Genetics & Genomics (Paediatrics)

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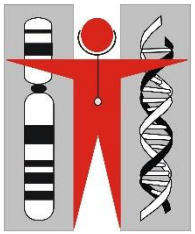
Consultant

Clinical Genetic Service

Department of Health



Existing service providers

Clinical Genetic Service, DH

- Providing territory-wide medical genetic services since 1981
- Genetic counseling unit
  - Diagnostic
  - Counselling
    - Family
- Genetic laboratory
  - Cytogenetics
  - Molecular genetics
- Genetic screening unit
  - Newborn screening

Caseload

- Genetic Screening Clinic
  - 2 sessions/week
    - CSWJCC
  - ~1250 attendance/year
- Genetic Counselling Clinic
  - 5.5 sessions/week
    - CSWJCC
    - QEH ACC
    - QEH L-blk
  - 4500 attendances/yr
    - ~1500 new
    - ~3000 follow-up

Existing service providers

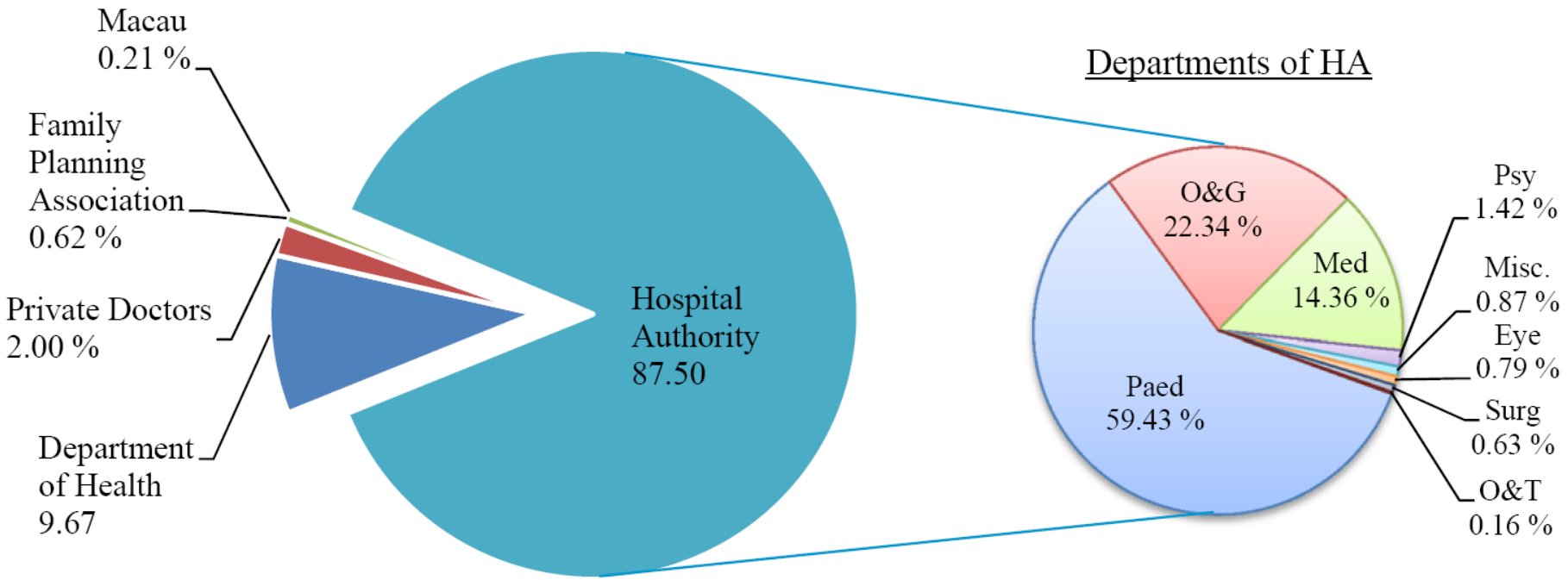
HKU/QMH/TYH

- Prenatal/preimplantation genetic clinic since 2010
- Clinical genetics clinic since 2010
  - 3 clinic sessions/wk
  - 600-700 cases/yr
- TYH Genetic laboratory
  - Cytogenetics
  - Molecular genetics
- In-patient consultation

