HONG KONG COLLEGE OF PAEDIATRICIANS
SUBSPECIALTY ACCREDITATION
CURRICULUM PROPOSAL - Paediatric Inherited Metabolic Medicine (PIMM)
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Introduction

Paediatric Inherited Metabolic Medicine (PIMM) is a well-recognized paediatric subspecialty. It encompasses prevention, recognition, diagnosis and management of all aspects of inherited metabolic disorders (IMD) that affect the body's normal biochemical processes. Many IMD patients have neurodevelopmental involvement and require life-long multidisciplinary family-centred care.

To equip a prospective metabolic paediatrician with attitude and skillsets for providing and driving high-standard professional PIMM services in Hong Kong, a PIMM subspecialty training programme is proposed as detailed in this document. The aim is to provide PIMM trainees an accredited curriculum, which provides a framework for training, articulating the standard required to work as a PIMM sub-specialist, and encouraging the pursuit of excellence in all aspects of clinical and wider practice.

The proposed training programme comprises of minimum of 36 months' training with 4 core plus 1 elective modules, which covers the essential facets of fundamental knowledge and skills and forms the solid foundation for the lifelong PIMM career development.

Eligibility

- Specialists holding the qualification of FHKAM (Paediatrics) or its equivalent
- Candidates who have completed 3 years basic training in general paediatrics and have passed the MRCPCH (UK) / HKCPaed Intermediate Examination are eligible to commence the PIMM subspecialty training during their higher training in General Paediatrics upon the approval by the PIMM Subspecialty Board. A maximum of 1 cumulated year's overlap is allowed during this period.

PIMM curriculum

The purpose of the PIMM curriculum is to support the trainee paediatrician in developing the knowledge, skills and attitudes required to work safely and confidently as a PIMM subspecialist.

PIMM subspecialists provide care for children, young people and families who have inherited disorders that affect the body's normal biochemical processes and lead to organ dysfunction. They have detailed knowledge of normal human biochemistry and physiology and the impact of IMD. They are skilful to apply this knowledge in the diagnostic process (including identifying novel disorders) and patient- and family-centred management.

PIMM subspecialists work closely with laboratory scientists, metabolic dietitians, pharmacist specialists, nursing and allied health teams. They strive to improve the early recognition and diagnosis of metabolic diseases, including through newborn screening. They keep up to date with the rapid developments and innovations in therapeutics for IMD, and provide consistently high quality PIMM services.

By the end of the PIMM training, a PIMM trainee must demonstrate the achievement of the following learning outcomes, which are mapped to the domains of learning outcomes as listed in the 2023 Hong Kong College of Paediatricians (HKCP) Curriculum Statement.

Learning outcomes of PIMM curriculum

PIMM Learning Outcomes		HKCP curriculum statement domains
1	Applies up-to-date understanding of the full range of metabolic conditions to the holistic metabolic assessment and diagnosis to patients with suspected and confirmed IMD	1, 2, 3, 4, 5, 10
2	Prevents, recognises, assesses and manages the full range of acute paediatric inherited metabolic emergencies	2, 3, 4, 5, 6, 7
3	Supports patients and families with suspected or confirmed IMD presenting clinically or detected through the newborn screening	1, 2, 3, 4, 5
4	Counsels families the inheritance of IMD within a cultural context	1, 2, 3, 4, 5
5	Provides consistently high quality PIMM service across specialties, disciplines, sectors, in both community and hospital settings	1, 2, 3, 4, 5, 6, 7, 8, 9, 10
6	Contributes to multi-centre collaborations and research	2, 5, 6, 8, 10, 11

PIMM training program at a glance

Module	Location in Hong Kong^	Duration
Core module 1@: Paediatric inherited metabolic medicine (clinical)	- Hong Kong Children's Hospital (HKCH) Metabolic Medicine Team - Other accredited PIMM training centre	Minimum 24 months (if being the only metabolic medicine team for training) Up to 6 months plus a minimum of 18 months in HKCH metabolic medicine team make up to a total of minimal 24 months
Core module 2: Paediatric inherited metabolic medicine (laboratory)	- Chemical pathology laboratories under Hospital Authority providing metabolic diagnostic services with training accreditation under The Hong Kong College of Pathologists - Newborn screening laboratory in Hong Kong Children's Hospital - Research laboratory on metabolic medicine (local: University of Hong Kong and Chinese University of Hong Kong; or overseas academic unit)	3 months
Core module 3: Paediatric neurology with preference to neuro-metabolic medicine	- Accredited Paediatric Neurology training centre under The Hong Kong College of Paediatricians	3 months
Core module 4: Clinical genetics & genomics	- Accredited Paediatric Genetic and Genomics training centre under The Hong Kong College of Paediatricians	3 months
Elective module@: Clinical specialties closely related to paediatric metabolic medicine	PIMM board recognized centre providing the following: - Paediatric subspecialty service closely related to PMM	3 months
	Total minimum duration	36 months

*An academic-oriented training with basic or clinical research component is highly encouraged.

