



UNION EUROPÉENNE DES MÉDECINS SPÉCIALISTES EUROPEAN UNION OF MEDICAL SPECIALISTS

Association internationale sans but lucratif

International non-profit organisation

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Syllabus for residents and trainees in Clinical Genetics

This syllabus is an outline and flexible summary of major and specific topics to be covered in some way in the training course of a resident. The basic goal of the syllabus is to help and ensure a fair and impartial understanding between the instructor and students such that there is minimal confusion in the topics, setting clear expectations of material to be learned. The syllabus provides neither a roadmap of course, nor organization/direction relaying the instructor's teaching philosophy to the trainees, as the syllabus is not a learning guide. Rather, the syllabus is a supporting reference material, content and priorities of training may vary in different training institutions.

Domain 1: Theoretical genetics / Basic science

1.1 Cellular and molecular mechanisms that underpin human inheritance

1.1.1 Basics

- 1.1.1.1 Nucleic acid structure, DNA and RNAs
- 1.1.1.2 Translation, protein structure
- 1.1.1.3 Chromosome structure and function (ploidy and cell cycle)
- 1.1.1.4 Monogenic vs. multifactorial inheritance
- 1.1.1.5 Mutations, variants, CNV
- 1.1.1.6 Cells, cell proliferation, cell specialization
- 1.1.1.7 Nuclear and mitochondrial genome
- 1.1.1.8 Gene editing, CRISPR

1.1.2 Genetics of hereditary metabolic disorders

- 1.1.2.1 Classification and molecular basis of metabolic disorders
- 1.1.2.2 Mitochondrial diseases – clinic biochemical and genetic features

1.1.3 Genetics of Malformation, developmental anomalies and rare intellectual disabilities

- 1.1.3.1 Identify normal developmental milestones and diagnose delayed development
- 1.1.3.2 Explain morphogenesis in terms of deformation, malformation, disruption and dysplasia
- 1.1.3.3 Have knowledge of common and rare dysmorphic syndromes
- 1.1.3.4 Genetic causes of intellectual disability (static and progressive)
- 1.1.3.5 Genetic contribution to autism and autistic spectrum disorders

1.1.4 Neurogenetics

- 1.1.4.1 Genetic aspects and clinical presentation of trinucleotide repeat disorders
- 1.1.4.2 Basic neuropathology and differential diagnosis of hereditary dementias

1.1.5 Neuromuscular genetics

- 1.1.5.1 Classification and molecular basis of common genetic neuromuscular disorders

1.1.6 Psychiatric genetics

- 1.1.6.1 Genetic contribution to psychiatric disease in adults

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1.1.7 Cancer Genetics

- 1.1.7.1 The genetic and environmental factors that affect risk of developing cancer
- 1.1.7.2 Current recommendations concerning tumour surveillance in cancer- prone families
- 1.1.7.3 Knowledge of clinical features of genetic cancer syndromes
- 1.1.7.4 Genetic mechanisms in neoplasia
- 1.1.7.5 Knowledge of molecular basis of cancer genetic syndromes
- 1.1.7.6 Knowledge of circulating tumour DNA in cancer detection

1.1.8 Cardiac Genetics

- 1.1.8.1 Knowledge of clinical features of inherited cardiac conditions (ICC) syndromes, including Marfan syndrome and related disorders
- 1.1.8.2 Knowledge of molecular basis of ICC syndromes
- 1.1.8.3 Current recommendations concerning cardiac surveillance in ICC families
- 1.1.8.4 Knowledge of genetic causes of sudden adult death

1.1.9 Skeletal Genetics

- 1.1.9.1 Classification and molecular basis of chondrodysplasia and bone disorders

1.1.10. Ophthalmologic Genetics

- 1.1.10.1 Classification and molecular basis of retinopathies, corneal dystrophies and lens disorders
- 1.1.10.2 Genetic contribution to ocular diseases in children and adults

1.1.11 Endocrine Genetics

- 1.1.11.1 Genetic contribution to endocrine diseases

1.1.12 Gastrointestinal Genetics

- 1.1.12.1 Genetic contribution to gastrointestinal diseases

1.1.13 Hematologic Genetics

- 1.1.13.1 Genetic contribution to hematologic diseases

1.1.14 Dermatologic Genetics

- 1.1.14.1 Classification and molecular basis of skin disorders
- 1.1.14.2 Genetic contribution to dermatologic diseases

1.1.15 Renal Genetics

- 1.1.15.1 Classification and molecular basis of diseases of kidney
- 1.1.15.2 Genetic contribution to kidney diseases

1.1.16 Urogenital Genetics

- 1.1.16.1 Genetic contribution to urogenital diseases

1.1.17 Pulmonary Genetics

- 1.1.17.1 Genetic contribution to pulmonary diseases

1.1.18 Hepatic Genetics

- 1.1.18.1 Genetic contribution to hepatic diseases

1.1.19 Genetics of Immunological and auto-inflammatory diseases

- 1.1.19.1 Genetic contribution to immunological and auto-inflammatory diseases

1.1.20 Connective tissue Genetics

- 1.1.20.1 Classification and molecular basis of connective tissue disorders

1.1.21 Gynaecological and Obstetric Genetics

- 1.1.21.1 Genetic contribution to Gynaecological and Obstetric diseases

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1.1.22 Prenatal and Reproductive Genetics

1.1.22.1 Genetic condition associated with infertility

1.1.23 Genetics of multi-systemic vascular diseases

1.1.23.1 Genetic contribution to multi-systemic vascular diseases

1.1.24 Genetics of craniofacial Anomalies and Ear Nose and Throat disorders

1.1.24.1 Genetic contribution to craniofacial Anomalies and ear nose and throat disorders

1.1.24.2 Classification and molecular basis of deafness

1.2 Embryology and genetics of human development

1.2.1 Understand the natural history of prenatally diagnosed conditions, including common single gene and chromosome abnormalities

1.2.2 Know the indications for and methods of preimplantation and prenatal diagnosis

1.2.3 Be informed of the latest advances in prenatal diagnosis such as testing free foetal DNA in maternal blood and the potential for non-invasive prenatal DNA diagnosis

