الهيئة السعودية للتخصصات الصحية
Saudi Commission for Health Specialties

Medical Genetics and Genomic

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2021


بسم الله الرحمن الرحيم

PREFACE

- The Saudi Commission for Health Specialties (SCFHS) is the national regulatory body of postgraduate training programs across all health professions in Saudi Arabia.
- The primary goal of this document is to enrich the training experience of postgraduate trainees by outlining the learning objectives to become independent and competent future practitioners.
- This curriculum may contain sections outlining certain regulations of training; however, such regulations must be sought from the "General Bylaws" and "Executive Policies" for training published by the Saudi Commission for Health Specialties (SCFHS), which can be accessed online through the official SCFHS website. In case of a discrepancy in regulation statements, the statement in the most recent bylaws and executive policies will be the reference to apply.
- As this curriculum is subjected to periodic refinements, please refer to the electronic version posted online for the most updated edition at www.scfhs.org.sa.
- For any further support please do not hesitate to contact us at: Curricula@scfhs.org.sa

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II. COPYRIGHT STATEMENTS

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We would also like to acknowledge that the CanMEDS framework is a copyright of the Royal College of Physicians and Surgeons of Canada, and many of the description's competencies have been acquired from their resources (Please refer to: CanMEDS 2015 physician competency framework; Frank JR, Snell L, Sherbino J, editors. CanMEDS 2015 Physician Competency Framework. Ottawa: Royal College of Physicians and Surgeons of Canada; 2015.).

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ISBN: 978-603-8408-62-9

III. FOREWORD

As you start reading, you may wonder how this curriculum came about. Through an ambitious joint effort, the Committee for the Clinical Genetics and Metabolic Disorders Fellowship and the Curriculum Development Unit at the Saudi Commission for Health Specialties (SCFHS) have collaborated to develop this comprehensive curriculum, in which you will learn about the ins and outs of this fellowship program offered at the SCFHS. The curriculum is focused on enlightening the trainee about what he/she needs to accomplish to obtain SCFHS certification after completing standardized training in an optimal milieu in medical genetics and genomics. This environment is designed for basic teaching and comprehensive training and also stresses round clinical experience.

This competency-based curriculum with explicit representation of learning domains (knowledge, skills, and attitude) will ensure that completion of the 3-year fellowship in this well-structured program will train you to provide the best clinical care for both children and adult patients with a wide array of clinical genetic and metabolic diseases by acquiring solid knowledge of the basic concepts of the specialty, understanding the complexities of genetic diagnosis and management through well-organized inpatient and outpatient clinical training, gaining pertinent genetic laboratory experience, and acquiring the basic principles and participating in medical genetic and genomic research.

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V. INTRODUCTION

1. Context of Practice

While medical innovations have progressed dramatically over the past century, over the last 20 years, there has not been a field of medicine that has seen an expansion of knowledge, development of new technologies, and adjunct use of computer science as tremendous as the field of medical genetics (1). Such expansion has brought much excitement and a completely new approach to the diagnosis, management, and treatment of a great number of genetic disorders. Genetic factors that play a significant role in many diseases are being increasingly recognized, even in diseases that are thought to be unrelated or only remotely related to genetic influences, such as infectious diseases, cancer, and adult-onset disorders. The Human Genome Project has already brought exciting new discoveries. It also raises many concerns and questions regarding the ethics of medical genetics practice. (2)

The Kingdom of Saudi Arabia (KSA) is one of the largest countries in the Middle East, with a population of over 32 million, and an overall rate of consanguinity exceeding 50% , of which at least half are considered first-cousin marriages (3). Along with the large family size, the high consanguinity rate has a deleterious impact on health by increasing the incidence and prevalence of autosomal recessive genetic disorders in this region, posing enormous challenges in the diagnosis and management of these patients (4). Due to the tribal founder effect, many monogenic disorders have high prevalence within certain tribes in certain regions of the country, e.g., propionic academia, very long chain acyl-CoA dehydrogenase deficiency (VLCAD), and HMG-COA lyase deficiency. Additionally, some genetic disorders, are prevalent all throughout the Kingdom. In addition, many novel genetic diseases have been described for the first time in the

Kingdom (5). As a result, there is an increasing need for well-organized and comprehensive medical genetic services across the country as well as accredited training programs to graduate clinical specialists in medical genetics and genomics who will provide care for these patients.

2030 Vision was adopted as a methodology and roadmap for economic and developmental action in the KSA. It sought to identify the general directions, policies, goals, and objectives of the Kingdom, where investment in higher education, healthcare, and scientific research has enabled the KSA to take large strides in all these areas. Over the past decades, there has been a remarkable upsurge of interest in studying genetic disorders in Saudi Arabia addressing the enormous burden of genetic diseases. As a result, the SCFHS believes that there is a great need for a fellowship training program in this field that accords with the current standard for genetics fellowship in leading centers in the world. The Saudi Genetics Fellowship Training Program began in the 1990s. Several healthcare centers providing leading comprehensive genetic services in the country have been granted accreditation for the fellowship program in the field of clinical genetics and metabolic disorders by the SCFHS. The training centers conducted the program under the supervision of the SCFHS in the form of an agreement program. The Saudi Genetics Fellowship Training Program relies on qualified personnel, well-equipped resources, and cutting-edge technologies to conduct and deliver state-of-the-art training.

Upon completion of this 3-year-program, the trainee must have the skills and knowledge to practice independently in order to accommodate the vast expansion in the field of monogenic and complex polygenic diseases, metabolic disorders, neurodevelopmental disorders, and specialized training in many areas of medical genetics, such as adult genetics, skeletal dysplasia, cancer genetics, and advanced molecular technologies, in addition to building leadership, research, and supervisory skills. Fellowship training primarily covers the basics of clinical genetics with an emphasis on the molecular, biochemical, and cytogenetic background of disease,

diagnosis and management of genetic disorders, and advanced treatment modalities, including enzyme replacement therapy, organ transplantation stem cell transplantation, and future therapies including targeted and gene therapy. Additionally, the availability of preventive interventions via prenatal diagnosis, preimplantation genetic diagnosis, premarital and preconception screening, and carrier screening are important segments of training(6).

1. Liu G, David BT, Trawczynski M, Fessler RG. Advances in pluripotent stem cells: History, mechanisms, technologies, and applications. Stem Cell Rev Rep. 2020;16(1):3-32.

2. Green ED, Watson JD, Collins FS. Human Genome Project: Twenty-five years of big biology. Nature. 2015;526(7571):29-31.

3. El-Mouzan MI, Al-Salloum AA, Al-Herbish AS, Qurachi MM, Al-Omar AA. Regional variations in the prevalence of consanguinity in Saudi Arabia. Saudi Med J. 2007;28(12):1881-4.

4. Bittles A. Consanguinity and its relevance to clinical genetics. Clin Genet. 2001;60(2):89-98.

5. Al-Owain M, Al-Zaidan H, Al-Hassnan Z. Map of autosomal recessive genetic disorders in Saudi Arabia: concepts and future directions. Am J Med Genet A. 2012;158A(10):2629-40.

6. Alsulaiman A, Hewison J. Attitudes to prenatal and preimplantation diagnosis in Saudi parents at genetic risk. Prenat Diagn. 2006;26(11):1010-4.

2. Goal and Responsibility of Curriculum Implementation

The ultimate goal of this curriculum is to guide trainees to become *competent* in their specialty. This goal requires a significant amount of effort and coordination on the part of all stakeholders involved in postgraduate training. As "*adult learners*," trainees must demonstrate full engagement with *proactive* roles through: careful understanding of learning objectives, self-directed learning, problem solving,

openness and readiness to apply what they have learned through reflective practice based on feedback and formative assessment, and self-wellbeing and seeking support when needed. The program director has a vital role in making the implementation of this curriculum the most successful. Training committee members, particularly program administrators and chief fellows, have a significant impact on program implementation. Trainees should be able to share responsibility in curriculum implementation. The Saudi Commission for Health Specialties (SCFHS) applies the best models of training governance to achieve the best quality of training. Academic affairs in training centers and regional supervisory training committees will play a major role in training supervision and implementation. The Clinical Genetic & Metabolic Disorders Fellowship Supervisory Committee will be responsible for ensuring that the content of this curriculum is constantly updated to reflect the best-accepted standards in postgraduate education in this specialty.

3. What is New in This Edition?

In this new edition incorporates revisions related to:

- Its transformation into a competency-based curriculum with an explicit representation of learning domains (knowledge, skills, and attitude).
- Changes in rotations, reflecting minor modifications in the rotations during the 2nd and 3rd years.

VI. ABBREVIATIONS USED IN THIS DOCUMENT

Abbreviation	Description
SCFHS	Saudi Commission for Health Specialties
F(1)	(First) Year of Fellowship
PT	Progress test
OSCE	Objective Structured Clinical Examination
CBE	Competency-Based Education
ITER	In-Training Evaluation Report
COT	Consultation Observation Tool
FTSC	Fellowship Training Supervisory Committee
NGS	Next Generation Sequencing
MLPA	Multiplex Ligation-Dependent Probe Amplification
PCR	Polymerase Chain Reaction
IEM	Inborn Error of Metabolism
PGD	Pre-Implantation Genetic Diagnosis
VCF	Variant Call Format
WES	Whole Exome Sequencing
WGS	Whole genome sequencing

VII. PROGRAM ENTRY REQUIREMENTS

Eligibility

Please refer to the updated executive policy of SCFHS on admission and registration.

