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]

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Training Programme: Genetics and Genomics (Paediatrics)

Contents

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Source of document

We gratefully acknowledge the inputs from our external referees and various stakeholders. The current document is shaped largely by the influence of the following essential documents with due consideration of our local setting and existing training facilities.

Hong Kong College of Paediatricians

- Guidelines on the Criteria for the Accreditation of a Paediatric Subspecialty Training Programme

The Hospital Authority of Hong Kong

- A review of Genetic and Genomic Services in Hong Kong

Preparatory Committee on the Strategies of Genetic and Genomic Services in Hong Kong (DH, HKAM, HA, CUHK, HKU)

- Report of Advisory Group for Preparatory Committee on Strategies of Genetic and Genomic Services in Hong Kong

Hong Kong Society for Paediatric Immunology and Infectious Diseases (HKSPID)

- A proposal of training curriculum for Paediatric Subspecialty Training Programmes: Paediatric Immunology and Infectious Diseases (Revised version 3)

Joint Royal College of Physicians Training Board, United Kingdom

- Specialty Training Curriculum for Clinical Genetics

The Royal Australasian College of Physicians

- Clinical Genetics Advanced Training Curriculum

Accreditation Council for Graduate Medical Education, USA (ACGME)

- Guidelines for combined training in Pediatrics and Medical Genetics leading to dual certification

Canadian College of Medical Geneticists

- CCMG General Knowledge Training Guidelines

Preface – History of development for Clinical Genetics & Paediatrics in Hong Kong

Paediatrics as a discipline in Hong Kong started during the 1960s when the Department of Paediatrics was established in the Queen Mary Hospital, with the corresponding academic department in the University of Hong Kong. In 1977, Professor Hutchison took up the Chair of Paediatrics in HKU. Realizing the importance of genetics for the university and Hong Kong as a whole, Professor Hutchison persuaded the Vice-Chancellor of HKU to invite Professor M Ferguson-Smith from the Department of Medical Genetics of the University of Glasgow to visit and advise the university on the feasibility of setting up a comprehensive genetic service. Following Professor Ferguson-Smith's report, the then Medical and Health Department arranged for Professor Paul Polani from the Paediatric Research Unit of Guy's Hospital to visit and submit a further report. As a result, a Clinical Genetic Service was established by the government in 1981; however Professor Hutchison felt "the opportunity to incorporate a strong university and research component seems to have been missed".

In the following four decades, another academic department in the Chinese University of Hong Kong and many more departments of paediatrics in regional hospitals came into being mirroring the rapid development of Hong Kong. The Academy of Medicine with specialty colleges was established in the early 1990s, with the statutory duty to ensure proper training and accreditation of paediatric specialists. With the anticipation of the establishment of the Hong Kong Children's Hospital in 2018, our College has started the next stage of our establishment in paediatric subspecialty development. In 2011, the Hospital Authority, in collaboration with Department of Health has commissioned a consultancy study on local genetics and genomics services, led by Professor Ron Zimmern, Public Health Genomics Foundation of the University of Cambridge. The scope of the study covers the review of genetics and genomics services in Hong Kong and recommendation for future development of such services across a range of specialties in the public, private and academic sectors.

In order to follow-up on the recommendation, a Preparatory Committee on the Strategies of Genetics and Genomic Services in Hong Kong consisting of representatives from the Department of Health, Hospital Authority, Hong Kong

Academy of Medicine, the Chinese University of Hong Kong and the University of Hong Kong was formed. The committee recognized an immediate need to build the capacity and critical mass of expertise in genetics that is up to the international standards for the development of genetics and genomics services in the territory. In this connection, a Genetics and Genomics Working Group was set up within the Hong Kong Academy of Medicine to advise and coordinate the necessary training and accreditation of clinical genetics and genomics by the relevant colleges. The Hong Kong College of Paediatricians is one of the five Colleges (the other colleges being Hong Kong College of Obstetrics and Gynaecology, Hong Kong College of Pathologists, Hong Kong College of Physicians, and Hong Kong College of Community Medicine) that recognized the importance of genetics and genomics and participated actively in the Working group.