^Overseas training in institutions with a recognized PIMM training programme is strongly recommended.

@Exposure to an adult metabolic medicine unit e.g. Princess Margaret Hospital is highly encouraged.

PIMM training syllabus

PIMM training syllabus supports the completion of the PIMM curriculum. It provides a reference guide on how to acquire the minimum set of competencies in specific domains, demonstrate the acquisition progress, and achieve the learning outcomes during the course of PIMM training and career development.

At the completion of training, trainees should demonstrate competencies as PIMM subspecialist. They should have acquired detailed knowledge of normal biochemistry and physiology as well as the impact of such disorders (as listed on the International Classification of Inherited Metabolic Disorders) on the body, and are able to critically apply the knowledge, to strive for early recognition, diagnosis, and effective management of the affected babies, children and young people as well as their families, including through screening, multidisciplinary care, and research.

The following minimum set of competencies are mapped to the domains of training competence as listed in the 2023 Hong Kong College of Paediatricians (HKCP) Curriculum Statement.

Minimum set of competencies of a PIMM subspecialist

Minimal Competencies of a PIMM subspecialist		HKCP curriculum statement domains
1	Applies up-to-date understanding of the full range of metabolic conditions to the holistic metabolic assessment and diagnosis to patients with suspected or confirmed IMD	1, 2, 3, 4, 5, 10
	A. Demonstrates sound understanding of the normal biochemistry and physiology: - Requirement, regulation and balance of fluid, electrolytes, acid/base and nutrients - Intermediary metabolism e.g. metabolic response to fasting, and metabolism of glucose, lactate, ammonia, amino acids, organic acids, fatty acids - Complex metabolite metabolism e.g. lipids, lipoproteins, cholesterols and other sterols, purines, pyrimidines, porphyrins, bilirubin - Vitamin, mineral/metal metabolism, including role of cofactors - Brain development and metabolism, including normal intellectual and psychological development, and role of blood brain barrier and neurotransmitters - Organelle functions and regulations, including mitochondria, endoplasmic reticulum, golgi, lysosome, peroxisome, ribosome - Enzyme biochemistry and tissue expression B. Demonstrates sound understanding of the impact and corresponding diagnostics of IMD: - Genetic, biochemical, pathological, and clinical changes as a result of IMD - Consequence of specific nutritional excess or deficiencies - Indications, selection, planning and interpretation of biochemical and genetic/genomic	

	investigations, functional studies, histology and histochemistries, newborn screening, and peri-/post-mortem tests, including the understanding of analytical, physiological and nutritional factors that influence the results C. Takes responsibility for appropriate investigations of the full range of IMD manifestations and holistic patient assessments in both acute and out-patient settings, and be aware of the possible non-IMD differential diagnoses: - Neurological manifestations including developmental delay / regression, seizures, movement disorders, myopathy, encephalopathy etc - Liver manifestations including cholestasis, hepatomegaly, acute or chronic liver dysfunction - Other organ manifestations e.g. cardiorespiratory, muscle, kidney, eye, endocrine, bone, skin, dysmorphism etc - Abnormal growth and nutritional status D. Undertakes bedside procedures including lumbar puncture, tissue biopsies, and dynamic tests	
2	Prevents, recognizes, assesses and manages the full range of acute paediatric inherited metabolic emergencies	2, 3, 4, 5, 6, 7
	A. Educates patients, families and schools on the risk factors and signs of impending metabolic decompensation, including advice and action plans for: - Dietetic, exercise and fasting precautions - Anaesthetic and surgical considerations - Intercurrent illnesses - Impending metabolic decompensation B. Prevents, recognises, assesses and manages the full range of acute metabolic emergencies including advice to clinicians from community and regional hospitals: - Appropriate choice and timing of investigations and monitoring of patient conditions - Timely and practical use of emergency fluid/electrolyte/nutritional management, assisted ventilation and dialysis - Timely prescription and administration of a range of specialised drugs for IMD - Monitoring and management of IMD drug associated reactions and side effects	
3	Supports patients and families with suspected or confirmed IMD presenting clinically or detected through the newborn screening	1, 2, 3, 4, 5
	A. Be skillful in counseling for a new diagnosis in simple terms: - Classification, investigations, monitoring, as well as possible clinical presentations and long-term complications of the suspected or confirmed IMD - Role and indications of treatment modalities e.g. diet, drug, transplantation, palliation	