1.2.4 Knowledge of the law pertaining to termination of pregnancy for foetal abnormality

1.2.5 Know the indications for, process and limitations of foetal post-mortem examination and issues of consent

1.2.6 Have knowledge of national guidelines on retention and storage of foetal tissues and the Human Tissues Act

1.3 Pharmacogenetics / Pharmacogenomics

1.3.1 Knowledge of genetic mechanisms that influence individual drug responses

1.3.2 Knowledge of the rare diseases associated adverse drug effects

1.3.3 new treatments

1.3.4 gene therapy

1.3.5 therapies based on knowledge of genes

1.4 Genetic epidemiology and biostatistics

1.4.1 The genetic characteristics of populations, common gene frequencies and disease prevalence

1.4.2 The factors that influence decisions to instigate programs of population screening for genetic diseases

1.4.3 Define sensitivity, specificity, and predictive values of screening tests. Knowledge of current screening programs

1.4.4 Knowledge of appropriate population-based registers

1.5 Population genetics

1.5.1 Calculate gene frequencies – understand the implications of the Hardy-Weinberg equilibrium

1.6 Risk assessment

1.6.1 Pedigree-based calculation of segregation ratios for structural chromosome abnormalities

1.6.2 Empiric risk calculations (occurrence and recurrence risks)

1.6.3 Perform Bayesian risk calculations including linkage-based risk calculations

1.7 Bioinformatics

1.7.1 Basic methods of medical statistics

1.7.2 Knowledge of the principles of Human Phenotype Ontology

1.7.3 Knowledge in the use of large data sets and “big data”

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1.8 Epigenetics

- 1.8.1 Principles of chromatin regulation
- 1.8.2 Other epigenetic mechanisms, such as miRNA
- 1.8.3 Epigenomics and disease susceptibility

1.9 Multifactorial disorders

- 1.9.1 Understand the genetic factors which influence the incidence and prevalence of common conditions
- 1.9.2 Understand the factors which influence health and illness – psychological, biological, social, cultural and economic especially poverty
- 1.9.3 Understand the influence of lifestyle on health and the factors that influence an individual to change their lifestyle
- 1.9.4 Understand the purpose of screening programmes
- 1.9.5 Understand the positive and negative effects of screening on the individual
- 1.9.6 Demonstrate in practice an appropriate knowledge of the influences of environment and behaviour on health including major factors such as poverty and poor housing, as well as those that might be overlooked

1.10 History of Genetics

- 1.10.1 Major milestones and developments in the history of genetics

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Domain 2: Clinical / Medical knowledge and specialist-level skills

2.1 Patterns of inheritance and methods for risk assessment

2.1.1 Be able to review medical records and identify information sources including databases and literature searches

2.2 Taking a detailed medical and family history and Pedigree construction and interpretation

2.2.1 Be able to take and analyse a clinical history in a relevant, succinct and logical manner

2.2.2 Be able to overcome difficulties of language, physical and mental impairment

2.2.3 Use interpreters and advocates appropriately

2.2.4 Elicit family history information in a sensitive and understanding manner

2.2.5 Draw complex pedigrees accurately, including consanguinity loops, recording appropriate information

2.2.6 Manages time and draws consultation to a close appropriately

2.2.7 Supplements history with standardised instruments or questionnaires when relevant

2.2.8 Manages alternative and conflicting views from family, carers, friends and members of the multi-professional team

2.2.9 Assimilates history from the available information from patient and other sources including members of the multi-professional team

2.2.10 Recognises and interprets appropriately the use of nonverbal communication from patients and carers

2.2.11 Focuses on relevant aspects of history

2.3 Diagnosis, investigation and genetic management of individuals with both common and rare inherited/genetic diseases and their families

2.3.1 Examination

2.3.1.1 Be able to perform a reliable and appropriate examination to elicit relevant signs of genetic disease

2.3.1.2 Perform examination appropriately in situations involving cultural sensitivity

2.3.1.3 Understand when additional specialist examination is required

2.3.1.4 Performs an examination relevant to the presentation and risk factors that is valid, targeted and time efficient

2.3.1.5 Recognises the possibility of deliberate harm (both self-harm and harm by others) in vulnerable patients and report to appropriate agencies

2.3.2 Investigations Including Imaging

2.3.2.1 Ability to prioritise investigations and interpret the results Ability to perform investigations competently where relevant

2.3.2.2 Ability to liaise and discuss investigations with colleagues and to order them appropriately

2.3.3 Diagnosis and Management

2.3.3.1 Present genetic information to a patient in a sensitive and understanding manner

2.3.3.2 Use computerized genetic databases and registers for information retrieval

2.3.3.3 Present undiagnosed cases to colleagues, including dysmorphology club meetings

2.3.3.4 Clearly and openly explain management options

2.3.3.5 Record concisely, accurately, confidentially and legibly the appropriate elements of the history, examination, results of investigations, differential diagnosis and management plan

2.3.4 Time Management

2.3.4.1 Recognise when he/she is falling behind and re-prioritise or ask for help

2.3.4.2 Organise and manage workload effectively and flexibly

2.3.4.3 Make appropriate use of other professionals and support workers

2.3.4.4 Employs techniques for improving time management

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2.3.5 Decision Making

- 2.3.5.1 Interpret clinical features, their reliability and relevance to clinical scenarios including recognition of the breadth of presentation of common disorders
- 2.3.5.2 Incorporates an understanding of the psychological and social elements of clinical scenarios into decision making
- 2.3.5.3 Construct a concise and applicable problem list using available information
- 2.3.5.4 Construct an appropriate management plan in conjunction with the patient, carers and other members of the clinical team and communicate this effectively to the patient, parents and carers securing their agreement to the course of action
- 2.3.5.5 Define the relevance of an estimated risk of a future event to an individual patient
- 2.3.5.6 Use risk calculators appropriately
- 2.3.5.7 Apply quantitative data of risks and benefits of screening and therapeutic intervention to an individual patient
- 2.3.5.8 Search and comprehend medical literature to guide reasoning
- 2.3.5.9 Generate hypothesis within context of clinical likelihood
- 2.3.5.10 Test, refine and verify hypotheses
- 2.3.5.11 Develop problem list and action plan

2.3.6 Ability to take samples for genetic analysis

- 2.3.6.1 Phlebotomy from adults and children, including those with special needs
- 2.3.6.2 Hair Root Extraction
- 2.3.6.3 Skin biopsy
- 2.3.6.4 Collection of other samples, such as buccal smears, urine samples, etc.

