UNION EUROPÉENNE DES MÉDECINS SPÉCIALISTES
EUROPEAN UNION OF MEDICAL SPECIALISTS

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UEMS SECTION OF MEDICAL GENETICS

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Proposal to: **UEMS Council and ETR Review Committee**

Incorporation of 'Genetics and Genomics' in all UEMS ETRs

Introduction / Background

- Modern science and technology advances in **genetics and genomics** have relevance for all branches and specialties within medicine and surgery of all ages, with increasing relevance for the management, screening, and therapy of many diseases.
- **'Genetics'** can be thought of as the effect of genetic variants / alterations on the health, life style and family planning of individuals and their families, and all related aspects of laboratory testing, counselling and clinical management.
- **'Genomics'** can be thought of as the changes of genomic material (in DNA or epigenetic) which contribute to disease processes and are therefore important to the understanding of the pathophysiology of many conditions and diseases, including cancer which affects all specialties.
- Currently, there is great variation in the extent to which **genetics and genomics** feature in UEMS ETRs, ranging from almost nothing to relatively sophisticated levels of knowledge expected of senior trainees.
- If UEMS ETRs are aiming to be world class, they should all include at least a basic understanding of **genetics and genomics**, similar to, or exceeding, the knowledge expected at undergraduate level.
- Thus, new and updated ETRs across specialties should consider accommodating basic **genetics and genomics** under the domains of '**knowledge'**, **'skills'**, and **'attitudes'**.

Knowledge

- In relation to inherited conditions, basic understanding of the main patterns of inheritance and simple recurrence risks associated with them:
  - Autosomal dominant
  - Autosomal recessive
  - X-linked
  - Mitochondrial

- Appreciation that phenotypic features and their variability may be influenced by:
    - Reduced penetrance
    - Different types of pathogenic variation in DNA
    - Somatic mosaicism
    - More than one gene as the underlying cause
    - Different environmental factors and lifestyle choices
    - X-inactivation in the case of X-linked diseases
- Basic understanding of different types of molecular pathology at the level of the genome:
    - Abnormal chromosome number (aneuploidy), microdeletions and microduplications (copy number variants, *aka* CNVs)
    - Pathogenic variants (formerly 'mutations') in DNA alter the reading frame and protein product in different ways (eg missense, nonsense, frameshift)
    - Trinucleotide repeat sequences giving rise to 'anticipation'
    - Rarely, altered or non-expression of genes through 'imprinting', mediated by DNA methylation
- A basic understanding of the genes and genetic mechanisms relevant to the more common conditions seen in the specialty, including the availability of genetic testing to confirm diagnosis
- Knowledge (or 'awareness' for some specialties) of the main genetic laboratory methods relevant for the diagnosis of the more common conditions seen in the different specialties, their possibilities and limitations, in particular
    - Classic karyotyping
    - Chromosomal microarray (*aka* CGH-array)
    - Single gene sequencing
    - Next Generation Sequencing, panel sequencing, exome and genome sequencing
- In cancer genetics:
    - Awareness that most common cancers arise due to the accumulation of genetic damage in cells and tissues
    - A proportion of common cancers are strongly inherited, primarily due to pathogenic variants in single genes

- Rare inherited family cancer syndromes may present in various ways and, when diagnosed, there are screening and/or genetic testing implications for other family members at risk

- Knowledge of the legal prerequisites of genetic / genomic testing

Skills

- Ability to take and interpret a basic family medical history (pedigree)
- Within the range of conditions encountered in the specialty, recognition of those that may be inherited and therefore hold risks and medical implications, including potential screening, for other family members
- Ability to recognise when a patient / family should be referred to the local / regional Genetic Service for further assessment / genetic testing / risk counselling
- Ability to obtain informed consent for genetic / genomic testing for the diagnosis of the more common conditions seen in the specialty

Attitudes

- Ability to work in multidisciplinary teams including Clinical / Medical Geneticists
- Awareness of the possible impact of an individual's genetic / genomic test result on other family members and future family planning
- Awareness of limitations in:
  - Knowledge of genetics and genomics
  - Interpretation of genetic data and reports
- Readiness to refer, when necessary / appropriate, to local / regional Clinical Genetics expertise

UEMS Section of Medical Genetics (Bureau, National Delegates, ETR Revision Group)

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