Website: www.scfhs.org.sa

1. Introduction to Learning Outcomes and Competency-Based Education

Training should be guided by well-defined "learning objectives" that are driven by targeted "learning outcomes" of a particular program to serve specific specialty needs. Learning outcomes are supposed to reflect the professional "competencies" and tasks that are to be "entrusted" to trainees upon graduation. This will ensure that graduates meet the expected demands of the healthcare system and patient care with respect to their particular specialty. Competency-based education (CBE) is an approach of "adult-learning" that is based on achieving pre-defined, fine-grained, and well-paced learning objectives that are driven by complex professional competencies.

Professional competencies related to healthcare are usually complex and pertain to a mixture of multiple learning domains (knowledge, skills, and attitude). CBE is expected to change the traditional way of postgraduate education. For instance, the time of training, though a precious resource, should not be considered a proxy for *competence* (e.g., time of rotation in certain hospital areas is not the primary marker of competence achievement). Furthermore, CBE emphasizes the critical role of informed judgment of learners' competency progress, which is based on a staged and formative assessment driven by multiple workplace-based observations. Several CBE models have been developed for postgraduate education in healthcare (example: CanMEDs by the Royal College of Physician and Surgeon of Canada (RCPSC), the CBME-Competency model by the Accreditation Council for Graduate Medical Education (ACGME), Tomorrow's Doctor in the UK, and multiple others). The following are concepts that enhance the implementation of CBE in this curriculum:

- **Competenc:** Competency is a cognitive construct that assesses the potential to perform efficiently in a given situation based on the standard of the profession. Professional roles (e.g., experts, advocates, communicators, leaders, scholars, collaborators, and professionals) are used to define competency roles in order to facilitate learning and assessment.
- **Milestones:** Milestones are stages of the developmental journey throughout the competency continuum. Trainees throughout their learning journey, from junior and throughout senior levels, will be assisted in transforming from (novice/supervised) to (master/unsupervised) practitioners. This should not undermine the role of supervisory/regulatory bodies regarding the malpractice of

independent practitioners. Milestones are expected to enhance the learning process by pacing training/assessment to match the developmental level of trainees (junior vs. senior).

- **Learning-Domains:** Whenever possible, efforts should be directed to annotate the learning outcomes with the corresponding domain (K = Knowledge, S = Skills, and A = Attitude). More than one annotation might be needed for a given learning outcome.
- **Content-Area Categorization:** It is advisable to categorize learning outcomes in broad content areas related to the practice of profession. For example, diagnostic versus therapeutic, simple versus complex, urgent versus chronic, etc.
- **Trainees** are expected to progress from the novice to the mastery level in a certain set of professional competencies. SCFHS endorsed CanMEDs for articulating professional competencies. This curriculum applies the principles of competency-based medical education. CanMEDsCanMeds/ACGME/OTHER represents a globally accepted framework that outlines competency roles. The CanMEDs 2015/ACGME 2018 framework was adopted in this section.[1]

2. Program Duration

The duration of this program is three years, as approved by the decree of the Executive Council of Training and Education.

[1] ** (Frank JR, Snell L, Sherbino J, editors. CanMEDS 2015 physician competency framework Ottawa: Royal College of Physicians and Surgeons of Canada 2015)*

3. Program Rotations (Months)

Training Year	Mandatory core rotations*		Elective rotations**	
	Rotation name	Duration	Rotation name	Duration
F1	- Clinical Service - Clinical Nutrition - Genetic Counseling - Leave	9 Months 1 Month 1 Month 1 Month	None	
F2	- Clinical Service - Biochemical Laboratory - Cytogenetic Laboratory - Molecular Laboratory - Leave	7 Months 1 Month 1 Month 1 Month 1 Month	- Elective *(e.g., Adult metabolic developmental, radiology, adult/ pediatric neurology, laboratory, mental health)*	1 Month
F3	- Clinical Service - Next Generation Seq - Research I - Research II - Prenatal Genetics - Leave	6 Months 1 Month 1 Month 1 Month 1 Month 1 Month	- Elective *(e.g., Adult metabolic developmental, radiology, adult/ pediatric neurology, laboratory, mental health)*	1 Month

*(*Mandatory core rotation: Set of rotations that represent program core components and are mandatory.*

***Elective rotation: Set of rotations that are related to the specialty, as determined by the scientific council/committee, and the trainee is required to do some of them.*

4. Mapping of Learning Objectives and Competency Roles to Program Rotations

This section aims to match the competencies and objectives of each rotation. Trainees and trainers should work together to achieve these objectives during teaching and formative assessments. Expectations should evolve as the training level progresses (training stage, milestones).

Rotation Name: Clinical Service Rotation

<table>
<tr><th>Rotation Setting</th><th>Training stage</th><th>Rotation duration</th><th>Competency roles**</th></tr>
<tr><td rowspan="2">Clinical Service</td><td>F1</td><td>9 months</td><td rowspan="2">Key Competencies
By the end of training, Clinical Biochemical Genetics trainees will be able to:

MEDICAL EXPERT
1. Recognize basic concepts of cell biology, molecular biology and biochemistry, and human genetics;
2. Describe normal physiology throughout development including prematurity and changes during childhood, ageing, and pregnancy;
3. Determine and recognize the common clinical phenotypes usually seen in genetic and metabolic disorders;
4. Recognize, whenever possible, the underlying biological/molecular pathway of the disease;
5. Use diagnostic facilities and therapeutic modalities for optimal and effective patient care, and access and relate relevant information to clinical practice;
6. Show the ability to examine, interpret, and understand laboratory data and results relevant to the investigation, and understand their clinical importance.
7. Maintain medical records that are consistent, up to date, informative, and legible.

COMMUNICATOR
1. Efficiently elicit and understand health and other information communicated by patients and their families;
2. Acquire relevant history from patients and families and their communities;
3. Efficiently and empathetically communicate health information (e.g., diagnosis, prognosis, treatment decisions, end of life decisions, genetic issues) to patients and their families;
4. Confer with the team and other relevant healthcare providers to provide optimal patient care;
5. Understand the psychosocial impact of the genetic condition on the patient and their family and take it into consideration when communicating with them;
6. Learn how to convey “bad” news to the patient and their families with empathy and respect.</td></tr>
<tr><td>F2</td><td>7 months</td></tr>
</table>

	F3	7 months	**COLLABORATOR** 1. Contribute efficiently as a team member with relevant healthcare providers in collaborative decision making for metabolic cases; 2. Contribute efficiently to other interdisciplinary team activities. **LEADER** 1. Work efficiently and professionally in a healthcare organization; 2. Apprehend and apply the important elements of the Quality Management system within the clinic and/or laboratory; 3. Apply healthcare resources efficiently to balance patient care, learning needs, and outside activities; 4. Manage time efficiently and highlight required activities. **HEALTH ADVOCATE** 1. Advocate for suitable clinical services for patients with genetic and metabolism disorders with the introduction of new programs, new technologies, and new treatments; 2. Identify and respond to those issues where advocacy is applicable. **SCHOLAR** 1. Undertake constant learning activities to sustain and advance professional knowledge; 2. Assist the learning of other healthcare professionals, students, laboratory colleagues, the public, and others about the practice of clinical biochemical genetics; 3. Contribute to the growth of new knowledge and the progression of the field of medical genetics. **PROFESSIONAL** 1. Exercise medicine with highest care with integrity, honesty, and compassion; 2. Show suitable personal and interpersonal professional behaviors.

Rotation Name: Molecular Laboratory Rotation

Rotation Setting	Training stage	Rotation duration	Competency roles**
Molecular Laboratory rotations	F2	1 month	Key Competencies By the end of training, Molecular Laboratory trainees will be able to: **MEDICAL EXPERT** 1. Recognize basic concepts in clinical molecular biology and medical genetics and genomics, e.g., how to read a mutation and how to differentiate pathogenic, benign, and variants of unknown significance; 2. Describe the pathobiology of human genetic disorders and their molecular genetic causes; 3. Show the ability to implement the basic molecular genetic testing; e.g., Polymerase Chain Reaction (PCR), Sanger sequencing, Multiplex Ligation-dependent Probe Amplification (MLPA); 4. Relay molecular genetic testing for human inherited disease to other molecular diagnostic testing applications; 5. Define the standard and advanced molecular biology techniques. **COMMUNICATOR** 1. Arrange for consultation on molecular diagnostic cases with healthcare providers, laboratory staff, patients, and others; 2. Incorporate clinical and laboratory information with the outcome of interpretation and decision making for suitable molecular genetic test utilization to support diagnosis; 3. Report the outcomes and interpretation of molecular diagnostic testing to applicable individuals. **COLLABORATOR** 1. Contribute efficiently as a team member with related healthcare providers in collaborative decision making for molecular diagnostic cases; 2. Facilitate decision making in inter-professional teams; 3. Participate efficiently in other interdisciplinary team activities. **LEADER** 1. Apprehend and apply the important elements of the Quality Management system within the laboratory; 2. Apply molecular genetic testing resources efficiently; 3. Direct staff, equipment, and sample resources to work efficiently and professionally in a healthcare organization;

			4. Manage time efficiently and highlight required activities. **HEALTH ADVOCATE** 1. Define particular public health practices or policies that influence the provision of molecular genetic testing services; 2. Identify the health necessities of individuals, communities, and populations served by molecular diagnostic testing. **SCHOLAR** 1. Undertake constant learning activities to sustain and advance the healthcare professional knowledge; 2. Assist the learning of other healthcare professionals, students, laboratory staff, the public, and others about molecular diagnostic testing; 3. Lead research projects and publish findings for the advancement of knowledge. **PROFESSIONAL** 1. Determine ethical practices and a sense of accountability in molecular diagnostic testing; 2. Show the personal and interpersonal professional behaviors expected of the molecular genetics diagnostician.