While most other subspecialties in Paediatrics stemmed from their counterparts in adult medicine, Clinical Genetics uniquely originates from Paediatrics and expands to adult medicine and other disciplines. It is the subspecialty concerned with the diagnosis of inherited disorders and birth defects, with the estimation of genetic risks and with genetic counselling of family members. The specialty is constantly changing and the clinical geneticists must take account of the new knowledge, molecular and other new areas of developments, including bioinformatics and next generation sequencing (hence “genomics”), and alter clinical practice accordingly. It is based on this understanding that our colleagues in Australasia, Europe and North America have already established their own comprehensive subspecialty training in their countries. With a population of 7 million in Hong Kong, we strongly believe that the subspecialty is needed and with open-mindedness, dedication and care we shall create a future secure for our next generation of subspecialists and patients/families which is the central idea of this training programme.

B Chung and I Lo
7 July 2015

Introduction

The programme of General Paediatrics has been steered by the College since its inception in 1991 with remarkable success. Now the College is entering into a new era and facilitates subspecialty groups to accomplish subspecialty accreditation, including the discipline of Genetics & Genomics [Paediatrics].

Genetic conditions/congenital malformations are defects of morphogenesis of organs, identifiable at birth. Very often patients with genetic conditions have multiple organ/system malfunction, and results in significant mortality and morbidity. Although each type of genetic condition is rare, the total numbers of infants that are born with rare disease make up to 2-3%. The WHO estimates that ~260,000 deaths worldwide were caused by birth defects in 2004. In Hong Kong, infant mortality rate (IMR) has dropped from 89.1 per 1,000 live births in 1946 to 1.8 in 2006 and the trend of proportional mortality has also changed significantly: congenital malformations and chromosomal abnormalities ranked top and accounted for one third of the proportional mortality in 2001-2006 (Department of Statistics, HKSAR). A territory-wide birth defect/ genetic disorder registry is not available in Hong Kong. We have done a territory-wide hospital-based study in 2005, showing that the annual hospitalization rate of children with birth defects/genetic disorders was 6 per 1,000 children (Chung et al. 25th International Congress of Paediatrics, 2007). This constituted 8.7% (~8,000 admissions) of paediatric hospitalizations in Hong Kong. Interestingly, the hospitalization rate is similar to Europe and North America but the etiologic pattern is very distinct. Since genetic conditions are heritable, family members can often be affected. Proper diagnosis leads to disease-specific therapeutics as well as primary, secondary and tertiary prevention of these devastating conditions for individuals, families and the wider society.

We envisage a genuine need in Hong Kong to establish a training programme encompassing clinical genetics and genomic medicine. In the Report of Advisory Group for Preparatory Committee on Strategies of Genetics and Genomic (GG) Services in Hong Kong, the “lack of formal and structured training for professionals particularly for clinicians” is identified as a major service gap and it is recommended that the imminent need of clinical genetics and genetic counselling as a specialty/subspecialty has to be further explored by the Academy with its Colleges. Several recommendations have been made by overseas professional societies e.g. Joint Royal College of Physicians Training Board (UK) and experts invited to provide

consultancy reports at different times, including Professor M Ferguson-Smith (1978), Professor P Polani (1980) and most recently Professor R Zimmern (2011).

The draft panel members are Fellows of the Hong Kong College of Paediatricians and are recognized at institutional level with the appointment of honorary professor, associate professor, consultants and specialist fellow in the respective subspecialty in Universities, Hospital Authority and Department of Health in Hong Kong. All of them are recognized locally, regionally, and/or internationally as the leading experts in clinical genetics. With widespread consultation to all stakeholders and overseas experts, we are able to have a consensus in the current document. With due consideration of workload statistics and training opportunities for trainees, we conclude that there will be one subspecialty board to accredit this programme in Hong Kong. Under the template of subspecialty training programme, there can be more than one training programmes involving different training centres or hospitals. However, the future Hong Kong Children's Hospital will serve as a major hub for clinical and laboratory training.

Programme Description

Clinical Genetics and Genomics (GG) is an internationally well-recognized subspecialty concerned with the diagnosis and management of inherited disorders and birth defects, including the estimation of genetic risks and genetic counselling of family members. Medical geneticists generally work in a multidisciplinary team in close collaboration with laboratory scientists, bioinformaticians, clinical co-workers, genetic counsellors and academic colleagues.

The "Genetics and Genomics [Paediatrics]" (GG[P]) subspecialty training programme offers a 3-year training curriculum in Hong Kong. The programme will be supported by other Colleges under the Hong Kong Academy of Medicine, with trainers from other Colleges helping to provide necessary training to equip a trained specialist with the knowledge, skills and attitudes in clinical genetics to provide high standard of professional service to patients and their families.

