required

2. **Exhibit appropriate personal and interpersonal professional behaviors.**
To achieve this, the Clinical Biochemical Genetics Trainee will be able to:

2.1 Demonstrate a professional attitude to the patient and family. This includes but not limited to timeliness in communication, responsibility for proper documentation and review of results

2.2 Demonstrate a professional attitude to clinical and laboratory colleagues, staff and trainees

2.3 Respect the opinions of fellow consultants and referring physicians in the management of patients and be willing to accept differences of opinion

2.4 Demonstrate understanding of the following professional skills in time management: recognition that the effective use of time depends on punctuality, planning, speed and accuracy in clinical / laboratory work

2.5. Demonstrate the ability to recognize and respond appropriately to abuse, gender bias, discrimination, intimidation, and disrespect.

REFERENCE

[1] Frank, JR. (Ed). 2005. The CanMEDS 2005 physician competency framework. Better standards. Better physicians. Better care. Ottawa: The Royal College of Physicians and Surgeons of Canada.

Canadian College *of* Medical Geneticists
Collège canadien *de* généticiens médicaux

CCMG General Knowledge Training Guidelines

Topic	Objectives	Specific knowledge and skills ***By the end of training, all Trainees will be able to:***
1. Human genome structure and heredity	Understand general concepts related to the human genome and inheritance of DNA	a. Describe the information content of the human genome and the elements that predispose to mutation b. Describe the structure of DNA, how it is replicated and maintained (DNA repair mechanisms) c. Describe the chromosomal structure of the human genome d. Describe stages of cell division for mitosis and meiosis e. Describe the medical relevance of mitosis and meiosis f. Describe human gametogenesis and fertilization and the transmission of genomic material
2. Human gene structure and function	Understand general principles of human genetics at the gene level	a. Explain the organization and structure of genes (exons, introns, promoter regions, enhancers, silencers, etc.) b. Explain basic gene expression: transcription through to translation c. Explain gene regulation including transcription, splicing, variation of gene expression between tissues and relevance to medicine and the role of non-coding RNAs d. Explain post-transcriptional mechanisms including post-translational modifications

3. Mendelian single gene inheritance	Understand general concepts of single gene disorders and factors influencing these disorders	a. Describe Mendel's laws of inheritance b. Describe basic principles of Mendelian inheritance c. Understand concepts of penetrance, expressivity, anticipation, hypomorphic alleles and pseudodeficiency d. Explain how epigenetic factors influence phenotype e. X-linked inheritance : describe the effect skewed X-inactivation may have on clinical phenotype in females f. Demonstrate ability to analyze pedigrees for inheritance patterns g. Give examples of conditions where genotype correlates with phenotypic severity
4. Molecular genetics concepts and testing methods	Understand general principles of molecular biology as applied to human health	a. Understand the basic principles of the polymerase chain reaction b. Understand the concepts of nucleic acid sequencing, including Sanger and massively parallel sequencing c. Understand the concepts of targeted assays versus scanning methods d. Understand the basic principles of nucleic acid hybridization assays (e.g. Southern blot, Northern blot) e. Understand the limitations associated with molecular methods (allele drop-out, primer polymorphisms, large deletions etc.) f. Describe the concept of sample identity testing and use as an adjunct method to establish relationship between samples (i.e. maternal cell contamination, sample identity matching) g. Describe mutations using appropriate nomenclature (e.g. HGVS)
5. Significance of gene mutations	Understand general concepts of pathogenicity of genetic variation	a. Describe different classes of gene mutations (missense, nonsense, frameshift, splicing) and their effect on transcription, translation and protein function b. Ascribe clinical significance to different types of gene variants
6. Non-mendelian inheritance	Understand principles of non-Mendelian inheritance	a. Describe common non-Mendelian inheritance and their etiologies, including uniparental disomy, imprinting, mosaicism, unstable triplet repeats, pseudoautosomal inheritance, etc.