Rotation Name: Next Generation Sequencing (NGS) Laboratory Rotation

Rotation Setting	Training stage	Rotation duration	Competency roles**
Next Generation Sequencing rotation	F3	1 month	Key Competencies By the end of training, Next Generation Sequencing Laboratory trainees will be able to: MEDICAL EXPERT 1. Recognize basic concepts in human molecular biology and advanced medical genetics and genomic testing, e.g., how to read a mutation and how to differentiate pathogenic, benign, and variants of unknown significance; 2. Describe the pathobiology of human genetic disorders and their molecular genetic causes. Know the limits of the NGS; 3. Show the ability to implement effective molecular NGS, e.g., pipelines, variant call format (VCF) file review; 4. Relay molecular advanced testing for human inherited disease to other NGS applications such as bioinformatics; 5. Define the standard and advanced human genomic data bases. COMMUNICATOR 1. Arrange for consultation on NGS diagnostic cases with healthcare providers, laboratory staff, patients, and others; 2. Incorporate clinical and laboratory information to support the selection of the most suitable molecular NGS test. 3. Report the outcomes and interpretation of NGS diagnostic testing to relevant individuals. COLLABORATOR 1. Contribute efficiently as a team member with related healthcare providers in collaborative decision making for molecular diagnostic cases; 2. Facilitate decision making in inter-professional teams; 3. Participate efficiently in other interdisciplinary team activities. LEADER 1. Comprehend and apply important elements of the Quality Management system within the laboratory; 2. Use molecular genetic testing resources efficiently;

			3. Direct staff, equipment, and sample resources to work efficiently and professionally in a healthcare organization; 4. Manage time efficiently and highlight required activities. **HEALTH ADVOCATE** 1. Define particular public health practices or policies that affect the provision of molecular genetic testing services; 2. Identify the health necessities of individuals, communities, and populations served by molecular diagnostic testing. **SCHOLAR** 1. Undertake constant learning activities to sustain and advance their professional healthcare knowledge; 2. Assist the learning of other healthcare professionals, students, laboratory staff, the public, and others about molecular diagnostic testing; 3. Lead research projects and publish findings for the advancement of knowledge. **PROFESSIONAL** 1. Determine ethical practices and a sense of accountability in molecular diagnostic testing; 3. Show the personal and interpersonal professional behaviors expected of the molecular genetics diagnostician.

Rotation Name: Cytogenetics Laboratory Rotation

Rotation Setting	Training stage	Rotation duration	Competency roles**
Cytogenetic Laboratory rotations	F2	1 month	Key Competencies By the end of training, Cytogenetics trainees will demonstrate the ability to: **MEDICAL EXPERT** 1. Recognize basic concepts in human cytogenetics, molecular biology and medical genetics and genomics, including chromosomal analysis, FISH, and SNP array; 2. Describe the pathobiology of human genetic disorders and their cytogenetic causes; 3. Relay cytogenetic testing for acquired disorders to other cytogenetic testing applications, including cancer cytogenetics; 4. Demonstrate knowledge of standard and advanced cytogenetic techniques; 5. Demonstrate the skill to implement effective cytogenetic testing. **COMMUNICATOR** 1 Arrange for consultation for cytogenetic cases to healthcare providers, laboratory staff, patients, and others; 2. Incorporate clinical and laboratory information on the outcome and its interpretation for appropriate decision making for suitable cytogenetic test utilization; 3. Report the outcomes and interpretation of cytogenetic testing to relevant individuals. **COLLABORATOR** 1. Contribute efficiently as a team member with related healthcare providers in collaborative decision making for cytogenetic cases; 2. Facilitate decision making in inter-professional teams; 3. Contribute efficiently to other interdisciplinary team activities. **LEADER** 1. Comprehend and apply the important elements of the Quality Management system within the laboratory; 2. Use cytogenetic testing resources effectively;

			3. Direct staff and equipment to work efficiently and professionally in a healthcare organization; 4. Manage time efficiently and highlight required activities. **HEALTH ADVOCATE** 1. Define particular public health practices or policies that affect the provision of cytogenetic testing services; 2. Identify the healthcare necessities of individuals, communities, and populations served by cytogenetic testing. **SCHOLAR** 1. Conduct constant learning activities to sustain and advance professional knowledge; 2. Assist the learning of other healthcare professionals, students, laboratory staff, the public, and others about cytogenetic testing; 3. Lead research projects and publish findings for the advancement of knowledge. **PROFESSIONAL** 1. Practice ethically and display a sense of accountability in cytogenetic testing; 2. Show the personal and interpersonal professional behaviors expected of the clinical cytogenetic diagnostician.

Rotation Name: Biochemical Genetics Laboratory Rotation

Rotation Setting	Training stage	Rotation duration	Competency roles**
Biochemical Genetics Laboratory rotations	F2	1 month	Key Competencies By the end of training, Laboratory Biochemical Genetics trainees will demonstrate the ability to: **MEDICAL EXPERT** 1. Recognize basic concepts in cell biology, human biochemistry, and medical genetics and genomics, including Tandem metabolic testing, urine organic acid analysis, and plasma amino acid analysis; 2. Describe the pathobiology of metabolic disorders and their biochemical genetic causes and treatment; 3. Relay biochemical genetic testing for human inherited disease to other biochemical genetic testing applications; 4. Demonstrate knowledge of standard and advanced biochemical genetic techniques; 5. Demonstrate the skills to implement effective biochemical genetic testing. **COMMUNICATOR** 1. Arrange for consultation on biochemical genetic cases with healthcare providers, laboratory staff, patients, and others; 2. Incorporate clinical and laboratory information to support result interpretation and decision making for appropriate biochemical genetic test utilization; 3. Report the outcomes and interpretation of metabolic testing to relevant individuals. **COLLABORATOR** 1. Contribute efficiently as a team member with related healthcare providers in collaborative decision making for metabolic cases; 2. Facilitate decision making in inter-professional teams; 3. Contribute efficiently to other interdisciplinary team activities. **LEADER** 1. Comprehend and apply the important elements of the Quality Management system within the laboratory; 2. Use biochemical genetic testing resources effectively; 3. Direct staff and equipment to work effectively and efficiently in a healthcare organization; 4. Manage time efficiently and highlight required activities.

			HEALTH ADVOCATE 1. Define particular public health practices or policies that affect the provision of biochemical genetic testing services; 2. Identify the health needs of individuals, communities and populations served by biochemical genetic testing. **SCHOLAR** 1. Undertake constant learning activities to sustain and advance their professional healthcare knowledge; 2. Assist the learning of other healthcare professionals, students, laboratory colleagues, the public, and others about biochemical genetic testing; 3. Lead research projects and publish findings for the advancement of knowledge. **PROFESSIONAL** 1. Practice ethically and display a sense of accountability in biochemical genetic testing; 2. Show the personal and interpersonal professional behaviors expected of the clinical biochemical genetics diagnostician.

Rotation Name: Genetic Counseling Rotation

Rotation Setting	Training stage	Rotation duration	Competency roles**
Genetic Counseling Rotation	F1	1 month	Key Competencies By the end of training, Genetic Counseling trainees will demonstrate the ability to: **MEDICAL EXPERT** 1. Demonstrate an understanding of the genetic counseling relevant to molecular biology, biochemistry, and human genetics; 2. Describe common patterns of Mendelian vs. non-Mendelian inheritance (autosomal dominant and recessive, X-linked, multifactorial, and the effect of maternal and paternal age) and demonstrate the ability to construct a pedigree; 3. Demonstrate an understanding of genetic, teratologic, metabolic, endocrine, immunologic, and infectious disorders that relate to pregnancy; 4. Demonstrate expertise and ability in implementing effective standards and advanced genetic testing techniques; 5. Identify resources in the community for diagnosis, genetic counseling, therapy, and psychosocial support of children with genetic defects and congenital anomalies **COMMUNICATOR** 1. Provide consultation for genetic cases to healthcare providers, patients, and others; 2. Integrate clinical and laboratory information to assist with result interpretation and decision making for appropriate genetic counseling utilization; 3. Report the results and interpretation of genetic testing to relevant individuals, including the prognosis and treatment of the genetic disorder; 4. Display an awareness of the unique personal, psychosocial, cultural, and ethical issues that surround individual patients with genetic disorders; 5. Demonstrate the ability to counsel patients with regards to prevention methods. **COLLABORATOR** 1. Participate effectively as a team member with relevant healthcare providers in collaborative decision making for genetic cases; 2. Mediate decision making in inter-professional teams;

			3. Work closely in a team-based approach with members of the genetic team including geneticists, nurses, and social workers. **LEADER** 1. Understand and apply the essential elements of the Quality Management system within the division; 2. Use genetic testing resources effectively for segregation analysis during counseling sessions; 3. Manage staff and equipment to work effectively and efficiently in a healthcare organization; 4. Manage time effectively and prioritize required activities. **HEALTH ADVOCATE** 1. Describe specific public health practices or policies that affect the provision of genetic counseling services; 2. Respond appropriately to the health needs of individuals, communities, and populations served by genetic counseling services; 3. Participate effectively in local, regional, and national specialty associations (professional or scientific) to promote better public healthcare for genetic disorders. **SCHOLAR** 3. Undertake ongoing learning activities to maintain and advance their professional knowledge related to genetic counseling field; 4. Facilitate the learning of other healthcare professionals, students, laboratory colleagues, the public, and others regarding genetic counseling services; 5. Conduct research projects and publish findings for the advancement of knowledge. **PROFESSIONAL** 1. Complete genetic reports, letters, and summaries in a timely fashion and maintain medical records that are consistently accurate, informative, and legible; 2. Practice ethically and display a sense of responsibility in genetic counseling service.