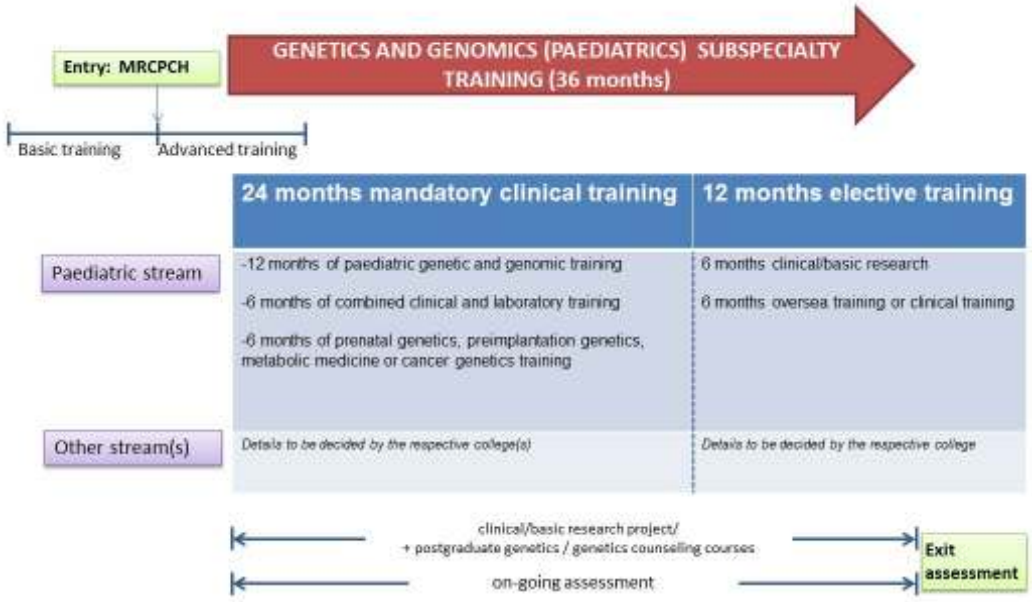
Under this training programme, candidates must have completed 3 years of basic training in General Paediatrics and passed the Joint MRCPCH(UK)/HKCP Intermediate Examination. Candidates will be eligible to commence the subspecialty training in medical genetics during their higher training in General Paediatrics; and a maximum of 1 year is allowed to overlap (the overlapping year), upon the approval of the Subspecialty Board of GG[P]. Applications from candidates seeking academically oriented training with an interest in basic or clinical research are encouraged.

The curriculum is designed to train candidates to provide outstanding clinical care and to develop the fundamental skills to pursue a life-long career in GG[P]. There are 24 months of mandatory clinical training consists of in-patient/out-patient clinical rotations. This include at least 12 months of paediatric genetics, 6 months of combined clinical and laboratory rotation (on either cytogenetics, molecular genetics, bioinformatics or metabolic genetics) and an additional 6 months of clinical training in paediatric genetics, prenatal genetics, preimplantation genetics, metabolic medicine, or cancer genetics.

During the 3rd year the trainee should receive 12 months of elective training that subject to approval by the training director. Six months of training in an overseas tertiary care centre with a recognized programme is strongly recommended. Elective training can be clinical or laboratory or research project-based. Obtaining the

qualification of a postgraduate diploma or degree (e.g. MSc, MPhil, PhD or MD) related to clinical/medical genetics may also be recognized as a completion of elective training for up to a maximum of 6 months subject to approval by the training director.

Training should be competence based that integrates clinical care with small group tutorials, clinical case write-ups, postgraduate courses, journal clubs, clinical/laboratory meetings, interdisciplinary conferences and grand rounds. During the training, a trainee has to pursue either a basic or clinical research project related to medical genetics or genetic counselling. In addition, a trainee is required to perform some administrative duties e.g. clinical audit or quality assurance activities under the supervision of trainers. Attendance of postgraduate training/short courses in genetic counselling/ communication skills, genomics/bioinformatics, molecular biology/developmental biology is strongly encouraged. Supervision is provided by trainers who have achieved stature as clinicians, educators and scientists. Assessment will be in the format of on-going assessment and exit examination (or other modalities).



Mission, Goals and Objectives

The mission of this GG[P] 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.

Specific goals:

Upon completion of training, GG[P] specialists will be able to (a) diagnose and manage genetic disorders; (b) provide genetic counselling to patients and families; (c) apply their 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; and (g) interact with other health care professionals in the provision of services for patients with genetic disorders.