2.3.7 Clinical Photography

- 2.3.7.1 Demonstrate ability to take photographs of sufficient quality for clinical use
- 2.3.7.2 Use of digital photography and storage of data

2.4 Therapeutic aspects and emerging therapies of genetic diseases

- 2.4.1 Prescribe and oversee enzyme replacement therapies for applicable disorders, including lysosomal storage disorders within a multidisciplinary clinical team consensus
- 2.4.2 Prescribe other repurposed drugs to specific genetic condition (e.g. losartan) within a multidisciplinary clinical team consensus
- 2.4.3 Develop a management strategy, including preventative surgery, for men and women with hereditary cancer

2.5 Risk assessment and role in genetic testing

- 2.5.1 Calculate genetic risk in single gene disorders by hand
- 2.5.2 Calculate genetic risk by use of a computer programme

2.6 Paediatric genetics including training in Dysmorphology (knowledge of common dysmorphic syndromes, their aetiology and the use of dysmorphology databases) and investigation of learning and intellectual disability in children

- 2.6.1 Be able to take a relevant history, and perform an appropriate examination, obtain illustrative photographs
- 2.6.2 Have a rational approach to investigation of children with delayed development and/or dysmorphic syndromes
- 2.6.3 Formulate differential diagnoses of unknown syndromes Utilise journals and databases used in syndrome identification
- 2.6.4 Cultivate critical assessment of database information and case reports to identify uncertainty and subjectivity in syndrome diagnosis
- 2.6.5 Be able to provide a diagnostic service within a multidisciplinary clinical team
- 2.6.6 Present and discuss cases with colleagues

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2.7 Adult genetics to include knowledge of late onset disorders and disorders with a significant genetic component presenting in adult life (including predictive testing)

- 2.7.1 Be able to take a relevant history, perform an appropriate examination and formulate clinical diagnoses
- 2.7.2 Be able to assess patients and families affected by genetic conditions
- 2.7.3 Judge when it is necessary to sustain supportive relationships with patients with chronic disease
- 2.7.4 Be able to discuss reproductive options (AID, ICSI, IVF, pre-implantation diagnosis) with the patient and their partner in a sensitive manner
- 2.7.5 Be able to discuss and formulate integrated care pathways and management plans with individuals/families
- 2.7.6 Verify diagnoses from old hospital records

2.8 Prenatal Genetics and knowledge about effects of teratogens in foetal development

- 2.8.1 Interpret family history data
- 2.8.2 Provide genetic advice and organize testing for women who may undergo preimplantation or prenatal diagnosis
- 2.8.3 Formulate differential diagnoses and assess prognosis in collaboration with the foetal medicine team
- 2.8.4 Assess risk to foetus when pregnancies are exposed to hazards such as congenital infections, alcohol, ionising irradiation or drugs
- 2.8.5 Assess clinical significance of chromosome, DNA and foetal imaging in the context of foetal abnormality
- 2.8.6 Evaluate foetal post-mortem findings
- 2.8.7 Interpret the reports of non-invasive prenatal testing (NIPT)

2.9 Genetic screening programmes

- 2.9.1 Team-working with database managers, genetic associates and nurse specialists in:
 - 2.9.1.1 ‘Cascade screening’ and provision of genetic services for extended families with common single gene disorders (cystic fibrosis, Xp21 muscular dystrophy, fragile X syndrome, Huntington’s disease)
 - 2.9.1.2 Family based screening for individuals at high risk of developing cancer
 - 2.9.1.3 Contribute to the maintenance of departmental genetic register systems
 - 2.9.1.4 Be able to explain the benefits and consequences of screening programmes

2.10 Examination of paediatric and adult patients, knowledge of dysmorphic signs, and main neurologic signs

- 2.10.1 Physical examination, body measurements and review of medical information

2.11 Gene therapy and its current and future applications

- 2.11.1 Be able to discuss the pros and cons of gene therapy in relation to a specific disorder and suggest clinical trials, if appropriate

2.12 Common diseases with a genetic component and oligo-/polygenic disorders

- 2.12.1 Be able to distinguish between classical Mendelian and oligogenic inheritance and be able to calculate the appropriate recurrence risk
- 2.12.2 Be able to recognize and counsel patients with a strong genetic component

2.13 Special areas of Clinical Genetics including

- 2.13.1 Inherited metabolic disorders
 - 2.13.1.1 Recognise family history data that suggest familial metabolic disease
 - 2.13.1.2 Be able to confirm clinical signs in affected individuals
 - 2.13.1.3 Be able to draw up a differential diagnosis and institute appropriate genetic testing
 - 2.13.1.4 Assessment of symptoms and signs in patients at risk of metabolic disorders
 - 2.13.1.5 Make timely, appropriate referrals to other specialists
 - 2.13.1.6 Identify at-risk patients and relatives who are eligible to therapeutic and preventional strategies

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2.13.2 Neurogenetics and neuromuscular genetics

- 2.13.2.1 Recognise family history data that suggest familial neurological disease
- 2.13.2.2 Be able to confirm clinical signs in affected individuals
- 2.13.2.3 Be able to draw up a differential diagnosis and institute appropriate genetic testing
- 2.13.2.4 Assessment of symptoms and signs in patients at risk of adult-onset neurogenetic disease
- 2.13.2.5 Application of protocols for pre-symptomatic diagnosis of Huntington's disease and other neurodegenerative disorders
- 2.13.2.6 Make timely, appropriate referrals to other specialists such as neurologists, psychologists, psychiatrists, speech therapists