		b. Describe implications of non-Mendelian inheritance on genetic diagnostic testing c. Describe implications of non-Mendelian inheritance for clinical genetics including pedigree analysis and counselling
7. Mitochondrial inheritance	Understand principles of mitochondrial inheritance	a. Describe the structure and inheritance of the mitochondrial genome and gene expression b. Understand the basis for clinical heterogeneity in mitochondrial DNA defects c. Describe the role of nuclear and mitochondrial genes in mitochondrial disease d. Describe general features of mitochondrial disorders (multisystemic etc.) e. Recognize maternal inheritance from pedigree information.
8. Pharmacogenomics	Understand principles of pharmacogenomics	a. Describe the concept of drug responsiveness risks and benefits based upon genotype
9. Cytogenetic concepts and tests	Understand general principles of human cytogenetics as applied to human health	a. Describe general principles of cytogenetic methods for chromosome analysis, including karyotyping, genomic copy number assessment and spectral karyotyping. b. Describe appropriate indications for cytogenetic testing c. Distinguish between common cytogenetic variants and pathogenic rearrangements d. Describe general concepts of autosomal and sex chromosomal abnormalities (aneuploidy, translocations, etc.) e. Describe the meiotic segregation of rearranged chromosomes and the effects of recombination events. f. Describe general concepts of sex chromosomal abnormalities (aneuploidy, translocations etc.) g. Describe parent of origin effects and relevance to chromosomal abnormalities h. Describe etiology of chromosome abnormalities (non-disjunction, breakage and repair, non-homologous recombination, uniparental disomy) i. Understand uses and limitations of cytogenetic tests including the limits of

		detection of mosaicism j. Understand the effect of mosaicism on phenotype k. Understand use of appropriate nomenclature (e.g. ISCN)
10. Genomic microarray and copy number analysis	Understand principles and techniques associated with change in copy number analysis	a. Describe the appropriate indications for copy number analysis b. Describe the different techniques that can be used to detect copy number changes including array (CGH, SNP), FISH, qPCR, MLPA, whole genome sequencing; describe the limitations of each including the types of mutations detected c. Understand and describe when additional studies are required to complement or confirm microarray results d. Describe the basic principles used to ascribe clinical significance to copy number changes
11. Cancer genetics	Understand general principles of human cancer cytogenetics and molecular pathology	a. Explain concepts of multistep pathogenesis of cancers including inherited predisposition, oncogene activation, tumor suppressor inactivation, alteration of cell cycle control and DNA repair genes b. Explain and contrast inherited versus somatic mutations c. Describe methods to detect gene expression d. Describe principles of recurrent rearrangement detection using molecular or cytogenetic methods e. Describe the relevance of cytogenetic and molecular analysis to cancer diagnosis, prognosis and monitoring
12. Biochemical genetics	Understand broad categories of Inborn errors of metabolism	a. Describe the structure and functional relationships of intracellular components: nucleus, Golgi, endoplasmic reticulum, mitochondria, lysosomes, peroxisomes b. Describe the different categories of proteins in a cell (structural, enzymes, transport, receptor proteins etc.), their modes of action and means of regulation c. Describe biochemical consequences of a primary enzyme block in a metabolic pathway and the way clinical and pathological signs may be produced. d. Describe the major categories of inborn errors of metabolism: amino acid

		disorders, urea cycle disorders, organic acid disorders, fatty acid oxidation defects, lysosomal storage disorders, mitochondrial disease and peroxisomal disorders e. Understand principles of newborn screening f. Understand concept of pseudo-deficiency (ie. lysosomal disorders) g. Understand the deleterious effects of toxic metabolites on the fetus (e.g., maternal PKU)
13. Complex disorders	Understand the genetic contribution to complex human disease	a. Describe qualitative and quantitative traits; provide examples b. Describe the effect of genetic and environmental modifiers on single-gene disorders c. Define the concepts of multifactorial inheritance including liability model, threshold effects, epistasis, heritability and concordance d. Describe evidence for a genetic contribution to complex traits and common disorders e. Contrast the relative recurrence risks for multifactorial inheritance with single gene disorders and factors that affect risk (such as degree of relationship, sex, severity)
14. Population genetics	Understand concepts of human population genetics	a. Describe key concepts of human genetic variation in populations, including the role of ethnicity and population isolates in human variation b. Describe the Hardy-Weinberg equilibrium c. Demonstrate ability to use the Hardy-Weinberg equilibrium to assess genetic risk d. Understand concepts of population screening and when appropriate to offer screening
15. Genetic counseling	Understand key concepts in genetic counseling and risk assessment	a. Describe common indications for genetic counseling b. Describe the purpose of genetic counseling in specific scenarios c. Describe concepts of counseling: non-directive, awareness of values and biases