Specific objectives:

At the completion of the GG[P] training, trainees should have mastered the following specific objectives as they pertain to each of the specific goals of the curriculum:-

1. Human genome structure and heredity

Objectives	Specific knowledge and skills	Trainers (by colleges)
Understand the general concepts related to human genome and heredity	a. Describe the information content of the human genome and the elements that predispose to mutation b. Describe the structure of DNA, how it is replicated and maintained (DNA repair mechanism) c. Describe the chromosomal structure of the human genome d. Describe the different stages of cell division in mitosis and meiosis and their medical relevance e. Describe human gametogenesis and fertilization	HKCPaed HKCOG

	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

	parallel sequencing c. Understand the concepts of targeted assays versus mutation scanning methods d. Understand the basic principles of nucleic acid hybridization assays e. Understand the limitations associated with different molecular methods (allele drop-out, primer polymorphisms, large deletion etc.) f. Describe the concept of sample identity testing and use as an adjunct method to establish relationship between samples (e.g. maternal cell contamination, sample identity matching) g. Describe mutations using appropriate nomenclature (e.g. HGVS)	

5. Significance of gene mutations

Objectives	Specific knowledge and skills	Trainers* (by colleges)
Understand the general concepts of pathogenicity of genetic variations	a. Describe different classes of gene mutations (missense, nonsense, frameshift, splicing) and their effect on transcription, translation and protein function b. Ascribe clinical significance to different types of gene variants	HKCPaed HKCOG HKCPath

6. Non-Mendelian inheritance

Objectives	Specific knowledge and skills	Trainers (by colleges)
Understand the principles of non-Mendelian inheritance	a. Describe the different types of non-Mendelian inheritance and their etiologies, including uniparental disomy, imprinting, and mitochondrial inheritance, etc. b. Describe implications of non-Mendelian inheritance on pedigree analysis, diagnostic testing and counselling	HKCPaed

7. Mitochondrial inheritance

Objectives	Specific knowledge and skills	Trainers* (by colleges)
Understand the principles of mitochondrial inheritance	a. Describe the structure and inheritance of the mitochondrial genome and gene expression b. Understand the basis for clinical heterogeneity in mitochondrial DNA defects c. Describe the role of nuclear and mitochondrial genes in mitochondrial disease d. Describe general features of mitochondrial disorders e. Recognize maternal inheritance from pedigree information	HKCPaed HKCPath

8. Pharmacogenomics

Objectives	Specific knowledge and skills	Trainers* (by colleges)
Understand the principles of pharmcogenomics	a. Describe the concept of drug responsiveness risks and benefits based upon genotype	HKCPaed HKCPath HKCPhy

9. Cytogenetics

Objectives	Specific knowledge and skills	Trainers* (by colleges)
Understand the general principles of human cytogenetics as applied to medicine	a. Describe the general principles of cytogenetic methods for chromosome analysis, including karyotyping, genomic copy number assessment, etc. b. Describe appropriate indications for cytogenetic testing c. Distinguish between common cytogenetic variants and pathogenic rearrangements d. Describe general concepts of autosomal and sex chromosomal abnormalities e. Describe the meiotic segregation of rearranged chromosomes and the effects of recombination events f. Describe the general concepts of sex	HKCPaed HKCOG

	chromosomal abnormalities g. Describe parent-of-origin effects and relevance to chromosomal abnormalities h. Describe etiology of chromosome abnormalities (non-disjunction, breakage and repair, non-homologous recombination, uniparental disomy) i. Understand the uses and limitations of cytogenetic tests including the limits of detection of mosaicism j. Understand the effect of mosaicism on phenotype k. Understand use of appropriate nomenclature (e.g. ISCN)	

10. Chromosomal microarray and copy number analysis

Objectives	Specific knowledge and skills	Trainers* (by colleges)
Understand the principles and techniques used in the detection of copy number changes	a. Describe the appropriate indications for copy number analysis. b. Describe the different techniques that can be used to detect copy number changes including chromosomal microarray, FISH, qPCR, MLPA, whole genome sequencing, etc., and describe the limitations of each including the types of mutation detected c. Understand and describe when additional studies are required to complement or confirm microarray results d. Describe the basic principles used to ascribe clinical significance to copy number changes	HKCPaed HKCOG