2.13.3 Cardiovascular genetics

- 2.13.3.1 Be able to take a relevant history, perform an appropriate examination
- 2.13.3.2 Work with bereaved families following sudden adult death
- 2.13.3.3 Use of Ghent criteria for diagnosing Marfan syndrome
- 2.13.3.4 Assessment of screening protocols for at-risk relatives
- 2.13.3.5 Coordinate diagnostic and predictive genetic testing in ICC families
- 2.13.3.6 Identify at-risk patients/trios eligible to participate in prevention strategies (e.g. therapeutic trials)

2.13.4 Cancer genetics

- 2.13.4.1 Be able to take a relevant history, perform an appropriate examination and undertake risk estimation using a variety of methods
- 2.13.4.2 Use of cancer registers and other sources to verify diagnoses
- 2.13.4.3 Use disease registers to support follow-up of affected and at-risk patients
- 2.13.4.4 Assessment of screening protocols for at-risk relatives
- 2.13.4.5 Identify at-risk patients and relatives who are eligible to participate in trials of cancer prevention strategies
- 2.13.4.6 Rare cancers; differences and similarities with rare diseases. Types (classification: Pediatric cancers, Haematologic rare neoplasms; Sarcomas; Rare thoracic cancers; Neuroendocrine tumours; Head & neck cancers; Central nervous system tumours; Rare female genital cancers; Rare urological and male genital tumours; Endocrine gland tumours; Digestive rare cancers; Rare skin cancers & non-cutaneous melanoma)

2.13.5 Reproductive genetics

- 2.13.5.1 Be able to provide preconceptional genetic counselling to couples with sub-/infertility and organize genetic testing
- 2.13.5.2 Be able to provide preconceptional genetic counselling to couples with genetic and inherited disorders for their reproductive choices including invasive diagnosis, non-invasive testing, and assisted reproductive technologies (ART)
- 2.13.5.3 Be able to inform on the different ART options according to the national legislation and European guidelines including preimplantation genetic testing

2.13.6 Skeletal Genetics

- 2.13.6.1 Be able to formulate a differential diagnosis of a fetus suspected of having a skeletal dysplasia or dysostosis and assess whether the condition is compatible with postnatal survival
- 2.13.6.2 Be able to formulate a differential diagnosis for a child with a congenital limb, axial, and/or craniofacial malformation, including teratogenic causes, syndromic causes, and skeletal dysostoses/dysplasias
- 2.13.6.3 Be able to evaluate radiographs and other imaging studies and know when to order further biochemical or molecular genetic tests, as well as which tests are appropriate for a given situation

2.13.7 Ophthalmologic Genetics

- 2.13.7.1 Be able to formulate a differential diagnosis for a child with microphthalmia/ anophthalmia/ coloboma with or without a congenital anomaly of the central nervous system
- 2.13.7.2 Be able to delineate ocular from oculocutaneous albinism
- 2.13.7.3 Be able to apply diagnostic criteria to establish the diagnosis of various genetic syndromes with supporting ophthalmologic features
- 2.13.7.4 Clinical trials in gene-replacement treatment strategies for childhood heritable retinal dystrophies

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2.13.8 Endocrine Genetics

- 2.13.8.1 Be able to formulate a differential for a child with short stature
- 2.13.8.2 Be able to evaluate a child with ambiguous genitalia, and formulate differential diagnosis
- 2.13.8.3 Be able to recognize Albright's hereditary osteodystrophy
- 2.13.8.4 Be able to counsel families with a child with 21-hydroxylase deficiency and adults with infertility, including Klinefelter syndrome, mosaic Turner syndrome, and androgen insensitivity syndrome.
- 2.13.8.5 Be able to formulate a differential diagnosis for sex reversal
- 2.13.8.6 Be able to counsel families with multiple endocrine neoplasia (MEN) I or II
- 2.13.8.7 Be able to evaluate the child with thyroid abnormalities and hearing loss

2.13.9 Gastrointestinal Genetics

- 2.13.9.1 Be able to provide a differential diagnosis for congenital anomalies such as intestinal aganglionosis, pyloric stenosis, intestinal malrotation, etc.
- 2.13.9.2 Formulate a differential diagnosis for patients with hereditary pancreatitis
- 2.13.9.3 Recognize the need for a cancer control plan for extra intestinal cancers in polyposis syndromes (e.g., breast cancer in Peutz-Jeghers syndrome)
- 2.13.9.4 Recognize that meconium ileus and malabsorption are features of cystic fibrosis

2.13.10 Hematologic Genetics

- 2.13.10.1 Recognize stigmata of Fanconi anemia
- 2.13.10.2 Evaluate genetic causes of familial neutropenia syndromes (e.g., cyclic or severe congenital neutropenia, and Shwachman-Diamond syndrome), and disorders of neutrophil function (e.g., chronic granulomatous disease)
- 2.13.10.3 Differential diagnosis for genetic red cell membrane disorders such as hereditary spherocytosis
- 2.13.10.4 Diagnose and counsel patients with sickle cell trait, beta thalassemia trait, and the various forms of alpha thalassemia trait
- 2.13.10.5 Plan laboratory assessments for pregnant women with microcytic anemia
- 2.13.10.6 Counsel families with hemoglobinopathy

2.13.11 Dermatologic Genetics

- 2.13.11.0 Be able to formulate a differential diagnosis for a patient with an ichthyosiform disorder
- 2.13.12.0 Recognize the features of skin fragility and blistering associated with epidermolysis bullosa
- 2.13.13.0 Formulate a differential diagnosis for a patient with abnormal ectodermal structures (hair, teeth, nails, sweat glands)
- 2.13.14.0 Formulate a differential diagnosis for a patient with premature aging, photosensitivity, vascular lesions or multiple cutaneous neoplasms or hamartomas
- 2.13.15.0 Order appropriate genetic testing for suspected genodermatoses
- 2.13.16.0 Recognize the cutaneous features that are associated with multisystem disorders

2.13.12 Nephrologic Genetics

- 2.13.12.1 Provide genetic counselling for an individual who has or is at risk for infantile or adult polycystic kidney disease
- 2.13.12.2 Recognize the genetic aetiologies that contribute to nephrotic and renal tubular disorders
- 2.13.12.3 Recognize the differential diagnosis between Alport syndrome and other renal disorders
- 2.13.12.4 Apply diagnostic criteria to establish diagnosis of disorders including Bardet-Biedl syndrome, tuberous sclerosis complex, von Hippel-Lindau syndrome, Meckel syndrome, Zellweger syndrome