CUHK/PWH

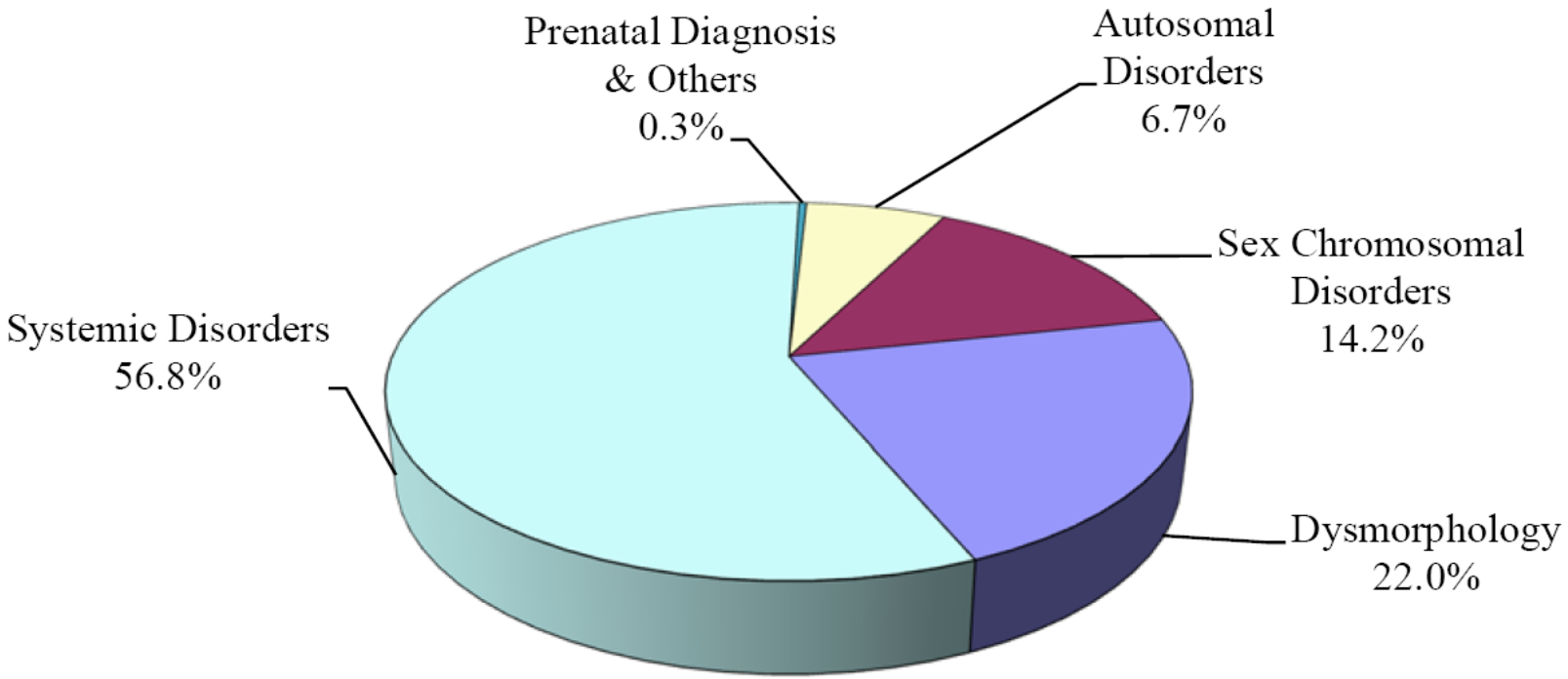
- Prenatal diagnosis clinic
  - ~100 cases/yr
- Paediatric genetic/metabolic clinic
  - ~100 cases/yr
- Laboratory
  - Cytogenetics
  - Molecular genetics
  - Biochemical genetics

Patients (CGS)



