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TM26_Dip_ScClin

Teach Mob – Visiting Professors

Academic year 2014/2015

1st term
<p>COURSE TITLE Medical Genetics</p>
<p>Scientific area Internal Medicine</p>
<p>Department of Clinical and Biological Sciences</p>
<p>Language used to teach English</p>
<p>Course summary From the lab to the clinics: basic concepts and real examples of pedigrees, transmission modes, mutations, penetrance, variable expressivity, pleiotropy, linkage, one gene-one disease vs many genes-many diseases, somatic and germ-line mosaicism, polymorphisms, and the concepts of population genetics that are relevant in medicine. From the clinic to the lab: clinical reasoning in medical genetics, types of biological samples, diagnostic and presymptomatic gene testing, genetic counselling, available databases, guidelines, rules of privacy and informed consent. Cancer genetics: genetics of hereditary breast and ovary cancer, Lynch syndrome, familial polyposis, multiple endocrine neoplasia, complex cases, collaboration between specialities, role of follow up. Genetic diseases in neurology, cardiovascular and respiratory medicine, nephrology, ophthalmology, otorhinolaryngology. Prenatal genetics and chromosomal pathology. Future perspectives: pharmacogenetics, testing susceptibility to complex disease.</p>
<p>Learning objectives At the end of the course the student will be able to –recognize indications and applicative modalities of genetic tests and the role of pre- and post.- test genetic counselling. - to understand the role of gene mutations and polymorphisms in the pathogenesis of monofactorial as well as complex disease.</p>
<p>Tutorship activities A short attendance to genetic counselling sessions and in the lab will be offered to each student, under guidance of an Assistant Professor in Medical Genetics and a second certified MD consultant.</p>
<p>Lab activities Discussion of molecular findings in real clinical cases referred for inherited cancer syndromes or rare</p>

genetic diseases (e.g. Uromodulinopathies, primary Hyperoxaluria, Brugada Syndrome and other inherited arrhythmogenic syndromes)

Visiting Professor Profile

The successful candidate will possess a solid experience in research and the clinical practice of Medical Genetics, and be currently in charge of an official teaching appointment of Medical Genetics in a six years MD program, at least as Lecturer or Assistant Professor.

Competence in genetic counselling and genetic testing for rare disease, particularly if focused in introducing Next Generation Sequencing in the clinics, will be particularly appreciated.

Contact person at the Department

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