11. Cancer genetics

Objectives	Specific knowledge and skills	Trainers* (by colleges)
Understand the general principles of human cancer cytogenetics and	a. Explain concepts of multistep pathogenesis of cancers including inherited predisposition, oncogene activation, tumour suppressor	HKCPaed HKCPhy HKCPath

molecular pathology	inactivation, alteration of cell cycle control and DNA repair genes b. Explain and contrast inherited and somatic mutations c. Describe methods to detect gene expression d. Describe principles of recurrent rearrangement detection using molecular or cytogenetic methods e. Describe the relevance of cytogenetic and molecular analysis to cancer diagnosis, prognosis, treatment and monitoring	

12. Biochemical genetics

Objectives	Specific knowledge and skills	Trainers* (by colleges)
Understand broad categories of Inborn error of metabolism	a. Describe the structural and functional relationships of intracellular components: nucleus, Golgi, ER, mitochondria, lysosomes, peroxisomes, etc. b. Describe the different categories of proteins in a cell (structural, enzymes, transport, receptor proteins, etc.), their modes of action and means of regulations c. Describe the biochemical consequences of a primary enzyme block in a metabolic pathway and the way clinical and pathological signs may be produced. d. Describe the major categories of inborn errors of metabolism: amino acid disorders, urea cycle disorders, organic acid disorders, fatty acid oxidation defects, lysosomal storage disorders, mitochondrial disease and peroxisomal disorders e. Understand the principles of newborn screening	HKCPaed HKCPath

13. Complex disorders

Objectives	Specific knowledge and skills	Trainers* (by colleges)
Understand the genetic contribution to complex human disease	a. Describe qualitative and quantitative traits with examples b. Describe the effect of genetic and environmental modifiers on single-gene disorders c. Define the concepts of multifactorial inheritance d. Describe evidence for a genetic contribution to complex traits and common disorders e. Contrast the relative recurrence risks for multifactorial inheritance with single gene disorders and factors that affect risk	HKCPaed HKCCM

14. Population genetics

Objectives	Specific knowledge and skills	Trainers* (by colleges)
Understand the concepts of human population genetics	a. Describe key concepts of human genetic variation in populations, including the role of ethnicity and population isolates in human variation b. Describe the Hardy-Weinberg equilibrium and its application to assess genetic risk c. Understand the concepts of population screening and when appropriate to offer screening	HKCPaed HKCCM

15. Genetic counselling

Objectives	Specific knowledge and skills	Trainers* (by colleges)
Understand the key concepts in genetic counselling and risk assessment	a. Describe common indications for genetic counselling b. Describe the purpose of genetic counselling in specific scenarios c. Describe concepts of counselling: non-directive, awareness of values and biases	HKCPaed HKCOG HKCPhy

Risk assessment and calculations

Objectives	Specific knowledge and skills	Trainers* (by colleges)
Understand the general concepts related to genetic risk calculation and assessment	a. Basic Bayesian analysis: demonstrate ability to modify a priori risk by one conditional factor b. Calculation of odds ratios c. Understand basic test performance characteristics: sensitivity, specificity, positive and negative predictive value	HKCPaed HKCOG HKCCM

16. Developmental genetics and birth defects

Objectives	Specific knowledge and skills	Trainers* (by colleges)
Understand the key steps in human development	a. Understand key concepts in developmental biology as it relates to normal and abnormal human morphogenesis b. Understand the concepts of morphogenesis, differentiation, pluripotency, specification, determination, embryonic induction, competency, and signal transduction c. Describe the processes involved in early embryogenesis: fertilization to gastrulation d. Describe the major embryonic cell lineages and the distribution in the fetus and extra-embryonic tissues e. Understand the contribution of postmortem examination and imaging in syndromal diagnosis	HKCPaed HKCOG HKCPath HKCR

17. Prenatal/ Preimplantation diagnosis

Objectives	Specific knowledge and skills	Trainers* (by colleges)
Understand the principles of prenatal screening, prenatal diagnosis and related methodologies	a. Articulate the principles of a prenatal screening program b. Differentiate the prenatal screening from prenatal diagnoses c. Describe the advantages, disadvantages and limitations associated with prenatal karyotyping,	HKCOG

	and other modes of prenatal molecular diagnoses d. Describe the advantages and disadvantages of amniocentesis and chorionic villus sampling e. Describe the risk, benefits, limitations and controversies surrounding the use of emerging technologies such as (i) Non-invasive prenatal testing (ii) Array CGH on prenatal sample (iii) Exome sequencing on prenatal sample (iv) Preimplantation genetic diagnosis f. Understand the impact of teratogen exposure and maternal disease on fetal development g. Describe genetic factors that contribute to recurrent pregnancy loss and subfertility	