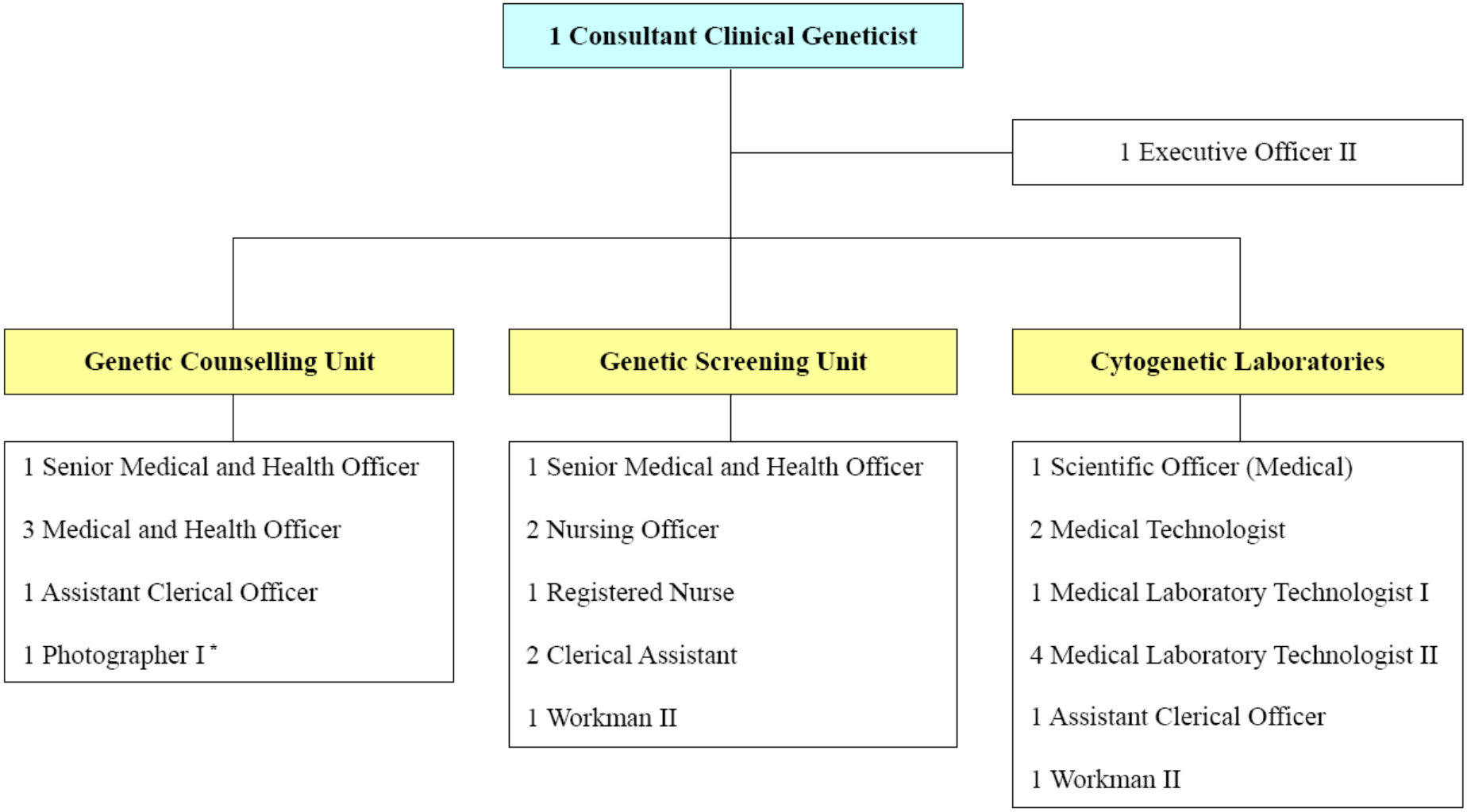
Source of Referrals from Different Institutes in 2015

Spectrum of genetic disorders (CGS)