Rotation Name: Prenatal and PGD Rotation

Rotation Setting	Training stage	Rotation duration	Competency roles**
Prenatal and PGD Rotation	F3	1 month	Key Competencies By the end of training, prenatal and PGD trainees will demonstrate the ability to: MEDICAL EXPERT 1. Demonstrate an understanding of the basic sciences relevant to maternal fetal medicine and how to apply these techniques for the prevention of genetic disease; 2. Demonstrate and understand knowledge of maternal, placental, fetal, and newborn anatomy, embryology, genetics, pharmacology, biochemistry, endocrinology, microbiology, physiology, and pathology; 3. Demonstrate an understanding of genetic, teratologic, metabolic, endocrine, immunologic, and infectious disorders that relate to pregnancy; 4. Demonstrate expertise and ability in implementing effective standard and advanced prenatal genetic techniques; 5. Demonstrate an understanding of indications and limitations of the different methods of prenatal diagnosis, both non-invasive and invasive, including CVS, amniocentesis, cordocentesis, maternal serum screening, non-invasive prenatal testing (NIPT), and microarray. COMMUNICATOR 1. Provide consultation on prenatal genetic cases with healthcare providers, laboratory staff, patients, and others; 2. Integrate clinical and laboratory information to assist with result interpretation and decision making for appropriate prenatal genetic test utilization; 3. Report the results and interpretation of genetic testing to relevant individuals; 4. Demonstrate an awareness of the unique personal, psychosocial, cultural, and ethical issues that surround individual patients with obstetric problems; 5. Demonstrate the ability to counsel patients regarding termination methods.

			COLLABORATOR 1. Participate effectively as a team member with relevant healthcare providers in collaborative decision-making for prenatal cases; 2. Mediate decision-making in inter-professional teams; 3. Work closely in a team-based approach with members of the Perinatal Clinic Team including Nurses, Sonographers, Genetic Counselors, and Social Workers. **LEADER** 1. Understand and apply the essential elements of the Quality Management system within the division; 2. Use biochemical genetic testing resources effectively; 3. Manage staff and equipment to work effectively and efficiently in a healthcare organization; 4. Manage time effectively and prioritize required activities. **HEALTH ADVOCATE** 1. Describe specific public health practices or policies that affect the provision of prenatal and PGD genetic testing services; 2. Respond responsibly to the health needs of individuals, communities, and populations served by prenatal and PGD genetic testing; 3. Participate effectively in local, regional, and national specialty associations (professional or scientific) to promote better healthcare for women (if applicable). **SCHOLAR** 1. Conduct ongoing learning activities to maintain and advance professional knowledge; 2. Facilitate the learning of other healthcare professionals, students, laboratory colleagues, the public, and others regarding prenatal genetic service; 3. Conduct research projects and publish findings for the advancement of knowledge. **PROFESSIONAL** 1. Complete reports, letters, and summaries in a timely fashion and maintain medical records that are consistently accurate, informative, and legible. 2. Practice ethically and display a sense of responsibility in prenatal and PGD genetic testing; 3. Show the personal and interpersonal professional behaviors expected of the clinical and laboratory prenatal and PGD genetic diagnostician.

Rotation Name: Clinical Nutrition Rotation

Rotation Setting	Training stage	Rotation duration	Competency roles**
Clinical Nutrition rotations	F1	1 month	Key Competencies By the end of training, Clinical Nutrition trainees will be able to: **MEDICAL EXPERT** 1. Explain general concepts in human nutrition, especially those pertinent to genetic and metabolic disorders; 2. Explain the normal physiology of digesting, including nutritional changes during childhood, in both healthy and sick individuals; 3. Demonstrate knowledge and skills in the nutritional management of inborn errors of metabolism (IEM); 4. Demonstrate the ability to examine, interpret, and understand laboratory data and results relevant to the nutrition of IEM, and understand their clinical significance. **COMMUNICATOR** 1. Effectively elicit and understand health and other information related to metabolic diet communicated by patients and their families; 2. Effectively communicate health information (e.g., types of formula, metabolic diet calculation, sick days formula) to patients and their families; 3. Consult with the metabolic team and other relevant healthcare providers to provide optimal patient nutritional care. **COLLABORATOR** 1. Participate effectively as a team member with relevant healthcare providers in collaborative decision making for metabolic cases; 2. Contribute effectively to other interdisciplinary team activities. **LEADER** 1. Understand and apply the essential elements of the Quality Management system within the clinical departments; 2. Manage staff, equipment, and sample resources to work effectively and efficiently in a healthcare organization; 3. Manage time effectively and prioritize required activities. **HEALTH ADVOCATE**

			1. Describe specific public health practices or policies that affect the provision of nutrition services; 2. Respond appropriately to the health needs of individuals, communities, and populations served by molecular diagnostic testing; 3. Advocate for appropriate clinical services for patients with inborn errors of metabolism, including the introduction of new programs, new technologies, and new diet treatments; 4. Recognize and respond to those issues where advocacy is appropriate. **SCHOLAR** 1. Undertake ongoing learning activities to maintain and advance their professional knowledge; 2. Facilitate the learning of other healthcare professionals, students, laboratory staff, the public, and others regarding IEM nutritional management; 3. Conduct research projects and publish findings for the advancement of knowledge. **PROFESSIONAL** 1. Practice ethically and display a sense of responsibility in practice; 2. Show suitable personal and interpersonal professional behaviors toward the clinical team.

REFERENCE: 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.

IX. CONTINUUM OF LEARNING

The clinical genetics and metabolic disorders fellowship training program extends over three years, during which the fellows are exposed to learning opportunities at different levels, including clinical, laboratory, metabolic nutrition, genetic counseling, and prenatal genetic services. The training program also provides research opportunities and promotes administrative and leadership skills. Different rotations are distributed over the three years of the program, including F1, F2, and F3 years, to provide a continuum of learning reflecting the competency roles of the rotation and the year of training.

X. TEACHING METHODS

The teaching process in postgraduate fellowship training programs is based mainly on the principles of "adult learning theory." The trainees must feel the importance of learning and play active roles in the content and process of their own learning. The training program implements the adult learning concept in each feature of the activities where the fellows are responsible for their own learning requirements. The training time includes the following three formal teaching activities:

- Program-Specific Learning Activities
- Universal Topics
- General Learning Opportunities

1.1. Program-Specific Learning Activities

Program-specific activities are educational activities that are specifically designed and intended for teaching trainees during their training time. The trainees are required to attend these activities, and non-compliance can subject trainees to disciplinary actions. Program administration should support these activities by providing protected time for trainees to attend these activities and allow them to participate in such activities.

A) Program Academic Half-Day (Mandatory)

Every week, at least 2 hours of formal training time (commonly referred to as the *academic half-day*) should be reserved. Formal teaching time is an activity that is planned in advance with an assigned tutor, time slots, and venue. Formal teaching time shall exclude bedside teaching, clinic postings, etc. The academic half-day covers the core clinical genetics and metabolism topics to ensure that important clinical aspects of the specialty are well taught. The academic day should include at least one

- Case presentation (One hour)
- Didactic lecture (One hour)

Since many genetic disorders are considered rare, “case presentation” represents an important tool for formal teaching, where illustrative cases with unique clinical, radiological, and laboratory findings are discussed along with their genetic testing results. Case presentations should occupy at least one hour of the academic day. Likewise, another hour at least of the academic half-day should be of the didactic lecture style. It is recommended that lectures be conducted in an interactive, case-based discussion format. The learning objectives of each core topic need to be clearly defined, and it is preferable to use pre-learning material. The core specialty topics are delivered through didactic lectures every week. It is recommended that the fellowship supervisory committees work together in coordination with academic and training affairs, program directors, and chief fellows to ensure the planning and implementation of academic activities, as indicated in the curriculum. There should be an active involvement of the trainee in the development and delivery of the topics under faculty supervision; the involvement might be in the form of delivery, content development, research, etc. The supervising educator should ensure that each topic is stratified into the three categories of the learning domain: knowledge, skill, and attitude. The recommended number of half-days conducted annually is 40 sessions per training academic year, with time reserved for other forms of teaching methods such as journal clubs and clinical/practical teaching. It is advised that the fellowship training committee, program directors, and chief fellows, in coordination with academic and training affairs and the fellowship supervisory committees, work together to ensure the planning and implementation of academic activities, as indicated in the curriculum. This should aim for the efficient use of available resources and to optimize the exchange of expertise.

Core topics to be discussed during academic half-day activity should include different areas in clinical genetics and metabolism, including but not limiting to:

- Basic human genetics;
- Principles of cytogenetic techniques;
- Principles of molecular genetics techniques;
- Biochemical genetics;
- Clinical genetics;
- Developmental genetics;
- Cancer genetics;
- Prenatal genetics;
- Genetic counseling;
- Adult genetics;
- Population genetics;
- Diagnosis and management of metabolic disorders;
- Metabolic emergencies, diagnosis, and management;
- Novel therapeutic approaches of genetic disorders;
- Ethics in medical genetics.

The following table illustrates relevant topics for the half-day activities over the course of 12 months, taking into consideration the level (F1, 2, and 3). The program, although not meant to be in the same chronological order as depicted in the table, should be designed for the three years of training to cover all topics listed above and to match different rotation goals and objectives. Topics are denoted I, II, and III to indicate the different lecture sessions within the same topic given in the first, second, and third years of training, respectively.