18. Clinical genetics

Objectives	Specific knowledge and skills	Trainers (by colleges)
Understand the broad categories of genetic conditions and their methods of assessment	a. Describe methods of assessment of phenotypic variations, syndrome identification and diagnosis, including generally accessible computer diagnostic aids e.g. OMIM b. Understand the concept of syndrome and be able to give examples of syndromes associated with the following clinical manifestations (i) Dysmorphology (ii) Cancer (iii) Neurogenetic conditions (iv) Cardiac genetic conditions (v) Imprinting disorders (vi) Inborn errors of metabolism (vii) Chromosomal syndromes and genomic disorders characterized by recurrent microdeletion/duplications c. Describe and understand the distinction between genetic screening and genetic testing d. Describe and understand the distinction	HKCPaed

	between genetic testing for the purpose of diagnosis and predictive testing to assess risk for predisposition to monogenic or complex genetic diseases as well as their applications and limitations	

19. Ethics

Objectives	Specific knowledge and skills	Trainers* (by colleges)
Understand the general ethical principles related to the practice of clinical genetics	a. Describe privacy and confidentiality principles as it relates to genetic practice (e.g. communication with health care providers, reporting, database searches) b. Describe ethical issues that relate to genetic testing in childhood c. Describe informed consent and its role in genetic testing d. Describe issues that relate to consenting for genomic analysis e. Explain incidental finding and provide examples from common tests in which an incidental finding may be uncovered f. Outline the implications related to reporting and/or not reporting incidental findings and secondary findings (actively searching for disease-related pathogenic mutations not related to a patient's indication for testing) g. Describe ways to reduce risk of incidental findings h. Describe principles of biobanking	HKCPaed HKCOG HKCCM

*As described in the previous section, a Genetics and Genomics (GG) Working Group was established under the Hong Kong Academy of Medicine to advise and coordinate training and accreditation of clinical genetics and genomics by the respective Colleges. With the difference in training structure, the Working Group concluded that it would be more feasible for individual college to design its own GG curriculum and to decide if clinical genetics is to be recognized as a subspecialty. The Academy will supervise the overall development and help to build and coordinate a

comprehensive, systematic, cross-discipline GG training programme with a list of training resources that can be shared and accredited among the colleges. Training sessions including lectures, seminars, practical trainings, clinical and laboratory rotations shall be made available to trainees under different colleges and these sessions will be offered every year/every other year to allow fellows with different point of entry to structure their individualized training rotation with higher flexibilities.

Methodology for Training in Genetics and Genomics

[Paediatrics]

In order to achieve the goals and objectives for the competence based fellowship program the following experiences have been established for the purpose of teaching GG[P] Fellows. These include 24 months of mandatory clinical training and 12 months of elective training. Other activities include:

- Didactic conferences
- Continuing medical education and participation in professional societies
- Development of teaching skills
- Case reports write-up

(A) Clinical rotation and patient care experience

GG[P] trainees must have the opportunity to manage a number of patients and families sufficient to allow them to develop an understanding of the wide variety of medical genetic problems, including Mendelian disorders, inborn errors of metabolism, chromosomal disorders, multifactorial disorders, syndromes, congenital malformations, other birth defects and other genetically determined conditions. These patients or families may be evaluated in outpatient and inpatient settings. It is expected that at least 12 months in the first 2 years will be spent in the evaluation, treatment and care of patients in the paediatric age group. As clinical genetics and genomics involves families and individuals of all ages, GG[P] trainees must be competent to work not only with children but also adults and must have an opportunity to gain understanding about family dynamics in relation to diagnosis, counselling and management. Therefore, fellows are encouraged to do up to 6 months of elective clinical rotation in prenatal genetics, preimplantation genetics, cancer genetics, etc. Importantly, the development of mature clinical judgment requires that fellows, properly supervised, be given responsibility for patient care commensurate with their ability. This can only be achieved only the fellow is involved in the decision-making process and in the continuity of patient care. GG[P] trainees must be given the responsibility for direct patient care in all settings, including diagnostic and therapeutic planning and management, subject to review and approval by the supervisors. Continuity care experiences must continue throughout this 24 months mandatory clinical training period.

