6.1. *Proposed creation of a MJC Rare and Undiagnosed Diseases ***

This item was proposed by UEMS Section of Clinical Genetics

Béla Melegh
president
UEMS Council meeting
16-17th October, 2015
Warsaw

UNION EUROPÉENNE DES MÉDECINS SPÉCIALISTES
EUROPEAN UNION OF MEDICAL SPECIALISTS
SECTION OF CLINICAL GENETICS

Dr Edwin Borman
Secretary General
Union Européenne des Médecins Spécialistes
European Union of Medical Specialists
Rue de l'Industrie, 24
1040 – BRUSSELS

25 August, 2015

Dear Professor Borman,
Dear Secretary General,

On behalf of the Clinical Genetics section (CGS) of the UEMS, hereby I contact you with the request to create an MJC for "Rare & Undiagnosed Diseases". Attached please find a short summary to underline the rationale of this action, with the list of sections of the UEMS in annex who already expressed their interest. Please put into the agenda of Warsaw meeting this aim to enable us to present the objectives to the National Representatives and to the representatives of the sections.

Yours sincerely,

Béla Melegh
president of CGS

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Establishment of UEMS MJC for "Rare and Undiagnosed diseases"

Background, specific challenge:
In most of the EU countries a disease considered as rare (or orphan) disease if the disease affect not more than 5 per 10 000 persons in the general population. This group, however, is quite large, as it is estimated that rare diseases include about 6 000 to 8 000 separate entities, affect altogether more than 30 million people in the EU countries. It was already recognized, that due to the small and dispersed patient populations of the individual specific rare diseases the international collaborations in the diagnosis, care, treatment, and research efforts are crucial. The rare disease task recently has been modified and the task extended to include the whole spectrum of the undiagnosed diseases as well. This task involves most if not all medical specialties. About 80-90% of this disease group has genetic background, about 10-15% is non-genetic amongst the non-communicable part, and 5-10% has infectious origin. Thanks to advances of the new tools like the array technology and the next generation sequencing, great progress is seen in the understanding the molecular pathogenesis of the diseases of genetic origin, the research and the diagnosis is tightly linked in this field. Albeit the European Commission recognized the significance of this issue, and numerous huge systemic research projects have been granted, moreover, the implementation of European Reference Network for rare diseases is now on the way, almost any step towards a systemic and harmonized EU compatible training scheme has been devoted.

Scope:
The aim of this effort includes the incorporation of the principles of methods of genomics and/or other –omics and/or other high-throughput approaches used in the molecular characterization of rare and undiagnosed diseases into the training of medical experts of various sub-specialties. As the undiagnosed rare diseases may range from groups of disorders with relatively common and phenotypically well described diseases to groups of diseases with extremely rare incidence rate an almost unknown molecular basis, care should be taken to the appropriate training of the experts about common standards and terminologies for rare disease classification and also support their training on appropriate bioinformatics tools and incentives to facilitate data sharing, including the management of the existing resources used for depositing data generated by the different platforms.

Expected impact:

- Provide better understanding of genomic methods for the correct diagnosis of undiagnosed rare diseases for which there is no or unsatisfactory diagnosis available.
- Contribute thereby towards the multidisciplinary harmonization of the essential knowledge base of the new generation genomic approaches.
- Foster dissemination of novel scientific results and knowledge exchange between specialties.
- Provide better knowledge for improved family counselling as well as to improve follow-up for patients.
- Help to develop knowledge management strategies, with the view of facilitating models of care and access to the data gathered by different rare disease networks.
- Help putting into the right practice on the EC regulations on the *in vitro* diagnostic medical devices, with special focus on the genetic diagnostic ones.
- Contribute to the development of the best practice to regulate how and by whom patients are counselled before a genetic test.

Type of action: MJC

The goals related to this topic are optimally provided by the MJC conditions. The MJC proposal should enable and foster exchange between stakeholders from countries and regions with different practices and strategies of rare disease task.

Interest of UEMS Sections & Boards about creation of the Rare and Undiagnosed Diseases MJC:

Founder:
Section of Clinical Genetics, Bela Melegh
Charter members:
Section of Internal Medicine, Reinold Gans
Section of Medical Oncology, Serdar Turhal
Section of Child and Adolescent Psychiatry, Dame Sue Bailey
Section of Dermatology and Venereology, Magdalena Czarnecka-Operacz
Section of Infectious Diseases, Jean-Paul Stahl
Section of Pediatric Surgery, Gian Battista Parigi
Section of Neurology, Patrick Kras

Personal: Giorgio Berchicci, and Helena Alves (Section of Laboratory Medicine)