2.13.13 Urogenital Genetics

- 2.13.13.1 Be able to formulate a differential diagnosis for a child with a congenital anomaly of the urogenital tract

2.13.14 Pulmonary Genetics

- 2.13.14.1 Formulate a differential diagnosis, for hereditary pulmonary emphysema
- 2.13.14.2 Formulate a differential diagnosis, for idiopathic pulmonary hypertension
- 2.13.14.3 Counsel patients with idiopathic pulmonary fibrosis
- 2.13.14.4 Counsel families with or at risk for cystic fibrosis
- 2.13.14.5 Counsel patients with alpha-1-antitrypsin deficiency

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- 2.13.15 Hepatic Genetics
 - 2.13.15.1 Formulate a differential diagnosis for patients with biliary atresia or arteriohepatic dysplasia
 - 2.13.15.2 Counsel families with hepatic disorders
- 2.13.16 Genetics of Immunological and auto-inflammatory diseases
 - 2.13.16.1 Formulate a differential diagnosis for a child with severe combined immune deficiency
 - 2.13.16.2 Formulate a differential diagnosis for a patient with hypogammaglobulinemia
 - 2.13.16.3 Formulate a differential diagnosis for a patient with chronic granulomatous disease
 - 2.13.16.4 Recognize the signs of hereditary angioedema
 - 2.13.16.5 Formulate a differential diagnosis for an adult with auto-inflammatory disease
- 2.13.17 Connective tissue Genetics
 - 2.13.17.1 Conduct a physical examination appropriate for evaluation of an individual with a suspected connective tissue disorder, including appropriate body measurements (arm span, upper/lower segment ratios, Beighton score, arachnodactyly, hindfoot valgus, pes planus, pectoral abnormalities, etc.)
 - 2.13.17.2 Formulate a differential diagnosis for a patient with joint laxity
 - 2.13.17.3 Formulate a differential diagnosis for a patient with Marfanoid habitus
 - 2.13.17.4 Formulate a differential diagnosis for a patient with aortic dilatation using family history, medical history, and physical examination
 - 2.13.17.5 Apply diagnostic criteria to establish a diagnosis of Loeys-Dietz syndrome, including use of imaging (such as evidence of vascular tortuosity)
 - 2.13.17.6 Establish the specific type of EDS based on diagnostic criteria
 - 2.13.17.7 Apply clinical and laboratory criteria to establish a diagnosis of Stickler syndrome
- 2.13.18 Gynecological and Obstetric Genetics
 - 2.13.18.1 Recognize the stages of embryonic development and their relationship to teratogenic windows in the context of maternal teratogens such as alcohol, medications, or viral exposures
 - 2.13.18.2 Recognize the range of normal variation in foetal ultrasound images, the associations of normal variants with and the limitations of ultrasound as a screening modality
 - 2.13.18.3 Counsel and initiate the appropriate prenatal genetic tests when a structural malformation and/or growth abnormality is identified by foetal ultrasound
- 2.13.19 Genetics of multi-systemic vascular diseases
 - 2.13.19.1 Formulate a differential diagnosis for multi-systemic vascular diseases
- 2.13.20 Genetics of craniofacial Anomalies and Ear, Nose and Throat disorders
 - 2.13.20.1 Formulate a differential diagnosis for craniofacial Anomalies and ear nose and throat disorders
 - 2.13.20.2 Formulate a differential diagnosis in new-borns identified with congenital deafness either through new-borns screening or clinically
 - 2.13.20.3 Interpret audiologic tests and distinguish different patterns of hearing impairment, including sensorineural and conductive
 - 2.13.20.4 Formulate a management plan for a child or an adult with congenital or progressive hearing impairment
- 2.13.21 Genetics of malformation, developmental anomalies and rare intellectual disabilities
 - 2.13.21.1 Determine if a congenital anomaly represents a malformation, deformation, disruption, or dysplasia
 - 2.13.21.2 Explain the difference between a syndrome, sequence, and association
 - 2.13.21.3 Explain congenital anomalies in terms of dysfunction of normal development, both at the level of the embryo and at the level of cellular mechanisms of morphogenesis
 - 2.13.21.4 Explain how foetal exposures/environment can adversely affect foetal growth and/or development
 - 2.13.21.5 Explain how prenatal studies can facilitate diagnostic evaluation
 - 2.13.21.6 Define developmental milestones and growth parameters and recognize patterns of abnormal development
 - 2.13.21.7 Formulate a differential diagnosis and testing strategy for a patient with one or more major anomalies
 - 2.13.21.8 Recognize the specific patterns of dysmorphic features that allow for clinical diagnosis of recognizable genetic conditions
 - 2.13.21.9 Formulate a differential diagnosis for a patient with hypotonia and dysmorphic features
 - 2.13.21.10 Formulate a differential diagnosis for a patient with disordered growth

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- 2.13.21.11 Formulate a differential diagnosis for a patient with autism and dysmorphic features
- 2.13.21.12 Apply diagnostic criteria to establish diagnosis of congenital anomaly syndromes

2.13.22 Psychiatric genetics

- 2.13.22.1 Formulate a genetic differential diagnosis based on DSM criteria
- 2.13.22.2 Recognize that some disorders, including Huntington disease, metachromatic leukodystrophy, some forms of porphyria, and Wilson disease may present with psychiatric symptomatology before other symptoms.
- 2.13.22.3 Be able to diagnose, manage and counsel individuals with these disorders
- 2.13.22.4 Be aware that inborn errors of metabolism, particularly syndromes elevating ammonia levels, may be associated with altered behaviours that are symptomatic of acute decompensation
- 2.13.22.5 Apply knowledge of the features, consequences, and guidelines for management of foetal alcohol syndrome and foetal alcohol spectrum disorder (FASD)
- 2.13.22.6 Evaluate syndromic aetiologies based on presentation, including sex and age of onset of symptomatology
- 2.13.22.7 Recognize the cardinal features and implement management recommendations for microdeletion syndromes associated with behavioural psychopathology as a primary or major component
- 2.13.22.8 Recognize that poorly controlled metabolic disorders often have prominent psychiatric consequences