Types of Disorders for Genetic Counselling in 2015

Establishment and Organization Chart of Clinical Genetic Service
2015



CGS in HKCH

- The major, if not only, centre to deliver medical genetics/genomics services and training in HK
  - Advantages
    - More direct communication and interaction with other disciplines
    - Closer collaboration
      - Service delivery
      - Research
    - Concentration of complex cases, expertise and genetics/genomics technologies

Service Delivery in HKCH

- In-patient consultation/counselling
- Out-patient clinic (genetics/genomics)
- Combined clinic with other specialties/subspecialties

Training opportunities

- Pre-membership
  - 3 month elective during basic training (one more choice besides CAS and MCHC)
- Post-membership/Pre-fellowship
  - 3-6 months during higher training (optional)
  - 6-12 months (pre-G&G subspecialty training)
- Post-fellowship
  - Short term attachment for other subspecialty trainees
  - E.g. Developmental, neurology, endocrine, etc.
  - Genetics & Genomics subspecialty training (3 yrs)
    - Inter-collegial
    - Pathology/OG/Adult medicine

Hong Kong College of Paediatricians

A proposal of training curriculum for Paediatric Subspecialty Training Programme:

Genetics & Genomics [Paediatrics]

遺傳學與基因學 [兒科]

Dr. Chung Hon-Yin, Brian

Dr Lam Tak-Shum, Stephen

Dr Lo Fai Man, Ivan

Dr Luk Ho Ming

Dr Tsui Kan Ming

	and the transmission of genomic material	

2. Human gene structure and function

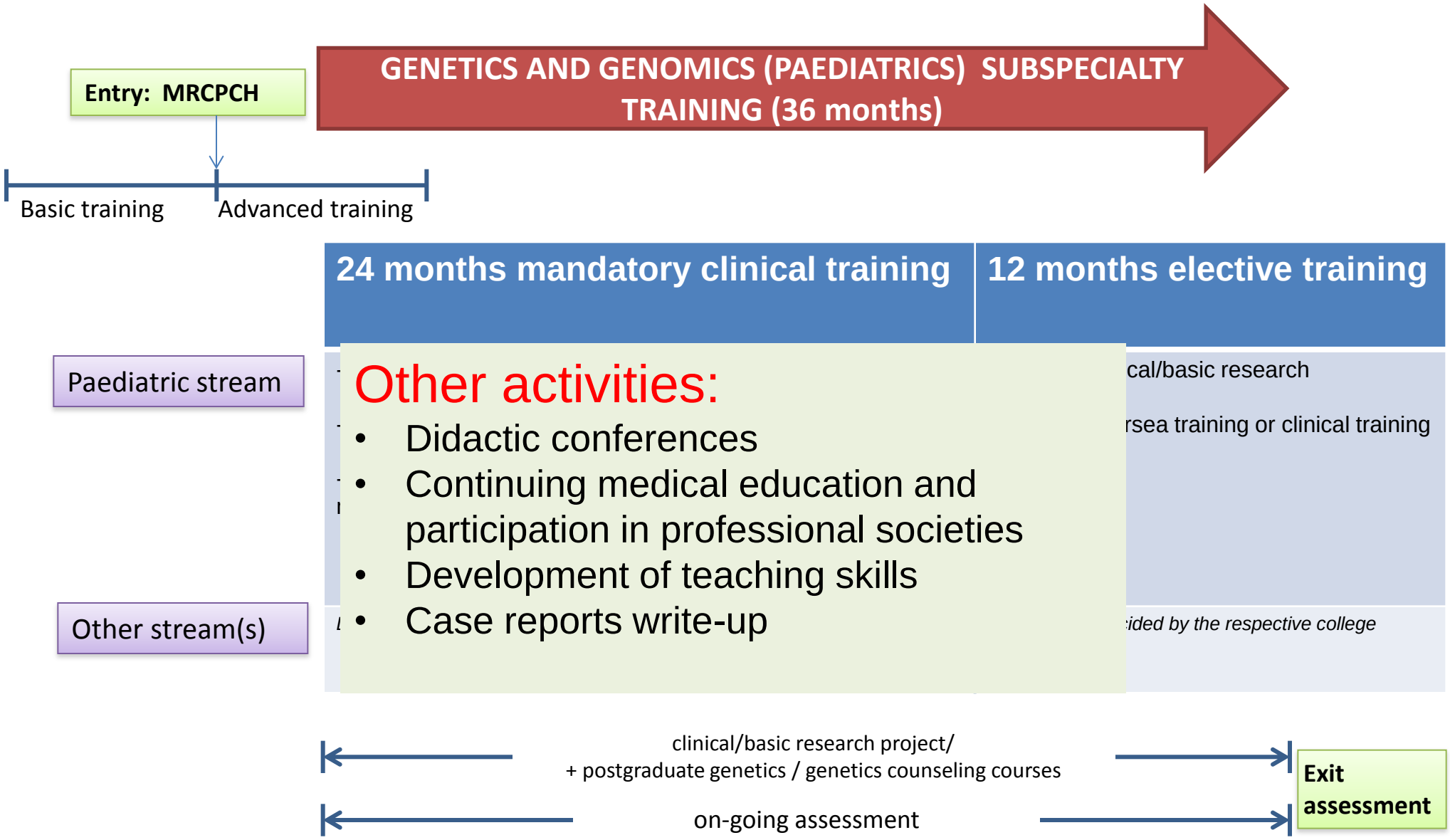
Objectives	Specific knowledge and skills	Trainers (by colleges)
Understand the general principles of human genetics at gene level	a. Explain the organization and structure of genes b. Explain basic gene expression: transcription through to translation c. Explain gene regulation including transcription, splicing, variation of gene expression between tissues and relevance to medicine d. Explain post-transcriptional mechanisms including post-translational modifications	HKCPaed