Academic week	Time	Didactic Topic/Session	Presenter	Moderator
1	13:00–13:05	Welcome to the program		
	13:05–14:00	Modes of inheritance I, II, III		
2	13:00–14:00	Basics of human genetics I, II, III		
3	13:00–14:00	Approach to the dysmorphic child I, II, III		
4	13:00–14:00	Basics of metabolic nutrition I, II, III		
5	13:00–14:00	Metabolic nutrition: disorder-based I, II, III		
6	13:00–14:00	Approach to a patient with metabolic emergencies, hyperammonemia I, II, III		
7	13:00–14:00	Approach to a patient with metabolic emergencies, metabolic acidosis I, II, III		
8	13:00–14:00	Interpretation of biochemical testing I, II, III		
9	13:00–14:00	Overview of carbohydrate metabolism disorders I, II, III		
10	13:00–14:00	Overview of amino acid metabolism disorders I, II, III		
11	13:00–14:00	Overview of energy disorders I, II, III		
12	13:00–14:00	Overview of organelle disorders (lysosomal, peroxisomal, others) I, II, III		
13	13:00–14:00	Overview of other metabolic disorders I, II, III		
14	13:00–14:00	General principles of treatment of metabolic disorders I, II, III		
15	13:00–14:00	Clinical cytogenetics I, II, III		
16	13:00–14:00	Cytogenetic techniques I, II, III		

17	13:00–14:00	Molecular genetics I, II, III		
18	13:00–14:00	Molecular genetic techniques I, II, III		
19	13:00–14:00	Introduction to NGS and bioinformatics I, II, III		
20	13:00–14:00	Basics of cancer genetics I, II, III		
21	13:00–14:00	Common cancer genetic disorders I, II, III		
22	13:00–14:00	General clinical genetics I, II, III		
23	13:00–14:00	Prenatal testing and reproductive genetics I, II, III		
24	13:00–14:00	Clinical metabolism I, II, III		
25	13:00–14:00	Genetic counselling I, II, III		
26	13:00–14:00	Skeletal dysplasia I, II, III		
27	13:00–14:00	Clinical genetics: dysmorphology I, II, III		
28	13:00–14:00	Clinical genetics: congenital defects I, II, III		
29	13:00–14:00	Population genetics and risk assessment I, II, III		
30	13:00–14:00	Neurodevelopmental genetics I, II, III		
31	13:00–14:00	Clinical genetics: dysmorphology, I, II, III		
32	13:00–14:00	Clinical genetics: monogenic disorders I, II, III		
33	13:00–14:00	Clinical genetics: imprinting disorders I, II, III		
34	13:00–14:00	Clinical genetics: adult disorders I, II, III		
35	13:00–14:00	Biochemical disorders: adult disorders I, II, III		

36	13:00–14:00	New diagnostic modalities in genetics I, II, III		
37	13:00–14:00	Clinical genetics: adult disorders I, II, III		
38	13:00–14:00	Biochemical disorders: adult disorders I, II, III		
39	13:00–14:00	Therapeutics in clinical genetics I, II, III		
40	13:00–14:00	Ethical issues and genetic counseling I, II, III		

B) Case Presentation:

A weekly case presentation constitutes a case-based teaching session. Fellows and consultant geneticists are encouraged to select interesting cases with relevant teaching points that are normally encountered during hospital consultations or outpatient services. Case presentation sessions should be interactive to help trainees learn the basic principles of human genetics, diagnostic approaches, and management issues in interesting and challenging cases.

1.2. Universal Topics

Universal topics are educational activities developed by SCFHS and are intended for all specialties. Priority will be given to topics as follows:

- High value,
- Interdisciplinary and integrated,
- Requiring expertise that might be beyond the availability of the local clinical training sites.

Universal topics have been developed and made available by SCFHS, such as e-learning via personalized access to the online modules for each trainee. Each universal topic has a self-assessment at the end of the module. As indicated in the Executive Policies of Continuous Assessment and Annual Promotion, universal topics are mandatory components of the criteria for the annual promotion of trainees from their current level of training to the subsequent level. Universal topics are distributed over the entire training period. The table below shows

the universal topic modules assigned in each training year/stage of the fellowship program.

Training Year	UT module	Subjects	Objectives
F1	Module 1	Medical Fundamentals (4 hours)	Blood transfusion Safe drug prescribing
	Module 2	Acute Care (4 hours)	Management of electrolyte imbalance
F2/F3	Module 3	Ethics and Healthcare (4 hours)	The goal of this learning is to develop the understanding of medical ethical principles and ability to apply these principles to a broad range of decision-making scenarios

1.3. General Learning Opportunities (Highly Recommended):

Formal training time should be supplemented by other practice-based learning (PBL), such as:

- Journal club (at least quarterly)
- Grand rounds (at least once per month)
- Continuous professional activities (CPD) relevant to specialty, such as conferences or workshops within the specialty (annually)
- Morbidity and Mortality (M&M; every 6 months)

N.B. Grand rounds are special presentation provided by experts in various specialties related to genetics.

XI. ASSESSMENT AND EVALUATION

1. Purpose of Assessment

Assessment plays a vital role in the success of postgraduate training. Assessment will guide trainees and trainers to achieve defined standards, learning outcomes, and competencies. In addition, the assessment will provide feedback to learners and faculty regarding curriculum development, teaching methods, and quality of the learning environment. A reliable and valid assessment is considered vital to ensure curriculum alignment between the objectives, learning methods, and assessment methods. Finally, regular assessments assure patients and the public that health professionals are safe and competent to practice.

Therefore, assessment can serve the following purposes:

a. **Assessment for learning**: As trainers will use information from trainees' performance to inform their learning for improvement, this enables educators to use information about trainees' knowledge, understanding, and skills to provide feedback to trainees about learning and ultimately how to improve the outcome of learning.
b. **Assessment as learning**: This involves trainees in the learning process, enabling them to monitor their own progress. Trainees use self-assessment and educators' feedback to reflect on their progression, developing and supporting their metacognitive skills. Assessment of learning is crucial in helping trainees become lifelong learners.

c. **Assessment of learning**: This serves to demonstrate the achievement of learning. It is a graded assessment that usually counts toward the trainee's end-of-training degree.
d. **Feedback and evaluation**: Assessment outcomes will represent quality metrics that can improve learning experience.

Miller's Pyramid of Assessment provides a framework for assessing the trainees' clinical competences, and serves as a road map for the trainers to select assessment methods to target different clinical competencies, including "knows," "knows how," "shows how," and "does." Appendix $$.

For the sake of organization, assessment will be further classified into two main categories: *formative* and *summative*.

2. Formative Assessment

2.1 General Principles

Trainees, as adult learners, should strive for feedback throughout their journey of competency from "novice" to "mastery" levels. Formative assessment (also referred to as continuous assessment) is the component of assessment that is distributed throughout the academic year aiming primarily to provide trainees with effective feedback. Every three months, at least one hour should be assigned by trainees to meet with their mentors/program director, in order to review performance reports (via ITERs). Input from the overall formative assessment tools will be utilized at the end of the year to make the decision to promote each individual trainee from the current to subsequent training level. Formative assessment will be defined based on scientific (council/committee) recommendations (usually updated and announced for each individual program at the start of the academic year). According to the executive policy on continuous assessment (available online: www.scfhs.org), formative assessment will have the following features based on Miller's pyramid (Appendix §§):

a. Multisource: a minimum of four tools.
b. Comprehensive: covering all learning domains (knowledge, skills, and attitude).
c. Relevant: focusing on workplace-based observations.
d. The fellowship training is quite homogenous across the three years of training, and therefore, the Competency-milestone-oriented assessment is not applicable to the program.

Trainees should play an active role in seeking feedback during training. However, trainers are expected to provide timely and formative assessments. The SCFHS will provide an e-portfolio system to enhance communication and analysis of data arising from formative assessments.

Trainers and trainees are directed to follow the recommendations of the scientific council regarding the updated forms, frequency, distribution, and deadlines related to the implementation of evaluation forms.

2.2 Formative Assessment Tools

Learning Domain	Formative Assessment Tools	Important details (e.g., frequency, specifications regarding the tool)
Knowledge	- Annual Written Progress Test (promotion), F3 exempted from the promotion exam	Multiple choice question exam once a year
Skills	- Logbook - Research Activities	- During the training program, the fellow should register for a minimum of 70 cases per year while in training, making the total cases required during the 3 years of training at least 210 cases. <u>The total 210 cases obtained during the three-year fellowship-training period should be distributed across the following categories:</u> - 70 patients with suspected/diagnosed metabolic condition. - 70 patients encountered with diagnosis of clinical genetics/dysmorphology. - 20 case of biochemical analysis interpretation. - 20 cases of karyotyping/microarray. - 20 cases for molecular diagnostic interpretation. - 10 from other fields of genetics. In addition to the numerical requirement, logbooks will be evaluated for overall case diversity, complexity of case experience, and distribution of diagnoses within each category. The logbook should be discussed with the program director every 6 months. <u>Research is evaluated over the three years of training as follow</u>: 1st year: - Planning: Critical thinking around clinical questions and reviewing the literature and available resources. - Finalize the hypothesis for the research project and the research methodology. - Write the proposal and get approval. 2nd year: - Start working on the research project; collect the data and present it at meetings.

		3rd year: - Complete the study. - Analyze the data. - Complete writing and provide evidence of submission to a journal at least 3 months prior to the exams. - Presentations/publications.
Attitude	- ITER: In-Training Evaluation Report	Monthly after each rotation. If two or three consecutive rotations involve the same area and same mentor(s), then one evaluation should be sufficient.