16. Risk assessment and calculations		a. Basic Bayesian analysis: demonstrate ability to modify *a priori* risk by one conditional factor b. Calculation of Odds ratios c. Understand basic test performance characteristics: sensitivity, specificity, positive predictive and negative predictive value
17. Developmental genetics and birth defects	Understand key steps in human development	a. Understand key concepts in developmental biology as it relates to normal and abnormal human morphogenesis b. Understand the concepts of morphogenesis, differentiation, pluripotency, specification, determination, embryonic induction, competency, and signal transduction. c. Describe the processes involved in early embryogenesis: fertilization to gastrulation d. Describe the major embryonic cell lineages and the distribution in the fetus and extra-embryonic tissues
18. Prenatal diagnosis	Understand principles of prenatal screening, prenatal diagnosis and related methodologies	a. Articulate the principles of a prenatal screening program b. Differentiate prenatal screening from prenatal diagnosis c. Describe the advantages, disadvantages and limitations associated with prenatal karyotyping, and prenatal rapid aneuploidy detection (RAD) by qfPCR or iFISH d. Describe the advantages and disadvantages of amniocentesis and chorionic villus sampling. e. Describe the risks, benefits, limitations and controversies surrounding the use of emerging technologies such as: • Non-invasive prenatal testing • array CGH on a prenatal sample • exome sequencing on a prenatal sample f. Describe genetic factors that contribute to recurrent pregnancy loss g. Understand the impact of teratogen exposure (e.g. infection, alcohol, medications) and maternal disease (eg. maternal PKU) on fetal development

19. Clinical Genetics	Understand broad categories of genetic conditions and their methods of assessment.	a. Describe methods of assessment of phenotypic variations, syndrome identification and diagnosis, including generally accessible computer diagnostic aids (e.g. OMIM) b. Understand the concept of syndrome and be able to give examples of syndromes associated with the following clinical manifestations: • dysmorphology • Cancer • Neurogenetic conditions • Cardiac genetic conditions • Imprinting disorders • Inborn errors of metabolism (major categories) • Chromosomal syndromes and genomic disorders characterized by recurrent microdeletions and microduplications c. Describe and understand the distinction between genetic screening and genetic testing d. Describe and understand the distinction between genetic testing for the purpose of diagnosis and predictive testing to assess risk for predisposition to monogenic or complex genetic diseases as well as their applications and limitations.
20. Ethics		a. Describe privacy and confidentiality principles as it relates to general practice (e.g. communication with health care providers, reporting, database searches), b. Describe ethical issues that relate to genetic testing in childhood c. Describe informed consent and its role in genetic testing d. Describe issues that relate to consenting for genomic analysis e. Explain incidental finding and provide examples from common tests (not large scale NGS-based sequencing) in which an incidental finding may be uncovered f. Outline the implications related to reporting and/or not reporting incidental findings and secondary findings (actively searching for disease-related pathogenic mutations not related to a patient's indication for testing) g. Describe ways to reduce the risk of incidental findings

		h. Describe the principles of biobanking

Recommended Texts and References:

Thompson and Thompson: Genetics in medicine 7th Edition
Strachan and Read: Human Molecular Genetics
Leonard, Debra: Molecular Pathology in Clinical Practice
Firth, Helen and Hurst, Jane: Oxford Desk Reference Clinical Genetics
Milunsky and Milunsky: Genetic Disorders and the Fetus
Moore, Persaud: The Developing Human
Emery and Rimoin: Essential Medical Genetics
Gardner: Chromosome abnormalities and Genetic Counseling.