3. Mendelian inheritance

Objectives	Specific knowledge and skills	Trainers (by colleges)
Understand the general concepts of single gene disorders and factors modifying these disorders	a. Describe the Mendel's laws of inheritance b. Describe the basic principles of Mendelian inheritance c. Understand concepts of penetrance, expressivity, anticipation, hypomorphic alleles and pseudodeficiency d. Explain how epigenetics influence phenotype e. X-linked inheritance: describe the effect of skewed X-inactivation may have on phenotype in females f. Demonstrate ability to infer inheritance patterns by pedigree analysis g. Give examples of genotype-phenotype correlation in medical conditions	HKCPaed HKCPhy

4. Molecular genetics concepts and testing methods

Objectives	Specific knowledge and skills	Trainers* (by colleges)
Understand the general principles of molecular technology as applied to medicine	a. Understand the basic principles of the polymerase chain reaction b. Understand the concepts of nucleic acid sequencing including Sanger and massively	HKCPaed HKCPath HKCOG


Entry: MRCPCH
GENETICS AND GENOMICS (PAEDIATRICS) SUBSPECIALTY TRAINING (36 months)
Basic training
Advanced training
24 months mandatory clinical training
12 months elective training
Paediatric stream
Other stream(s)
cal/basic research
rsea training or clinical training
ided by the respective college
Other activities:
• Didactic conferences
• Continuing medical education and participation in professional societies
• Development of teaching skills
• Case reports write-up
clinical/basic research project/
+ postgraduate genetics / genetics counseling courses
on-going assessment
Exit assessment

Training objectives

The mission of this Genetics and Genomics (Paediatrics) subspecialty training programmes is to produce paediatricians who:

1. Are clinically competent in the field of clinical genetics and genomics;
2. Are capable to serve children in Hong Kong in a variety of settings; and
3. Possess attitudes and skills of life-long learning to build upon their knowledge, skills and professionalism.

Curriculum

a) Diagnose and manage genetic disorders
b) Provide genetic counselling to patients and families
c) Apply knowledge of genetic disorders with respect to the heterogeneity, variability and natural history in patient-care decision making
d) Elicit and interpret individual and family medical histories
e) Interpret cytogenetic, molecular genetics, and specialized laboratory testing information
f) Explain the causes and natural history of genetic disorders and genetic risk assessment
g) Interact with other health care professionals in the provision of services for patients with genetic disorders