The evaluation of each component will be based on the following equation:

Percentage	< 50%	50–59.4%	60–69.4%	> 70%
Description	Clear fail	Borderline fail	Borderline pass	Clear pass

Final year (F3) formative assessment

In addition to the formative tools (logbook, research, and ITER), the final year (F3) should assume/conduct a special assignment, as shown below:

1. Quality project: The fellow should have a significant role in at least one healthcare quality project.
2. Chief-fellow position.
3. Scientific contribution through significant participation in at least one of the following:
   A. Creating and/or maintaining patient database.
   B. Participating in writing at least 2 manuscripts or a book.
   C. Poster presentation in a specialty-related conference.
   D. Heavily participating in community-based activities within the specialty (awareness day or public education).)

(Special permission should be obtained from the Supervisory Fellowship Committee for any special assignment at the SCFHS other than those listed above).

3. Summative Assessment

3.1 General Principles

Summative assessment is a component of assessment that aims primarily to make informed decisions on trainees' competency. In comparison to formative assessment, *summative assessment* does not aim to provide constructive feedback. For further details on this section, please refer to the general bylaws and executive policy of assessment (available online: www.scfhs.org). To be eligible to set for the final exams, a trainee should have been granted a "*Certification of Training-Completion.*"

3.2 Certification of Training-Completion

To be eligible to sit for final specialty examinations, each trainee is required to obtain the *Certification of Training-Completion.* Based on the training bylaws and executive policy (please refer to www.scfhs.org), trainees will be granted the Certification of Training-Completion once the following criteria are fulfilled:

a. Successful completion of all training rotations.
b. Completion of all the training requirements (e.g., logbook, research, others) as outlined in FITER, as approved by the scientific council/committee of specialty.
c. Clearance from SCFHS training affairs ensuring compliance with tuition payments and the completion of universal topics.

The Certification of Training-Completion will be issued and approved by the supervisory committee or its equivalent according to SCFHS policies.

3.3 Final Specialty Examinations

The final specialty examination is the summative assessment component that grants trainees the specialty's certification. It has two elements:

a. Final written exam: In order to be eligible for this exam, trainees are required to hold the Certification of Training-Completion.

b. Final clinical/practical exam: Trainees will be required to pass the final written exam in order to be eligible to set for the final clinical/practical exam.

Examination Format:

- The final clinical genetics and metabolism fellowship examination shall consist of one paper test with 80–120 multiple-choice questions (single best answer out of four options). Ten unscored items are added for pretesting purposes.
- The end-of-year assessment exam is composed of OSCE/OSPE with 7–10 cases
- Passing Score: Please refer to the SCFHS website for updated passing criteria.

Blueprint Outlines: The content of the following table is for demonstration purposes only. Please refer to the most up-to-date version published on the SCFHS website.

The blueprints of the final written and clinical/practical exams are shown in Table..

Example of Final Written Exam Blueprint

*Main blueprint framework adapted from the Medical Council of Canada Blueprint Project

Categories	Basic Medical Science	Approach to Diagnosis	Management
Basics of Genetics 20 %	10	5	5
Clinical Genetics 25%	5	10	10
Clinical Metabolic 25%	5	10	10
Molecular Genetics 10%	5	5	0
Cytogenetics 6%	3	3	0
Genetic Counselling 5%	3	0	2
Cancer Genetics 5%	2	2	1
Research, Ethics, Professionalism, and Patient Safety 4%	2	0	2
Total 100%	35%	35%	30%

The Clinical Genetic and Metabolic Disorders final clinical examination shall consist of:

1- 4 Structured Oral Exam (SOE) stations.
2- Each station for 10–15 minutes.
3- Structured Oral Exam (SOE) stations with 2 examiners each.
4- All stations shall be designed to assess integrated clinical encounters.
5- SOE stations are designed with preset questions and ideal answers.

Example of Final Clinical Exam Blueprint

		DIMENSIONS OF CARE				
		Health Promotion & Illness Prevention Station	Acute Station	Chronic Station	Psychosocial Aspects Station	# Stations
DOMAINS FOR INTEGRATED CLINICAL ENCOUNTER	Patient Care Station		1			1
	Patient Safety & Procedural Skills Station			1		1
	Communication & Interpersonal Skills Station	1				1
	Professional Behaviors Station				1	1
	Total Stations	1	1	1	1	4

Note:

Blueprint distributions of the examination are for demonstration only and may differ up to +/3% in each category, and the number of stations may differ in accordance with assessment department regulations.

For further details on the final exams, please refer to general bylaws and executive policy of assessment (available online: www.scfhs.org).

For SOE example and relevant definitions, refer to *Appendix F*

Learning Domain	Summative Assessment Tools	Passing Score
Knowledge	- Final Written Examination	At least borderline pass on each tool in accordance with the standard-setting method used by the executive administration of assessment
Skills	- Objective Structured Clinical Examinations (OSCE) - Structured Oral Examinations (SOE)	At least borderline pass on each tool in accordance with the standard-setting method used by the executive administration of assessment
Attitude	FITER: In-Training Evaluation Report	Successfully pass FITER

XII. PROGRAM AND COURSE EVALUATION

SCFHS applies variable measures to evaluate the implementation of this curriculum. The training outcomes of this program will be subject to the quality assurance framework endorsed by the Central Training Committee at the SCFHS. Trainees' assessment (both formative and summative) results will be analyzed and mapped to curriculum content. Other indicators that will be incorporated are as follows.

- Report of the annual trainees' satisfaction survey.
- Reports from trainees' evaluation of faculty members.
- Reports from trainees' evaluation of rotations.
- Reports from the annual survey of program directors.
- Data available from program accreditations.
- Reports from direct field communications with trainees and trainers.

Goal-Based Evaluation: The intended achievement of milestones will be evaluated at the end of each stage to assess the progress of the curriculum delivery, and any deficiency will be addressed in the following stage utilizing the time devoted to trainee-selected topics and professional sessions.

In addition to subject-matter opinion and best practices from benchmarked international programs, SCFHS will apply a robust method to ensure that this curriculum utilizes all the data available during the revision of this curriculum in the future.

XIII. POLICIES AND PROCEDURES

This curriculum represents the means and materials outlining learning objectives with which trainees and trainers will interact to achieve the identified educational outcomes. The Saudi Commission for Health Specialties (SCFHS) has a full set of “General Bylaws” and “Executive Policies” (published on the official SCFHS website) that regulate all processes related to training. General bylaws of training, assessment, and accreditation as well as executive policies on admission, registration, continuous assessment and promotion, examination, trainees’ representation and support, duty hours, and leave are examples of regulations that will be applied. Trainees, trainers, and supervisors need to apply this curriculum in compliance with the most up-to-date bylaws and policies that can be accessed online (via the official SCFHS website).

XIV. APPENDICES

A. Competency Matrix

B. Universal Topics Modules

C. Top Conditions and procedures in the Specialty

D. Examples of Formative Assessment Tools

E. Glossary & Relevant Definitions

F. Structured Oral Exam Example

G. References

Appendix A

Competency Matrix: To Map Competency, Learning Domain, and Milestones

Training Year level	*Competency-Roles* (with annotation of learning domains involved: K: knowledge, S: Skills, A: Attitude)	Professional Activities Related to Specialty					
All levels (F1, F2, F3)	Professional Expert	Elicit an accurate and relevant history according to the child's age and presenting complaints, including developmental and family history. (S)	Perform an accurate and focused physical examination and link it with the patient's symptoms. (S)	Identify the appropriate next steps in diagnosis and/or management, including dealing with emergency situations such as: Hyperammonemia, hypoglycemia, and metabolic acidosis (with high anion gap) (S)	Recognize the clinical presentation, epidemiology, course, and prognosis of common childhood genetic diseases in our population, and identify appropriate investigations and basic management for these conditions (K)	Recognize and describe the common dysmorphic features and know the differential diagnoses of those features. (S)	Identify the diseases covered by the National Newborn Screening Program. (K)
	Communicator	Demonstrate compassionate care toward the child with chronic illness and his family. (A)	Collect and synthesize relevant information on the child's condition from patients, their families or caregivers, and other healthcare professional. (S)	Facilitate a multidisciplinary meeting related to a suspected or confirmed childhood genetic/metabolic disease in an effective manner. (S)	Effectively communicate with patient, guardian, team member, showing writing, dictation, and presentation skills. (S)	Convey information that is relevant to the child's condition to patients, their families or caregivers, and other healthcare professionals (both oral and written) with appropriate level of supervision. (S)	

	Collaborator		Participate effectively and appropriately in an inter-professional healthcare team. (A)	Prevent, negotiate, and resolve inter-professional conflicts that may arise while caring for children (e.g., with nurses). (S)	Communicate effectively with other healthcare professionals (e.g., during consultation requests or replies). (S)		
	Advocate		Identify and appropriately respond to individual patient issues while caring for children with suspected or confirmed genetic/metabolic diseases (e.g., social difficulties). (A)	Identify opportunities for advocacy and health promotion (e.g., compliance issues). (A)	Describe the impact of chronic childhood genetic/metabolic diseases on children's participation in normal activities (e.g., at home, school, social settings, and/or work). (S)		
	Leader	Prioritize and manage their time appropriately when conducting assessments of children with suspected or confirmed genetic/metabolic diseases and prioritize their consultation and clinical tasks. (S)	Plan relevant elements of healthcare delivery or academic activity (e.g., patient visit, round's time, and teaching session). (S)	Work collaboratively and efficiently with other members of the healthcare team regarding problems that arise. (S)	Identify medically appropriate investigations for children with suspected or confirmed genetic/metabolic diseases in an ethical and resource-effective manner. (S)	Recognize the impact of drug/benefit coverage on the management of children with suspected or confirmed genetic/metabolic diseases. (K)	

