

UEMS—SECTION OF MEDICAL GENETICS

History of the Section up to 2021

The UEMS Section of Medical Genetics was born in 2013 (initially the Section of Clinical Genetics) after a long and drawn out gestation period. Dr Ulf Kristoffersson (Lund) here provides a personal account of those early beginnings:

“Back in 1997 when the ESHG set up its Public and Professional Policy Committee (PPPC), where I was a founding member, one agenda issue was how to achieve EU-recognised status for our medical speciality according to the then valid EU Directive. Professor Peter Farndon (Birmingham) found via his contacts in the UK that persons involved in the 1999 update of the Directive had discussed Clinical Genetics as a potential newly recognised specialty. We also learned that there would not be an update for several years. Thus, the PPPC left the issue behind to focus on its other activities.

In 2004 I was representing the PPPC at the annual ESHG Board meeting in Munich. During the meeting Professor Jen-Jacques Cassiman (JJC; Leuven) informed the group that he had met Dr Bernard Maillet (BM; Antwerp), the then Secretary-General of UEMS, and that they might be able to help us. As a result, I was tasked to contact him. JJC also informed us that a prerequisite was a European recognised training programme (ETR). So, a working group was set up consisting of myself and Professor Dian Donnai (Manchester), later replaced by Dr Helen Kingston (HK; Manchester), to draft an ETR.

In the spring of 2005 I had the first meeting with BM in Brussels and it was suggested to form a Multidisciplinary Joint Committee (MJC). An MJC is formed by at least three member sections on a topic of common interest, and the UK was invited to the UEMS Council meeting in October to present the case. As a consequence, the Section of Obstetrics and Gynaecology, together with the Sections of Paediatrics and Dermatology, formed a MJC which was approved by UEMS Council in October 2006. I was appointed Chair with HK as Secretary, and she proved to be a much valued side-kick during those years. As the MJC Chair I was recruited to serve as assessor for EACCME accreditations, which provided some funding.

The MJC had a draft ETR approved by the ESHG and UEMS Council, thus providing a platform to approach the EU Commission. However, this turned out to be complex. Our contacts within UEMS used their network but we were told that the Directive 2005/36/ECEU would not be updated in the near future. But with the formation of the MJC a number of people expressed interest in helping to make a Section possible. They joined the MJC and we started to update the ETR. In parallel, within the EuroGentest network and the OECD Working Group on OECD Guidelines for Quality Assurance in Genetic Testing, discussions took place on how to proceed towards EU recognition, and a document was produced describing the need for a European specialty for Medical Genetics. We were learning that progress would not be possible without bringing the issue to a governmental, political level. After a EuroGentest meeting in Brussels, Professor Milan Macek (MM; Prague) and I discussed if we could depend on the OECD recommendations together with the upcoming Directive on Rare Diseases to get things moving, and MM made further contacts.

During the French presidency of the EU the profile of Rare Diseases was raised. Professor John Burn (Newcastle) was then the ESHG President (2006-07) and Professor Arnold Munnich (Paris) was able to organise a breakfast meeting between them and the French Deputy Minister of

Health. Some six months later, during the Czech EU presidency, MM was summoned to a meeting with the Health Department in Prague, who had inherited the issue initiated by the French, and he very happily pursued the task. This continuity was necessary because typically the Commission responds when submissions are made by consecutive national presidencies. Thus, a proposition to add Medical Genetics to the Directive was put forward and endorsed in 2009.

The next step was to form a Section within the UEMS. New sections are formed on the suggestion of a full member, i.e., the national medical association of a member country. It was necessary for the ETR to be endorsed by the national specialist organisations, which led to a discussion on the length of training. A compromise of four years was reached. In 2011 the Swedish Medical Association proposed to the UEMS Council the creation of a new section, namely Clinical Genetics. The MJC had provided the necessary documentation and in 2012 the UEMS Council adopted the new section with acclaim.”