(B) Combined clinical/laboratory rotations and the correlation of laboratory and clinical experience

Experience in cytogenetics, molecular genetics and clinical biochemical genetic laboratories must be integral components of each clinical genetics and genomics program and GG[P] trainees must have regular opportunities to develop their abilities to understand and critically interpret laboratory data. Since copy number variant analysis and next-generation sequencing technologies are already widely used in the clinical setting, the experience to work on genomic "big data" with bioinformaticians is also becoming an essential part of the laboratory experience for GG[P] trainees. Trainees should develop an understanding of the appropriate use of laboratories during diagnosis, counselling and management of patients with genetic disorders. It is expected that 6 months in the mandatory clinical training will be spent in combined clinical and laboratory rotation. There must also be a minimum of 2 continuous weeks in each type of laboratory and GG[P] trainees must not be assigned clinical responsibilities at the same time they are participating in the required laboratory experiences except for their continuity clinic experiences.

(C) Research experience

An active research component must be included within the fellowship programme. A meaningful research experience must be provided with appropriate protected time for each GG[P] trainee to achieve a level of competency to initiate independent clinical or laboratory-based research project. Exposure to research should be initiated early in the fellowship to allow adequate insight into the areas of potential research in preparation for the ultimate selection of his/her research mentor for the remainder of the training programme. The immediate goal of research experience is for the GG[P] trainee to learn sound methodology in designing and performing research studies and the scientific way of interpreting and reporting research data. During this phase of training, the GG[P] trainee will work under close guidance of his/her research mentor for a minimal period of 6 months in the third year of the training.

(D) Didactic conferences

Conference will be held on a regular basis with attendance required of all GG[P] trainees and trainers. At a minimum there should be at least one monthly clinical

case conference and one bimonthly journal club. Basic science or clinical research conference may be held quarterly. Some of these conferences will be organized by trainers from our college while others are provided by other Colleges as part of the cross-discipline genetics and genomics training programme under the Genetics and Genomics Working Group under the Hong Kong Academy of Medicine. Fellows will be required to attend a minimum of 75% of each of the conferences.

(E) Continuing medical education and participation in professional societies

In addition to participating in the organized didactic conferences established within the fellowship programme as well as the cross-discipline genetics and genomics training programme under the Academy, it is also strongly encouraged that all fellows become members of local as well as regional and international human/medical genetics societies. Participation in the continuing medical education (CME) activities of these professional organizations will help to foster the standards of professionalism and augment the process of life-long learning. It is envisaged that the CME activities of these professional societies will form the basis of continuing professional development of the GG[P] specialists

(F) Development of teaching and administrative skills

The programme must cultivate a nurturing environment and ample opportunity to foster activities of learning and at the same time teaching. This includes the education of medical students, physicians, other allied health professionals, patients and families and to a broader sense the society of Hong Kong. Development of these competences requires the fellow to receive instruction and feedback in counselling and communication techniques. Genetic counselling is an integral part of clinical genetics training. Delivering health education talks to professionals and the public at large and contribution/participation of patient support groups will also be encouraged. In addition, a trainee is required to perform some administrative duties e.g. clinical audit, quality assurance activities, diagnostic performance of newly available genetic/genomic tests under the supervision of trainers.

(G) Case report write-up/Dissertations

Details of at least 10 interesting or complicated cases have to be presented. The cases reported need not to be confined to the period of higher and subspecialty training. The description and discussion of each case should be at least 1000 words

excluding the appended references. Examples of reportable cases may include but by no means restricted to those published in local and international peer-reviewed journals (e.g. American Journal of Medical Genetics (AJMG), European Journal of Medical Genetics). These 10 case reports are required as part of training of the curriculum.

For exit examination and assessment, at least 2 presentations (in local or international conferences) and at least 2 dissertations are necessary. One of the dissertations should be at least accepted for publications. The dissertation can be case report, case series, retrospective or prospective study.

Methods of Evaluation

In order for the training programme to achieve its goal and objectives, it is essential to establish an evaluation process incorporating interim and summative assessment of the trainees, and a reciprocal evaluation by the trainees of the programme itself and the trainers.