	Scholar	Reflect on their learning needs related to medical genetics. (K)	Formulate appropriate individualized learning questions related to childhood genetic/metabolic diseases. (S)	Access and critically evaluate medical information relevant to identified learning questions (e.g., review relevant literature). (S)	Evaluate the impact of practice changes (through reflective practice) on themselves and on patient care. (K)	Present at least once of a common topic with literature review.	Encouraged to participate in an ongoing research project in the Medical Genetics Division. (A)
	Professional	Demonstrate appropriate professional behaviors in practice, including honesty, integrity, commitment, compassion, respect, and altruism. (A)	Develop rapport, trust, and ethical therapeutic relationships with children and their families in inpatient and outpatient settings. (A)	Recognize and respond appropriately to ethical issues encountered in teaching and clinical practice. (A)	Recognize the principles and limits of patient confidentiality as defined by legal and professional practice standards. (A)	Balance personal and professional priorities to ensure personal health and commitment to professional obligations. (S)	

Appendix B

Universal Topics

Intent:

These are high-value interdisciplinary topics of outmost importance to the trainee. The reason for delivering the topics centrally is to ensure that every trainee receives high-quality teaching and develops essential core knowledge. These topics are common to all specialties. The topics included meet one or more of the following criteria:

- Impactful: Topics that are common or life-threatening
- Interdisciplinary: Hence topics that are difficult to teach in a single discipline
- Orphan: Topics that are poorly represented in the undergraduate curriculum
- Practical: Topics that trainees will encounter in hospital practice

Development and Delivery:

Core topics for the PG curriculum will be developed and delivered centrally by the Commission through an e-learning platform. A set of preliminary learning outcomes for each topic was developed. Content experts, in collaboration with the central team, may modify the learning outcomes.

These topics will be didactic in nature, with a focus on practical aspects of care. These topics will be more content-heavy than workshops and other face-to-face interactive sessions planned.

The suggested duration of each topic is 1:30 hours.

Assessment: The topics will be delivered in a modular fashion. At the end of each learning unit, there will be an online formative assessment. After completion of all topics, there will be a combined summative assessment in the form of context-rich MCQ. All trainees must demonstrate minimum competency in the summative assessment. Alternatively, these topics can be assessed in a summative manner, along with a specialty examination.

Some ideas may include case studies, high-quality images, worked examples of prescribing drugs in disease states, and Internet resources.

Module 1: Inborn error of metabolism (IEM) Disorders

1. Recognition and management of inborn error of metabolism (IEM) emergencies
2. Management of suspected complications (acidosis, hyperammonemia, and hypoglycemia etc.)
3. Management of fluid in hospitalized patients
4. Management of acid-base electrolyte imbalances
5. Comorbidities of common IEM disorders (seizures, cardiomyopathy, liver failure, etc.)

Recognition and Management of Metabolic Emergencies: At the end of the learning unit, you should be able to:

a) Describe pathogenesis of common IEM emergencies including their complications
b) Identify risk factors and groups of patients vulnerable to such emergencies
c) Recognize a patient presenting with IEM emergencies
d) Institute immediate management
e) Refer the patient to appropriate next level of care
f) Counsel patient and families to prevent such emergencies

Management of Complications: At the end of the learning unit, you should be able to:

a) Describe the pathogenesis of important complications of common IEM
b) Screen patients for such complications
c) Provide preventive measures for such complications
d) Treat such complications
e) Counsel patients and families, with special emphasis on prevention

Management of Fluid in Hospitalized Patients: At the end of the learning unit, you should be able to:

a) Review the physiological basis of water balance in the body
b) Assess a patient for his/her hydration status
c) Recognize a patient with over and underhydration
d) Order fluid therapy (oral as well as intravenous) for a hospitalized patient

e) Monitor fluid status and response to therapy through history, physical examination, and selected laboratory investigations

Management of Acid-Base Electrolyte Imbalances: At the end of the learning unit, you should be able to:

a) Review the physiological basis of electrolyte and acid-base balance in the body
b) Identify diseases and conditions that are likely to cause or be associated with acid/base and electrolyte imbalances
c) Correct electrolyte and acid-base imbalances
d) Perform careful calculations, checks, and other safety measures while correcting the acid-base and electrolyte imbalances
e) Monitor response to therapy through history, physical examination, and selected laboratory investigations

Module 7: Ethics and Healthcare

1. Occupational hazards of HCW
2. Evidence-based approach to smoking cessation
3. Patient advocacy
4. Ethical issues: transplantation/organ harvesting; withdrawal of care
5. Ethical issues: treatment refusal; patient autonomy
6. Role of doctors in death and dying

Occupation Hazards of Healthcare Workers (HCW): At the end of the learning unit, you should be able to:

1. Recognize common sources and risk factors of occupational hazards among the HCW
2. Describe common occupational hazards in the workplace
3. Develop familiarity with legal and regulatory frameworks governing occupational hazards among HCW
4. Develop a proactive attitude to promote workplace safety
5. Protect yourself and colleagues against potential occupational hazards in the workplace

Evidence-Based Approach to Smoking Cessation: At the end of the learning unit, you should be able to:

a) Describe the epidemiology of smoking and tobacco usage in Saudi Arabia
b) Review the effects of smoking on the smoker and family members
c) Effectively use pharmacological and non-pharmacological measures to treat tobacco usage and dependence
d) Effectively use pharmacological and non-pharmacological measures to treat tobacco use and dependence among special population groups such as pregnant women, adolescents, and patients with psychiatric disorders

Patient Advocacy: At the end of the learning unit, you should be able to:

a) Define patient advocacy
b) Recognize patient advocacy as a core value governing medical practice
c) Describe the role of patient advocates in the care of the patients
d) Develop a positive attitude toward patient advocacy
e) Be a patient advocate in conflicting situations
f) Be familiar with local and national patient advocacy groups

Ethical Issues: Transplantation/Organ Harvesting; Withdrawal of Care: At the end of the learning unit, you should be able to:

a. Apply key ethical and religious principles governing organ transplantation and withdrawal of care
b. Be familiar with the legal and regulatory guidelines regarding organ transplantation and withdrawal of care
c. Counsel patients and families in the light of applicable ethical and religious principles
d. Guide patients and families to make informed decision

Ethical Issues: Treatment Refusal; Patient Autonomy: At the end of the learning unit, you should be able to:

a) Predict situations where a patient or family is likely to decline prescribed treatment
b) Describe the concept of "rational adults in the context of patient autonomy and treatment refusal
c) Analyze key ethical, moral, and regulatory dilemmas in treatment refusal
d) Recognize the importance of patient autonomy in the decision-making process

e) Counsel patients and families declining medical treatment in the light of best interest of patients

Role of Doctors in Death and Dying: At the end of the learning unit, you should be able to:

a) Recognize the important role a doctor can play during the dying process
b) Provide emotional as well as physical care to a dying patient and family
c) Provide appropriate pain management to a dying patient
d) Identify suitable patients and refer the patient to palliative care services

Appendix C

Top Conditions and Procedures in the Specialty

Top Conditions and Procedures in the Specialty Top Ten Causes of Mortality in Saudi Arabia*			
Disease; Conditions		Relative Frequency	Cumulative Frequency
1.	Fatty acid oxidation defects	NA	NA
2.	Organic acidemia	NA	NA
3.	Maple syrup urine disease	NA	NA
4.	Urea cycle disorders	NA	NA
5.	Lysosomal storage disorders	NA	NA
	Peroxisomal disorders	NA	NA
	Cytogenetic disorders	NA	NA
	Neurogenetic disorders	NA	NA
	Mitochondrial disorders	NA	NA
	Cardiogenetic disorders	NA	NA
*In some specialties it is the overall mortality pattern that is important. However, for others, it might be diseases that are important. The numbers shown here are fictional.			
Top Ten Causes of Out-Patient Consultations by Specialty in Saudi Arabia			
Disease; Conditions		Relative Frequency	Cumulative Frequency
1.	Neurometabolic/neurogenetic disorders	NA	NA
2.	Organic acidemia/urea cycle disorders	NA	NA
3.	Congenital birth defects	NA	NA
	Energy metabolism (e.g., mitochondrial) disorders	NA	NA
	Lysosomal storage disorders	NA	NA
	Dysmorphic disorders	NA	NA
	Cytogenetic disorders	NA	NA
	Genetic counseling	NA	NA

	Adult genetic disorders (e.g., cancer genetics)	NA	NA
	Genetic hearing and/or vision impairment	NA	NA

Top Ten Causes of In-Patient Admissions by Specialty in Saudi Arabia			
Disease; Conditions		Relative Frequency	Cumulative Frequency
1.	Metabolic crisis: Organic acidemia	NA	NA
2.	Metabolic crisis: Urea cycle disorders	NA	NA
	Metabolic crisis: Maple syrup urine disease	NA	NA
	Metabolic encephalopathy and seizures	NA	NA
	Pancreatitis associated with metabolic diseases	NA	NA
3.	Chest infection	NA	NA
	Pre-surgical procedure	NA	NA
	Clinical genetic work-up	NA	NA
	Clinical metabolic work-up	NA	NA
	Adult metabolic patients with comorbidities (e.g., renal, hematological, cardiac)	NA	NA

Top Ten Procedures/Surgeries Performed by Specialty	
Name of Procedures/Surgeries	Approximate Frequency
IV line managements (port-a-cath insertion)	
TPN and lipid management	
Skin biopsy	
Gastrostomy tube insertion	
Scoliosis surgery	
Congenital heart surgery	
Cleft lip/palate surgery	
Other congenital defect surgeries	
Urological surgery	
Other orthopedic surgery	

