Co-funded by the Tempus Programme of the European Union

MEDGEN

Developing Medical Genetics Education Through Curriculum Reforms And Establishment Of Postgraduate Training Programs

THE CONTENT OF SPECIAL MEDICAL GENETICS COURSES FOR NON CLINICAL GENETICIST RESIDENTS

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Competencies

At the completion of residency training, a medical resident should be able to:

- ✓ Perform an appropriate multigenerational family history and identify patients who have a personal medical condition and/or family history that indicates the risk of a genetically linked disorder, and provide appropriate counseling
- ✓ Effectively interview patients in order to obtain information about relevant genetic, environmental, and behavioral risk factors
- ✓ Understand the important social, behavioral, and psychological implications health- related genetic information can have for individuals and families

Knowledge

The resident should demonstrate the ability to apply knowledge of the following:

- ✓ Basic human genetics terminology, principles of human and medical genetics, and basic patterns of biological inheritance and variation
- ✓ The role of genetic factors in health maintenance and disease prevention
- ✓ The difference between clinical diagnosis of disease and identification of genetic predisposition to disease
- ✓ The role of behavioral, social, and environmental factors that modify or influence genetics in the manifestation of disease
- ✓ The implementation of relevant practice guidelines or consensus statements
- ✓ The range of genetic approaches to treatment of disease (including pharmacogenomics and gene therapy)
- ✓ The indications and resources for genetic testing and referral to genetic specialists
- ✓ The genetic testing and recording of genetic information regarding:
    - Screening for genetic abnormalities
    - Prenatal/preconception testing
    - Presymptomatic genetic testing
    - Carrier testing for genetic disorders
    - Confidentiality
    - Risk assessment
    - Informed consent

Skills

The resident should demonstrate the ability to independently perform or appropriately refer the following:

- ✓ Gather genetic family history information (including an appropriate multi-generational family history)
- ✓ Identify patients who would benefit from genetic services
- ✓ Explain basic concepts of probability, disease susceptibility, and the influence of genetic factors on maintenance of health and development of disease
- ✓ Appropriately seek assistance from and refer to genetics experts and peer support resources
- ✓ Obtain current information about genetics for self, patients, and colleagues
- ✓ Provide culturally appropriate information about the potential risks, benefits, and limitations of genetic testing
- ✓ Safeguard the privacy and confidentiality of the genetic information of patients
- ✓ Inform patients of potential limitations of maintaining privacy and confidentiality of genetic information

- ✓ Educate patients about availability of genetic testing and/or treatment for conditions seen frequently in practice
- ✓ Provide patients with an appropriate informed consent process to facilitate decision making related to genetic testing
- ✓ Utilize appropriate genetic and genomic information resources

Structure of Courses

Each course covers

- ✓ theoretical lessons,
- ✓ workshops, case studies,
- ✓ shadow clinical and clinical laboratory training,
- ✓ simulation of practical replacement, including genetic counselling

A. 2 week course “Clinical Cancer Genetics” for Oncologists

Topics
Cancer Genetics
Familial Adenomatous Polyposis
Lych Syndrome
Hereditary Breast and Ovarian Cancer Syndrome
Gynecological cancer
Multiple Endocrine Neoplasia Syndrome
Neurofibromatosis
Targeted therapy in oncology

B. 3 week course “Birth Defects and Congenital Disorders” for gynecologists, obstetricians, neonatologists, pediatricians

Topics
Congenital malformations (major and minor; single and multiple anomalies)
Teratogenicity and related causes of dysmorphic features
Chromosomal abnormality syndromes
Skeletal dysplasia
Single gene mutations as cause of birth defects and congenital disorders

C. 3 week course “Inborn Errors of Metabolism” for neonatologists, pediatricians, neurologists

Topics
Organic acidemias
Fatty acid oxidation defects
Primary lactic acidoses
Aminoacidopathies
Urea cycle defects
Disorders of carbohydrate metabolism
Lysosomal storage disorders

D. 1 week course “Autoinflammatory syndromes” for pediatricians, rheumatologists

Topics
Hereditary Periodic Fever Syndromes
Familial Mediterranean fever
TNF receptor-associated periodic syndrome
Hyperimmunoglobulinemia D with periodic fever syndrome
Cryopyrin-associated periodic syndromes

E. 1 week course “Multifactorial Diseases” for non-clinical geneticist residents

Topics
Marfan syndrome
Cystic Fibrosis
Polycystic kidney disease
Inherited cardiac disorders
Hypercholesterolemias
Hereditary deafness
Hereditary ophtalmological disorders
Cleft lip/palate
Diabetes mellitus
Metabolic syndrome
Asthma and allergy
Vascular disorders
Amyloidosis

F. 1 week course “Medical Genomics” for non-clinical geneticist residents

Topics
Genomic analysis: from karyotype to next generation sequencing
Next Generation Phenotyping with Face2Gene in the clinic, laboratory and for research
Classical and banding cytogenetics – techniques in pre- and postnatal diagnostics
Reporting in prenatal, postnatal and tumor cytogenetics
Molecular cytogenetics
FISH-applications in diagnostics
Array-CGH – principles and applications
PCR techniques and applications
NIPT
MLPA, related techniques and applications
Preimplantation genetic diagnosis and preimplantation genetic screening
Technical basis of Sanger sequencing
Technical basis of NGS

G. 3 week course “Clinical Laboratory Genetics” for laboratory clinician residents

Topics
Karyotyping in pre- and postnatal diagnostics
Molecular cytogenetics
Array-CGH, principles and applications
PCR techniques and applications
MLPA, related techniques and applications
Sanger sequencing, related techniques and applications
NGS, related techniques and applications

H. 3 week course “Prenatal Genetics” for gynecologists, obstetricians

Topics
Genetic diagnosis and genetic screening
Prenatal genetic diagnosis and genetic screening
Risk calculation based on genetic screening
Family risk calculation based on genetic diagnosis
Non-invasive genetic screening, including NIPT
Invasive cytogenetic diagnosis
Invasive molecular genetic diagnosis
Preimplantation genetic screening
Preimplantation genetic diagnosis