Domain 3: Genetic counselling and communication skills

3.1 Training in genetic counselling for all types of genetic disease and genetics-related situations encountered in clinical genetics practice. This includes pre- and post-testing counselling in relation to reproductive genetic diagnosis, prenatal diagnosis and for late onset such as neurogenetic and cancer genetic disorders, including predictive testing. Where applicable, training in co-counselling with other professionals such as genetic counsellors and specialists in other fields of medicine

- 3.1.1 Use of “non-directive” counselling skills
- 3.1.2 Effective use of co-counsellors
- 3.1.3 Communication of genetic information and risk to children and adolescents
- 3.1.4 Communication with adults and children with learning disability
- 3.1.5 Recognising which patients will benefit from referral on to psychological services

3.2. Understanding and handling emotional reactions and family crises in relation to the genetic diagnostic process

- 3.2.1 Be able to break bad news in steps appropriate to the understanding of the individual and be able to support distress
- 3.2.2 Demonstrate to others good practice in breaking bad news
- 3.2.3 Recognises the impact of the bad news on the patient, carer, supporters, staff members and self
- 3.2.4 Encourage questioning and ensure comprehension Respond to verbal and visual cues from patients and relatives
- 3.2.5 Act with empathy, honesty and sensitivity avoiding undue optimism or pessimism
- 3.2.6 Structures the interview inappropriately

3.3. Understanding ethical and legal issues and importance of consent and confidentiality

3.3.1 Principles of Medical Ethics and Confidentiality

- 3.3.1.1 Demonstrate knowledge of the principles of medical ethics Outline and follow the guidance given by the GMC on confidentiality
- 3.3.1.2 Define the provisions of the Data Protection Act and Freedom of Information Act
- 3.3.1.3 Define the principles of Information Governance
- 3.3.1.4 Define the role of the Caldicott Guardian and Information Governance lead within an institution, and outline the process of attaining Caldicott approval for audit or research
- 3.3.1.5 Outline situations where patient consent, while desirable, is not required for disclosure e.g. serious communicable diseases, public interest

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- 3.3.1.6 Outline the procedures for seeking a patient's consent for disclosure of identifiable information
- 3.3.1.7 Recall the obligations for confidentiality following a patient's death
- 3.3.1.8 Recognise the problems posed by disclosure in the public interest, without patient's consent
- 3.3.1.9 Recognise the factors influencing ethical decision making: including religion, personal and moral beliefs, cultural practices
- 3.3.1.10 Outline the principles of the Mental Capacity Act
- 3.3.2 Informed consent
 - 3.3.2.1 Know the process for gaining informed consent
 - 3.3.2.2 Understand process of consent for tissue/sample storage and use
 - 3.3.2.3 How to gain consent for a research project
 - 3.3.2.4 Outline the guidance given by the GMC on consent
- 3.3.3 Legal Framework for Practice
 - 3.3.3.1 All decisions and actions must be in the best interests of the patient
 - 3.3.3.2 Understand sources of medical legal information
 - 3.3.3.3 Understand disciplinary processes in relation to medical malpractice
- 3.4 Development of good communication skills with patients and families, colleagues in genetic centres and other specialists and healthcare professionals**
 - 3.4.1 Within a Consultation
 - 3.4.1.1 Be able to communicate effectively, both verbally and in writing to patients whose first language may not be that of the consultant in a manner that they understand
 - 3.4.1.2 Give clear information and feedback to patients and share information with relatives when appropriate
 - 3.4.1.3 Establish a rapport with the patient and relevant others
 - 3.4.1.4 Utilise open and closed questioning appropriately
 - 3.4.1.5 Listen actively and question sensitively to guide the patient and to clarify information
 - 3.4.1.6 Identify and manage communication barriers, tailoring language to the individual patient and others and using interpreters when indicated
 - 3.4.1.7 Deliver information compassionately, being alert to and managing their and your emotional response (anxiety, antipathy etc.)
 - 3.4.1.8 Use, and refer patients to, appropriate written and other evidence based information sources
 - 3.4.1.9 Check the patient's/carer's understanding, ensuring that all their concerns/questions have been covered
 - 3.4.1.10 Indicate when the consultation nearing its end and conclude with a summary and appropriate action plan; ask the patient to summarise back to check his/her understanding
 - 3.4.2 Complaints
 - 3.4.2.1 Manage dissatisfied patients / relatives
 - 3.4.2.2 Contribute to processes whereby complaints are reviewed and learned from
 - 3.4.2.3 Explain comprehensibly to the patient the events leading up to a medical error or serious untoward incident, and sources of support for patients and their relatives
 - 3.4.2.4 Deliver an appropriate apology and explanation (either of error or for process of investigation of potential error and reporting of the same)
 - 3.4.2.5 Distinguish between system and individual errors (personal and organisational)
 - 3.4.2.6 Show an ability to learn from previous error
 - 3.4.2.7 Recognise when something has gone wrong and identify appropriate staff to communicate this with
 - 3.4.3 Stress & Personal Health
 - 3.4.3.1 Develop appropriate coping mechanisms for stress and ability to seek help if appropriate
 - 3.4.3.2 Demonstrate the ability to recognise the manifestations of stress on self and others and know where and when to look for support
 - 3.4.3.3 Personal and professional roles and responsibilities. Prioritise tasks, having realistic expectations of what can be completed by self and others

Domain 4. Laboratory skills

4.1 Thorough knowledge of principles of classic laboratory techniques used in genetic diagnostic testing

- 4.1.1 Techniques for banding chromosome analysis in different tissues
- 4.1.2 Laboratory techniques and application of new cytogenetic tests e.g. FISH/CGH
- 4.1.3 Use of ISCN nomenclature
- 4.1.4 Molecular genetic techniques in common usage – (DNA extraction, PCR, DNA sequencing)
- 4.1.5 Application of DNA-based testing, for linkage and mutation detection
- 4.1.6 Potential application of new DNA and RNA technologies, and proteomics
- 4.1.7 Sensitivity and specificity of laboratory tests
- 4.1.8 Investigative approach to biochemical diagnosis of inborn errors of metabolism (via experience gained at metabolic disease clinics)
- 4.1.9 The operation of local and national antenatal and newborn genetic disease screening programmes

