

## **UEMS—SECTION OF MEDICAL GENETICS**

### **History of the Section up to 2025**

#### **1997-2004**

Written by Dr Ulf Kristoffersson

*“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 speciality. 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; Louvain) 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.*

#### **2005**

*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 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.*

#### **2006-2012**

*During the French presidency of EU the profile of Rare Diseases was raised. Professor John Burn (Newcastle) was then the ESHG President (2006-2007) and Professor Arnold Munnich (Paris) was able to organise a*

*breakfast meeting between them and the French Minister of Health. Some six months later, during the Czech 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.”*

## **2013-2016**

Written by 2021-2024 Bureau

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 29<sup>th</sup>, 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 Reis (Erlangen), and Alessandra Renieri (Siena). 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’ but in 2017, after debate within the UEMS Council and elsewhere, was changed to ‘Medical Genetics’ in harmony with the Directive.

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 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 Requirements (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.

## **2017-2018**

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.

## **2019-2020**

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 1<sup>st</sup> 2021.

## **2021-2024**

The Bureau consisted of Ute Moog (Heidelberg), President; Peter Turnpenny (Exeter), Secretary; Yvonne Arens (Maastricht) for 9 months, followed by Cristina Rusu (Iasi), Treasurer; Feliciano Ramos (Zaragoza), Vice President; and Jonathan Berg (Dundee), Chair of Examination Committee. New homepages for the Section and the ECMGG were created at <https://www.uems-genetics.org> and <https://www.uems-ecmgg.org>, respectively, and maintained by Adam Tobias (Glasgow).

The ECMGG exam was developed fully online, both for the multiple choice questions (MCQs) and the oral assessments, and CYIM (Cyber Imagination; <https://www.cyim.com/en>), based in Rennes, was chosen as the delivery partner to provide the proctored exam platform, as well the online database for managing MCQs from 2022. Since 2021 the exam has been delivered annually to a growing number of candidates. Statistical analyses indicate a high-quality exam. In 2024 the ECMGG underwent satisfactory appraisal by the UEMS-Council of European Specialist Medical Assessment (CESMA).

A revision of the ETR for the specialty of Medical Genetics was carried out from 2021 by a working group under the lead of Johannes Zschocke (Innsbruck) and officers of the SMG Bureau in close cooperation with the ESHG and the EBMG. Besides a general update, the examination for the ECMGG was integrated in the new version of the ETR and the syllabus completely restructured. This revised ETR was approved by the UEMS Council in October 2023. It is available for download at the UEMS-SMG website (<https://uems-genetics.org/Education.html>) and at the UEMS website ([European training requirements | uems.eu](https://www.uems.eu/European_training_requirements)).

A reliable structure of the Branch of Medical Geneticists of the EBMG was found ([https://www.ebmgeu/fileadmin/MGG/SOP\\_EBMG\\_BMG\\_220321\\_FINAL.pdf](https://www.ebmgeu/fileadmin/MGG/SOP_EBMG_BMG_220321_FINAL.pdf)), reflecting the considerable overlap between this Branch and the SMG, and the wish to expand on the collaboration with the ESHG. In 2024, a revised Memorandum of Understanding was signed between the UEMS SMG, the ESHG and the EBMG with the intention to further intensify and structure a collaboration between the three parties.

The number of member countries and national delegates to the Section increased, counting 29 countries and roughly 50 delegates at the end of 2024. Many of them agreed to represent the Section to other UEMS bodies or the ESHG.

In 2024, elections took place for three Bureau members for reasons of retirement or ending renewed terms. Shortly before business was handed over to the new Bureau, two papers dealing with the ETR version 2023 and the ECMGG exam were submitted to the *European Journal of Human Genetics*.