Interim assessment of the GG[P] trainees

Interim evaluation should occur at the completion of any substantive interaction with a specific trainer or specific rotation. For each rotation, an assessment form will be completed by the supervising trainer. The assessment form utilized is one distributed and recommended by the Hong Kong College of Paediatricians. All trainers must complete the form prior to the completion of rotation and review directly with the fellows. All completed assessment forms are returned to the Training Director for review and placed in the GG trainees' permanent file. These assessment forms are completed every 3 months, or sooner depending on the duration of the rotation. Completed assessment forms submitted to the Training Director are immediately reviewed upon their receipt and any forms with a less than satisfactory rating in any category will require an immediate meeting between the GG[P] trainees and the Training Director to identify causes for the less than satisfactory performance and suggest means for improvement. All trainees will be required to keep a case log book, identifying the patients they have managed and their roles in diagnosis, management, counselling. A copy of this log will be provided to the Training Director 6 monthly. AT least 3-monthly, all trainees will confer individually with the Training Director to review all of their performance. This meeting is to provide feedback to the trainees and to identify areas for enhancement.

Summative assessment of the GG trainees

The overall performance of each GG[P] trainee is reviewed annually by the Subspecialty Board designated Trainee Monitoring Committee comprising the Training Director and 2 subspecialty board members through assessment of the portfolio and a structured interview. This committee is asked to monitor the performance and assess the level of competence of each GG[P] trainee through a detailed and structured interview with specific objectives to attain in different domains. The GG[P] trainee needs to present and discuss the merits of the portfolio

based on his/her training in the past year. The committee's assessment is written and recorded in the programme files for future reference. Any adverse judgments regarding the trainee's performance or competence should be first directed to the Training Director. If the fellow feels that the annual review is not to their satisfaction, then the grievance can be addressed by an established appeal mechanism directed by the College.

Evaluation of the Programme and its trainers

All trainees are required to complete and return a programme and trainers evaluation form once every year. Evaluation forms are collected in a fashion to assure the anonymity of the trainees. GG[P] trainees are encouraged to maintain a high level of communication with the Training Director and trainers. Annual evaluation meetings to be attended by all trainees and subspecialty board members will be established. These meetings can be used to disseminate training information and gather timely feedbacks. The feedbacks received during informal and formal meetings, and the annual evaluation forms will be used to suggest and assist in programme changes.

Final Exit Assessment

The final Exit Assessment normally takes place in June and/or December each year. The GG[P] trainee is to submit a collection of case/case series (see above), at least 2 dissertations and attend a viva examination conducted by an Assessment Board. The Assessment Board comprises of either (i) 3 local Examiners, (ii) 2 local and 1 external examiners or (iii) 3 external examiners, under the approval of programme director. External Assessor who is usually a Programme/Training Director in Clinical Genetics & Genomics from another region, or an overseas expert of renown in Clinical Genetics and Genomics. The external examiner may participate in the viva examination in person or via teleconferencing. College subspecialty board representative is welcomed to sit in and observe the standard of examination or report fault to the subspecialty board but without the right to score candidate or veto. Trainees are expected to defend his/her dissertations and to be asked questions related to his/her GG[P] training. Those GG[P] trainees who are successful at the Exit Assessment will be invited to apply for College Subspecialty Fellowship.

Appendix

Comparison of GG[P] training programmes in different countries

	UK	US	Canada	Australia
College/Board	Joint Royal College of Physicians Training Board	American Board of Medical Genetics	Royal College of Physicians and Surgeons of Canada	Royal Australasian College of Physicians
Total duration of training to become a clinical geneticist	7 years (3+4)	4-5 years (2+2 or 5 for dual program)	5 years (2+3)	6 years (3 + 3)
Prerequisites	Core medical training (MRCP/MRCPCH)	Min. 2 years training in an ACGME-accredited program (med/paeds/OG)	2 years of rotations in med, paeds, mat-fetal medicine, general non genetic counselling, electives	3 years of basic training program under med/paeds
Clinical genetics training	Min. 3 years clinically based training and 1 year max for combination of research or degree study (e.g. MSc in genetics = 6m)	Min. 2 years or Dual board-approved combined training in 5y	3 years of rotations in medical genetics (18m), cytogenetics/molecular genetics/biochemical genetics lab (2m each), elective/research(12m)	3 years (2 years core training + 1 year non-core training)
Assessment for accreditation	work place based assessment and certificate examination of clinical genetics (Royal College of Pathologists)	work place based assessment and ACMG Board examination	work place based assessment and Royal college examination	Supervisor's report, 4 case-based discussion + 4 case reports + 1 research project + an approved university genetic course + tertiary counselling course (recommended)

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