4.2 Thorough knowledge of the new laboratory methods used in genetic diagnostic testing including chromosomal microarrays, whole genome sequencing and exome sequencing

- 4.2.1 Skill on chromosomal microarrays technology including SNP and CGH arrays
- 4.2.2 Skill on next generation sequencing technologies: gene panels
- 4.2.3 Skill on exome sequencing data and report
- 4.2.4 Skill on whole genome sequencing data and report

4.3 Interpretation of results from cytogenetic, molecular genetic, biochemical genetic and genomic analyses including array, exome and genome sequencing

- 4.3.1 Interpretation of clinical consequences of abnormal karyotypes, enzyme deficiencies and molecular test results
- 4.3.2 Liaise with molecular and cytogenetics scientists in analysis of test results
- 4.3.3 Provide advice to laboratory on the wording of reports to referring clinicians
- 4.3.4 Genetic risk calculation based on laboratory test results (e.g. MLINK, Bayesian analysis)
- 4.3.5 Interpretation and re-interpretation during the time of results of exome and genome sequencing
- 4.3.6 Provide counselling to individuals regarding the application of whole genome or whole exome sequencing
- 4.3.7 Explain to an individual contemplating whole exome or genome analysis the potential risks, benefits and limitations of the information that will be obtained and facilitate informed decision-making
- 4.3.8 Prioritize the information obtained from whole exome or genome analysis, including carrier status for recessive disorders, single gene disorders, pharmacogenetic traits, and alleles that confer risk of common disease, in providing feedback and counselling
- 4.3.9 Describe potential risks and benefits that may be associated with disclosing risks of adult-onset disorders in children
- 4.3.10 Utilize genomic databases and bioinformatics tools to filter results on genetic variants and assess their clinical significance
- 4.3.11 Explain the difference between variants of known clinical significance and variants of unknown clinical significance in providing counselling on whole exome or genome analysis
- 4.3.12 Explain the concepts of odds ratio and relative and absolute risk, and the limitations in interpretation of genotypic data regarding risk of common disease

4.4 The time spent and the practical expertise gained in laboratory work may vary between countries, but it should be sufficient to ensure highly specialized knowledge in the laboratory diagnostics (see also the ETR)

4.5 Knowledge of pre-analytical handling of samples and logistics

4.6 Awareness of quality issues in genetic testing

4.7 Knowledge of international nomenclature systems used in genetic reporting

4.8. Clinical Utility Gene Cards

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Domain 5. Ancillary competences

5.1 Maintaining Good Medical Practice

- 5.1.1 Develop a commitment to lifelong learning through continuing professional development and attend relevant courses and conferences
- 5.1.2 Evidence-Based Medicine
- 5.1.3 Able to critically appraise evidence
- 5.1.4 Ability to be competent in the use of databases, libraries and the internet
- 5.1.5 Able to discuss the relevance of evidence with individual patient
- 5.1.6 Ability to search the medical literature including use of PubMed, Medline, Cochrane reviews and the internet
- 5.1.7 Appraise retrieved evidence to address a clinical question
- 5.1.8 Apply conclusions from critical appraisal into clinical care
- 5.1.9 Identify the limitations of research
- 5.1.10 Contribute to the construction, review and updating of local (and national) guidelines of good practice using the principles of evidence based medicine
- 5.1.11 Participate in Audit and Clinical Governance
- 5.1.12 Clinical Governance
- 5.1.13 Be an active partaker in clinical governance
- 5.1.14 Assess and analyse situations, services and facilities in order to minimise risk to patients and the public
- 5.1.15 Audit
- 5.1.16 Involvement in on-going audit
- 5.1.17 Undertake at least one audit project
- 5.1.18 Design, implement and complete audit cycles
- 5.1.19 Contribute to local and national audit projects as appropriate (e.g. NCEPOD, SASM)
- 5.1.20 Support audit by junior medical trainees and within the multi- disciplinary team
- 5.1.21 Patient Safety
- 5.1.22 Maintain a portfolio of information and evidence, drawn from your medical practice
- 5.1.23 Reflect regularly on your standards of medical practice in accordance with GMC guidance on licensing and revalidation
- 5.1.24 Practise with attention to the important steps of providing good continuity of care
- 5.1.25 Accurate attributable note-keeping including appropriate use of electronic clinical record systems
- 5.1.26 Demonstrate leadership and management in the education and training of junior colleagues and other members of the healthcare team
- 5.1.27 Lead and participate in interdisciplinary team meetings Provide appropriate supervision to less experienced colleagues
- 5.1.28 Adhere to accepted consent and confidentiality procedures
- 5.1.29 Timely management of medical documentation and communication with patients families and professionals
- 5.1.30 Educating Patients About Disease, Investigations and Management
- 5.1.31 Identify opportunities to promote changes in lifestyle and other actions which will positively improve health and/or disease outcomes
- 5.1.32 Identify the interaction between mental, physical and social wellbeing in relation to health
- 5.1.33 Counsel patients appropriately on the benefits and risks of screening and health promotion activities
- 5.1.34 Identify patient's ideas, concerns and health beliefs regarding screening and health promotions programmes and be capable of appropriately responding to these

5.2 Information technology (IT) skills

- 5.2.1 Use of information technologies including online resources and databases related to human genetics
 - 5.2.1.1 Demonstrate competent use of databases and statistics programmes
 - 5.2.1.2 Undertake effective literature searches
 - 5.2.1.3 Access genetic web sites and specialist databases to undertake searches
 - 5.2.1.4 Produce effective computer assisted presentations
 - 5.2.1.5 Demonstrate competent use of tools for interpretation of exome and genome sequencing

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5.3 Ethics and law

5.3.1 Understand ethical and legal issues

5.3.1.1 Continuity of Care

5.3.1.1.1 Make adequate arrangements to cover leave

5.3.1.1.2 Practise with professionalism including integrity, compassion, altruism, continuous improvement, aspiration to excellence, respect of cultural and ethnic diversity, and with regard to the principles of equity