And so it was that once this milestone had been achieved, a call for national delegates from European member states was made – two delegates per member state – and the inaugural meeting was held in Brussels on November 29th, 2013. The first Board consisted of Ulf Kristofferson, President; Kristiina Aittomäki (Helsinki), Secretary; Helen Kingston, Treasurer; and members Milan Macek, Béla Melegh (Pécs), Feliciano Ramos (Zaragoza), André Ries (Erlangen), and Alessandra Renieri (Siena). In mid-2015 Béla Melegh took over from Ulf Kristofferson as the Section’s second President, Alessandra Renieri as Secretary, and Artúr Beke (Budapest) as Treasurer.

A section receives no money from UEMS and contributions are therefore required from member associations. One of the first tasks of the newly elected board was to find a subscription formula acceptable to all member associations, in addition to a small income from EACCME assessments. The Section was initially designated ‘Clinical Genetics’. In 2018, following the proposal of UEMS President, Romanu Krajewsky, the UEMS Council changed the Section name to ‘Medical Genetics’, in harmony with the Directive of Annex V.

The European Diploma in Medical Genetics and Genomics (EDMGG) is a development of UEMS Section of Medical Genetics (UEMS-SMG), implemented in collaboration with the European Board for Medical Genetics (EBMG) – Branch of Medical Genetics and Genomics (BMGG) – and the ESHG. The first aim of the SMG Board was to prepare a pan-European training guide, a syllabus, as a common goal of the SMG and EBMG-BMGG, which was coordinated by Professor Alessandra Renieri. In March 2015 a group met in Gdansk to develop the syllabus (Alessandra Renieri, Ulf Kristoffersson, Béla Melegh, Feliciano Ramos, and Peter Turnpenny [Exeter]). A Memorandum of Understanding was reached between the UEMS-SMG and EBMG-BMGG in 2015.

Completion of the syllabus was the first step towards developing a European Examination in Clinical Genetics, which was further promoted by the Section’s next President, Professor Béla Melegh, who took over from Ulf Kristoffersson in mid-2015. Work on the European Training Requirement (ETR) for the specialty was presented to the SMG Board at a meeting in Zaandam, Netherlands, in March 2016. At the same meeting it was decided that the syllabus could be a stand-alone document with the EBMG-BMGG but would also be part of the official European Diploma in Medical Genetics and Genomics (EDMGG) documents, as recommended by Professor Vassilios Papalois, UEMS Secretary-General at that time. In addition, Béla Melegh prepared a

draft of the 'Description of the Specialty of Medical Genetics', which later became part of the examination documents.

These documents were approved by the UEMS Council in March 2017, a necessary step before examination preparation could commence in earnest. At the SMG meeting in Copenhagen, May 2017, the process of developing the EDMGG continued, expressions of interest were received from a number of national delegates, and a 24-month project was envisaged in order to deliver the inaugural examination.

On June 23rd 2017 Béla Melegh and Peter Turnpenny were granted access to observe the UEMS Neurology Examination in Amsterdam. Thereafter, a call was issued to national delegates inviting commitment and attendance at the first examination workshop. Several members responded positively and, in addition, Dr Jonathan Berg (Dundee), with substantial experience in medical examining, was invited. The first workshop took place in October 2017, hosted by Ulf Kristoffersson in Lund. Subsequent workshops took place in Dundee and Heidelberg in preparation for a pilot EDMGG in Milan, June 2018, which proceeded successfully with eight candidates.

The next workshops took place in Maastricht and Zaragoza in preparation for the inaugural EDMGG in Gothenburg, June 2019, which successfully assessed 20 candidates, 15 of whom passed. After further workshops in Roskilde and Perthshire, preparations for the 2020 Examination, now designated the European Certificate in Medical Genetics and Genomics (ECMGG), were interrupted by the Covid-19 pandemic.

In October 2020 elections took place for a new Bureau (executive officers) for the Section of Medical Genetics, which assumed office on January 1st 2021.