Examples of Core Specialty Topics: Case Discussions; Interactive Lectures	
Topics	Comments
Approach to patient with hyperammonemia	
Approach to patient with metabolic acidosis	
Approach to patient with hypoglycemia	
Approach to patient with dysmorphic features	
Approach to patient with seizures or other neurological symptoms	
How to read a mutation	
How to read SNP arrays	
How to interpret metabolic results	
How to manage a sick metabolic child in the emergency room	

Examples of Core Specialty Topics: Workshops/Simulation	
Topics	**Comments**
Blood gas interpretation	
Newborn screening and Tandem MS interpretation	
Plasma amino acid interpretation	
Urine organic acid interpretation	
Genetic testing and next generation sequencing data interpretation (e.g., WES, WGS)	
Chromosomal testing interpretation (e.g., array CGH)	

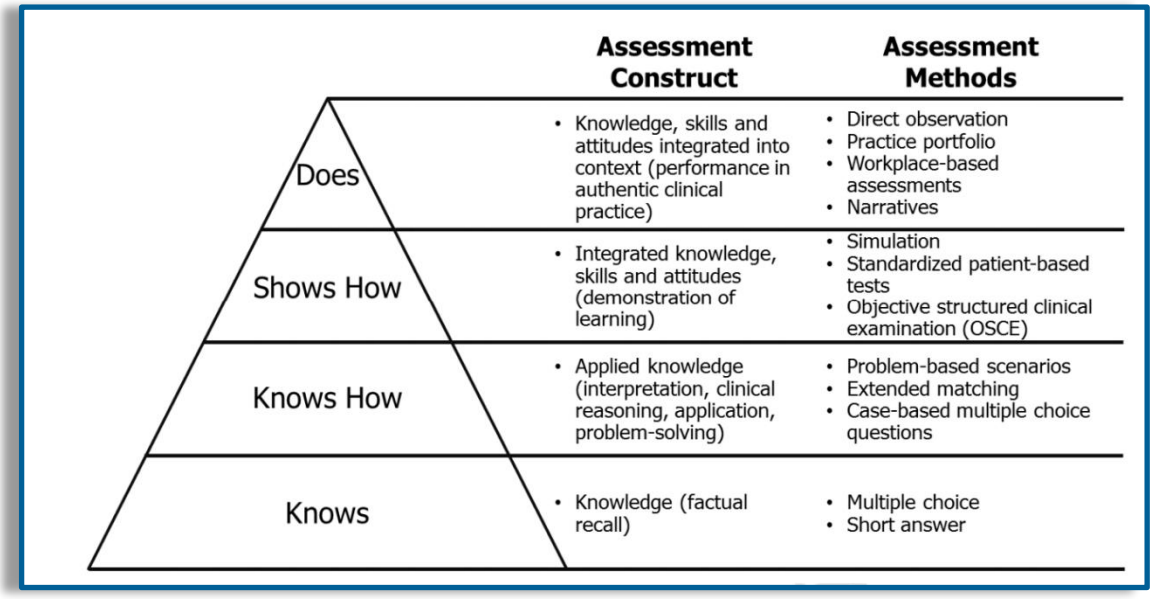
Appendix D

List of Formative Assessment Tools

(According to the Executive Policy on Continuous Assessment, a minimum of 4 tools is needed, and the three domains should be covered. The trainee should show competency in each assessment tool in order to be promoted to the subsequent training level; for further details, please refer to the policy at www.scfhs.org)

Appendix $$

Miller's Pyramid of Assessment provides a framework for assessing the trainees' clinical competences and serves as a road map for the trainers to select the assessment methods to target different clinical competencies, including "knows," "knows how," "shows how," and "does" (2).



(Figure 1. The Miller Pyramid)

1. Adapted from Walsh CM In-training gastrointestinal endoscopy competency assessment tools: Types of tools, validation, and impact. Best Pract Res Clin Gastroenterol 2016 Jun 1;30(3):357-74.
2. Miller GE. Assessment of clinical skills/competence/performance. Acad Med. 1990;65(9 Suppl):S63–7.

Appendix E

Glossary

Glossary	
Blueprint	Description correlating educational objectives with assessment contents. For example, the test blueprint defines the proportion of test questions allocated to each learning domain and/or content.
Competency	Capability to function within a defined professional role that implies entrustment of a trainee by graduation of the program with the required knowledge, skills, and attitude needed to practice unsupervised.
Specialty core content (skills, knowledge, and professional attitude)	A specific knowledge or skill or professional attitude that is specific and integral to the given specialty.
Formative assessment	An assessment that is used to inform the trainer and learner of what has been taught and learned, respectively, for the purpose of improving learning. Typically, the results of formative assessment are communicated through feedback to the learner. Formative assessments are not intended primarily to make judgments or decisions (though it can be as a secondary gain).
Mastery	Exceeding the minimum level of competency to the proficient level of performance, indicating rich experience with possession of great knowledge, skills, and attitude.
Portfolio	A collection of evidence of progression toward competency. It may include both constructed components (defined by mandatory continuous assessment tools in curriculum) and unconstructed components (selected by the learner).

Summative assessment	An assessment that describes the composite performance of the development of a learner at a particular point in time and is used to inform judgment and make decisions about the level of learning and certification.
Universal topic	A knowledge, skills, or professional behavior that is not specific to the given specialty but universal for the general practice of a given healthcare profession.

Dimensions of Care	Focus of care for the patient, family, community, and/or population
Health Promotion & Illness Prevention	The process of enabling people to increase control over their health and its determinants, and thereby improve their health. Illness prevention covers measures not only to prevent the occurrence of illness such as risk factor reduction but also arrest its progress and reduce its consequences once established. This includes but is not limited to screening, periodic health exam, health maintenance, patient education and advocacy, and community and population health.
Acute	Brief episode of illness within the time span defined by initial presentation through to transition of care. This dimension includes but is not limited to urgent, emergent, and life-threatening conditions, new conditions, and exacerbation of underlying conditions.
Chronic	Illness of long duration that includes but is not limited to illnesses with slow progression.
Psychosocial Aspects	Presentations rooted in the social and psychological determinants of health that include but are not limited to life challenges, income, culture, and the impact of the patient's social and physical environment.

Domains	Reflects the scope of practice & behaviors of a practicing clinician
Patient Care	Exploration of illness and disease through gathering, interpreting, and synthesizing relevant information that includes but is not limited to history taking, physical examination, and investigation. Management is a process that includes but is not limited to generating, planning, organizing care in collaboration with patients, families, communities, populations, and healthcare professionals (e.g., finding common ground, agreeing on problems and goals of care, time and resource management, and roles to arrive at mutual decisions for treatment)
Patient Safety & Procedural Skills	Patient safety emphasizes the reporting, analysis, and prevention of medical error that often leads to adverse healthcare events. Procedural skills encompass the areas of clinical care that require physical and practical skills of the clinician integrated with other clinical competencies in order to accomplish a specific and well-characterized technical task or procedure.

Communication & Interpersonal Skills	Interactions with patients, families, caregivers, other professionals, communities, and populations. Elements include but are not limited to active listening, relationship development, education, verbal, non-verbal, and written communication (e.g., patient centered interview, disclosure of error, informed consent).
Professional Behaviors	Attitudes, knowledge, and skills based on clinical and/or medical administrative competence, ethics, societal, and legal duties resulting in the wise application of behaviors that demonstrate a commitment to excellence, respect, integrity, accountability, and altruism (e.g., self-awareness, reflection, life-long learning, scholarly habits, and physician health for sustainable practice).

	Examination	Not done	Done
Examiner	Give 3 differential diagnosis of elevated Hcy		
Candidate	Cystathionine beta-synthase deficiency (High Meth and Hcy)	1	
	Methylenetetrahydrofolate reductase deficiency (low Methionine and high Hcy)	1	
	Disorders of intracellular cobalamin metabolism (C, D, F, E, and G) (some with elevated MMA)	1	
Examiner	Which kind of investigations are needed and why you order them?		
Candidate	Order some of following investigations. Tandem mass spectrometry, urine organic acid, plasma amino acid, vitamin B12, urine and serum methylmalonic acid, serum / RBC folate, CSF folate +/- CSF neurotransmitters, brain MRI, gene testing MTHFR, cobalamin complement probe assay to differentiate between the cobalamin disorders	2	
	Need to see the methylmalonic acid and methionine level to narrow the differential diagnosis	1	
Examiner	You have the result of plasma amino acid: low methionine (show it to the candidate), What is the diagnosis in this case?		
Candidate	Methylenetetrahydrofolate reductase deficiency	1	
Examiner	Outline the management for this patient, including medications		
Candidate	Admit the patient and order investigations.	1	

	The following medications are used in methylenetetrahydrofolate reductase deficiency a) Methionine supplementation b) Betaine c) Folic acid d) Folinic acid e) Hydroxocobalamin f) Aspirin g) Pyridoxine (Vitamin B6) h) Antiepileptic medications	2	
Examiner	(Total marks: /10)		
	Total station points: 10 Calculated MPL:		

Appendix F

Structured Oral Exam Example

Clinical Exam Clinical Genetic and Metabolic Disorders Fellowship Program	Station 1 Instructions to Resident

Clinical Scenario:

A 2-year-old male presenting with seizures and lethargy is known to have global developmental delay, progressive intellectual disability, hypotonia, and microcephaly, and family history revealed that the severely affected sibling had onset of intractable seizures at the age of 1 month and died at the age of 7 months, and that two relatives have a history of coronary artery disease, psychiatric illness, and early stroke. His initial metabolic work-up showed persistent total homocysteine elevations of 45–120 (normal ≤ 15.0 μmol/L)

Instructions to Resident:

YOU HAVE 15 minutes TO DO THE FOLLOWING:

Read the scenarios and answer the questions asked by the examiners.

Appendix G

References:

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