	B. Empowers families in the daily management, adapted to clinical settings and social context, including: - Provides empathetic support to families especially during difficult circumstances - Appreciates patient and families' understanding and psychological stress towards the metabolic disease, and the influence of ethnic and culture difference on their attitudes - Understands patient and families' challenge in facing progressive disorders especially those where natural history is difficult to predict - Explores effective palliative care for degenerative diseases	
4	Counsels families the inheritance of IMD within a cultural context	1, 2, 3, 4, 5
	A. Be skillful in genetic counseling in simple terms, adapted to cultural context: - The principles and impact of autosomal recessive, dominant, X-linked and mitochondrial DNA inheritance patterns - The utilization of pre-implantation / prenatal / postnatal / pre-symptomatic genetic diagnosis, taking cultural differences in attitudes in the consideration B. Be familiar with the currently available modalities of genetic/genomic analysis and principles of variant classification, to guide: - Appropriate modalities, samples, timing, and informed consents of the tests Explanation of the results and the possible need of data reanalysis or additional testing in the future	
5	Liaises effectively with clinicians and specialists (especially nurses, dietitians, pharmacists, laboratory scientists, allied health and social workers) in community, regional hospitals and specialist centres, to provide consistently quality service for patients with IMD.	1, 2, 3, 4, 5, 6, 7, 8, 9, 10
	A. Takes responsibility for the long-term holistic management of families with IMD: - Appropriately selects and prescribes supportive and specific treatment options including drug / diet / enzyme therapies, stem cell / organ transplantation and gene therapy - Understands the indications and interpretations of neuropsychometric assessments - Identifies the educational, social, psychological and palliative care needs early and initiates timely support B. Contributes to effective running and development of the metabolic unit: - Keeps the clinical team fully engaged with the rapid evolving development in diagnostics and therapeutics for PIMM and be a steadfast patient advocate - Develops clinical leadership, management and administrative skills e.g. business and budget planning, quality and safety, clinical audit, maintaining duty rotas and human resources	

	- Be interested in the overall organization of departmental activities and management directions - Upholds a constructive attitude to the process of decision making and accepts shared responsibility for the use of resources - Responds effectively to clinical complaints C. Contributes to effective and up-to-date local and regional multidisciplinary team (MDT) management: - Communicates effectively with patients and families, colleagues and working partners, research staff and administrators, to understand the perspectives of stakeholders in patient care - Upholds a collaborative problem-solving attitude towards colleagues and families, recognising and coping with stress in self and others - Understands local management structure and perspectives of other stakeholders including policy makers - Contributes to the formulation, coordination, and execution of effective MDT care plans	
6	Contributes to multi-centre collaborations and research	2, 5, 6, 8, 10, 11
	A. Be committed to continuing self-education and to teaching others - Be up-to-date with literature and online resources on IMD - Have a working knowledge on the design and execution of clinical studies and audit, including GCP (good clinical practice), critical appraisal, medical statistics, data organization and presentation, manuscript preparation, oral presentation - Contributes regularly to the organization of educational programmes and development of core teaching materials at both undergraduate and postgraduate levels - Teaches at various levels including undergraduate and postgraduate - Adheres to a positive empathetic approach to the supervision and motivation of junior medical staff and medical students B. Be active and supportive in research - Be familiar with the current state of clinical trials and novel therapies for untreatable IMD, with a good understanding of the role of disease registries, natural history studies, and clinical trials in the IMD management and treatment development - Participates in MDT discussions about local and international collaborations at clinical and research levels, including study recruitment	

Benchmark references

1. SSIEM Syllabus for training in clinical paediatric metabolic medicine (Updated 30 Jun 2017)
https://www.ssiem.org/index.php?option=com_content&view=article&id=41&Itemid=176

2. RCPCH Progress+ Paediatric Inherited Metabolic Sub-specialty Syllabus (Approved by GMC on 1 Aug 2023)
https://www.rcpch.ac.uk/sites/default/files/2023-07/progressplus-metabolic-medicine-syllabus-2023.pdf

2021 ICIMD Categories of Inherited Metabolic Diseases

1. Disorders of amino acid metabolism
2. Disorders of peptide and amine metabolism
3. Disorders of carbohydrate metabolism
4. Disorders of fatty acid and ketone metabolism
5. Disorders of energy substrate metabolism
6. mtDNA-related disorders
7. Nuclear-encoded disorders of oxidative phosphorylation
8. Disorders of mitochondrial cofactor biosynthesis
9. Disorders of mitochondrial DNA maintenance and replication
10. Disorders of mitochondrial gene expression
11. Other disorders of mitochondrial function
12. Disorders of metabolite repair / proofreading
13. Miscellaneous disorders of intermediary metabolism
14. Disorders of lipid metabolism
15. Disorders of lipoprotein metabolism
16. Disorders of nucleobase, nucleotide and nucleic acid metabolism
17. Disorders of tetrapyrrole metabolism
18. Congenital disorders of glycosylation
19. Disorders of organelle biogenesis, dynamics and interactions
20. Disorders of complex molecule degradation
21. Disorders of vitamin and cofactor metabolism
22. Disorders of trace elements and metals
23. Neurotransmitter disorders
24. Endocrine metabolic disorders

Reference:
An international classification of inherited metabolic disorders (ICIMD).
Carlos R Ferreira, Shamima Rahman, Markus Keller, Johannes Zschocke, ICIMD Advisory Group.
J Inherit Metab Dis. 2021 Jan;44(1):164-177.doi: 10.1002/jimd.12348.
https://pubmed.ncbi.nlm.nih.gov/33340416/