5.3.1.1.3 Work in partnership with patients and members of the wider healthcare team

5.3.1.1.4 Liaise with colleagues to plan and implement work rotas

5.3.1.1.5 Promote awareness of the doctor's role in utilising healthcare resources optimally and within defined resource constraints

5.3.1.1.6 Recognise and respond appropriately to unprofessional behaviour

5.3.1.1.7 Be able to handle enquiries from the press and other media effectively

5.3.1.1.8 Eliminate discrimination against patients from diverse backgrounds including age, gender, race, culture, disability and sexuality

5.3.1.1.9 Doctor-Patient Relationship

5.3.1.1.10 Develop a relationship that facilitates solutions to patient's problems

5.3.1.1.11 Deal appropriately with behaviour falling outside the boundary of the agreed doctor patient relationship in patients, e.g. aggression, violence, sexual harassment

5.3.1.1.12 Develop a self-management plan with the patient

5.3.1.1.13 Support patients, parents and carers where relevant to comply with management plans

5.3.1.1.14 Encourage patients to voice their preferences and personal choices about their care

5.3.1.1.15 Use assessment, appraisal, complaints and other feedback to discuss and develop an understanding of own development needs

5.3.1.2 Recognising Own Limitations

5.3.1.2.1 Know the extent of one's own limitations and the limitations of self-professional competence and know when to ask for advice

5.3.1.2.2 Recognise that personal beliefs and biases exist and understand their impact (positive and negative) on the delivery of health services

5.4 Biobanking

5.4.1 Understand principals of biobanking

5.4.2 Awareness of ELSI issues

5.5 Management training

5.5.1 Knowledge about general healthcare policy, goals and priorities

5.5.2 Understanding the organization and management of genetic services

5.5.3 Opportunities to participate in departmental/service activities related to organizational planning, financial management, and monitoring and maintaining quality standards

5.6 Development of multidisciplinary team working and leadership skills

5.6.1 Show leadership, delegate and supervise safely

5.6.2 Be able to communicate effectively

5.6.3 Recognise when input from another specialty is required for individual patients

5.6.4 Be able to work effectively with GPs, other medical and surgical specialists and other health care professionals

5.6.5 Behavioural management skills with colleagues to prevent and resolve conflict and enhance collaboration

5.6.6 Demonstrate the ability to facilitate, chair, and contribute to meetings

5.6.7 Prepare for meetings - reading agendas, understanding minutes, action points and background research on agenda items

5.6.8 Maintain and routinely practice critical self-awareness, including able to discuss strengths and weaknesses with supervisor, recognising external influences and changing behaviour accordingly

5.6.9 Create open and non-discriminatory professional working relationships with colleagues awareness of the need to prevent bullying and harassment

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5.6.10 Develop effective working relationships with colleagues and other staff through good communication skills , building rapport and articulating own view

5.6.11 Communicate effectively in the resolution of conflicts, and identifying and rectifying team dysfunction

5.7 Teaching

5.7.1 Develop teaching skills by participating in the education and training of various categories of staff

5.7.2 Be able to critically evaluate relevant educational literature

5.7.3 Vary teaching format and stimulus, appropriate to situation and subject

5.7.4 Provide effective feedback and appropriate after teaching, and promote learner reflection

5.7.5 Conduct developmental conversations as appropriate eg: appraisal, supervision, mentoring

5.7.6 Demonstrate effective lecture, presentation, small group and bed side teaching sessions

5.7.7 Participate in strategies aimed at improving patient education e.g. talking and listening at support group meetings

5.7.8 Be able to lead departmental teaching programmes including journal clubs

5.8 Involvement with patient groups, and patient/family education

5.8.1 Managing Long-Term Conditions and Promoting Patient Self-Care

5.8.2 Develop and agree a management plan with the patient (and carers), ensuring comprehension to maximise self-care within care pathways where relevant

5.8.3 Develop and sustain supportive relationships with patients with whom care will be prolonged and potentially life long

5.8.4 Promote and encourage involvement of patients in appropriate support networks, both to receive support and to give support to others

5.8.5 Encourage and support patients in accessing appropriate information

5.9 Supplementary to “ Education and Training”

5.9.1 Subspecialty training: Some trainees will elect to develop expertise in a subspecialty area such as cancer genetics, dysmorphology, neurogenetics, etc. This may also vary from country to country.

5.9.2 Knowledge and understanding of the principles of evidence-based medicine

5.9.3 Involvement and initiatives in courses programmes and social issues related to rare diseases.

5.9.4 Knowledge of patient registries, patient support organisations

5.10 Quality Assurance

5.10.1 Knowledge, skills and attitudes should form the basis of the training programme

5.10.2 A written agreed curriculum for the training period should be set up as a contract between the trainee and the supervisor if not otherwise determined by national regulations

5.10.3 Trainees should maintain a Training Logbook including details of clinical and laboratory experience, educational activities, research and publications

5.10.4 A mechanism should be in place for continuous assessment of trainees against agreed quality standards. Some countries will have a nationally prescribed system for assessment and certification

5.10.5 Specialist examination may be compulsory in some countries

5.10.6 EU-Certification in Clinical Genetics should be encouraged

5.11 Research

5.11.1 Medical genetics has a rapidly changing knowledge base and during specialty training the clinical/medical geneticist should be encouraged to participate in research. Some trainees may wish to participate in scientific projects and research leading to a higher academic degree. On completion of training some academic clinical/medical geneticists will continue to lead research programmes whilst many others will collaborate with laboratory based colleagues in the genetics team

5.11.2 Understand the principles of research methodology including clinical trials

5.11.3 Undertake systematic critical review of scientific literature

5.11.4 Ability to frame questions to be answered by a research project

5.11.5 Develop protocols and methods for research

5.11.6 Participate in collaborative research with clinical/scientific colleagues

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5.11.7 Be able to accurately analyse data

5.11.8 Write and submit a case report or original scientific research paper

5.11.9 Develop critical appraisal skills and apply these when reading literature