

EQA providers and other active in laboratory quality assurance are welcome. At present, participants from over 15 countries are registered; further places are available.

Programme and registration information is available on the Symposium web site at <http://www.eurogentest.org/QualitySymposium/index.xhtml>

Prof. Elisabeth Dequeker, Sarah Berwouts, Dr Michael Morris

Report from the Clinical Genetics EU Recognition Committee



*Dr. Ulf Kristoffersson,
Chair, Lund May 2009*

The committee has been very active through the Multidisciplinary Joint Committee - Clinical Genetics (MJC) of UEMS (WWW.UEMS.net). The educational programme for medical doctors' specialisation in clinical genetics has been endorsed by the Boards and Sections' Meeting in February and finally by the UEMS Council in April this year.

In parallel, Milan Macek of Prague has been very active during the Czech presidency of EU in order to enforce a recognition, in conjunction with the Recommendations on Rare Diseases that will be decided on by the ministers later this spring. Hopefully his work will bring our efforts closer to our task which is an EU recognition of Clinical Genetics as a medical speciality.

The committee will continue to work close with the MJC, who now will disseminate information about these guidelines to the national professional organisations and their section of clinical genetics in order to form a network of national contacts.

Proposal for a "EU Council recommendation on a European action in the field of rare diseases" and the amendment of the Directive 2005/26/EC with clinical-/medical genetics



*Professor Milan Macek
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Rare diseases and clinical- / medical genetics

EU has, together with the European Medicines Agency (<http://www.emea.europa.eu/>), defined a rare disease as one which affects fewer than 5 people per 10,000. Given the overall EU population size the number of sufferers is high, since there are over 7,000 known rare diseases. Most of these diseases are due to defined genetic defects, but environmental exposure during pregnancy or later in life, often in combination with genetic susceptibility, account for another common cause. A subset of these diseases comprises also rare complications of common diseases. While first symptoms may be detected at birth or in childhood, more than 50% of rare diseases appear during adulthood, and are often life-threatening or progressive and debilitating. Usually there is no effective treatment, but early diagnosis, followed by suitable medical and social care, can improve quality of life and life expectancy of those affected.

Although clinical-/medical genetics plays a crucial role in early diagnosis and management of rare diseases it has not been included in the list of "official" EU medical specialties listed in its "Directive 2005/36/EC of the European Parliament and of the Council from September 7, 2005 on the Recognition of professional qualifications" (<http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:L:2005:255:0022:0142:EN:PDF>). Inclusion of a speciality in this Directive assures free mobility of respective specialists within the EU by acknowledging their qualification achieved in a given member state at the EU-wide level, thereby permitting them to work in a EU member state of their choice.

EU Council recommendation on a European action in the field of rare diseases

Rare diseases constitute a serious public health concern and are considered a priority in the EU health and research programmes (http://ec.europa.eu/health-eu/health_problems/rare_diseases/index_en.htm). Following the very successful "Public Consultation" in which amongst others many clinical-/medical geneticists have voiced their opinion on a set of questions regarding improvement of diagnosis and care for rare diseases the European Commission published on November 11 / 2008 its "Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions on Rare Diseases: Europe's challenges {SEC(2008)2712; SEC(2008)2713} (http://ec.europa.eu/health/ph_threats/non_com/docs/rare_com_en.pdf).

Essentially, this important document stipulates a set of objectives and priorities related to a European action in the field of rare diseases which will be supported by the European Commission. According to the principles by which the EU is operating the Commission "Communication" should be accompanied by a subsequent EU Council "Recommendation", which lists priorities and objectives on which individual EU member states consensually agree upon and which ought to be practically implemented. Although a Council Recommendation is not legally binding it sets the frame for national

actions and represents a “background document” to which professional societies can refer to when pressing for national initiatives in a given area.

The Czech EU Council Presidency, for which I am serving as its scientific advisor for rare diseases, (www.eu2009.cz; January 1 – June 30/2009) has “inherited” from the previous French Presidency (www.eu2008.fr) a draft Proposal “EU Council recommendation on a European action in the field of rare diseases” (http://ec.europa.eu/health/ph_threats/non_com/docs/rare_rec_en.pdf). This document has already underwent four “examinations” of its text by EU Public Health committees at the EU Council in Brussels. It will most likely be adopted by the upcoming meeting of the Employment, Social Policy, Health and Consumer Affairs Council (EPSCO) in Luxemburg (June 8-9/2009) by EU27 health ministers. The Czech EU Council Presidency is doing its best to meet this deadline and to assure a smooth ratification process. Adoption of the Council Recommendation will be of great importance not only for rare diseases, but also for clinical-/medical genetics. This document in its Recital 15 states, inter alia, that „expertise should travel rather than patients themselves“. This statement provides the rationale for amending Directive 2005/36, since obviously our specialty is crucial for diagnosis and management of the majority of rare diseases. Furthermore, if clinical-/medical geneticists are to travel in order to provide their expertise in other EU member states, our specialty has to be included in this Directive. Thus, we are in fact in a rather unique position to achieve this otherwise politically and practically difficult goal, since this Directive has not been amended since 2005!

Union of European Medical Specialists consensus document on postgraduate training

Another critical piece of work, that has facilitated the process of amendment of Directive 2005/36, was done by prof. Ulf Kristoffersson (Lund) who has represented our field in the Joint Interdisciplinary Committee of the European Union of Medical Specialists (www.uems.net). During the last several years and after many meetings, negotiations and/or presentations of a draft clinical-/medical genetics “consensus” postgraduate curriculum to representatives of other medical disciplines the UEMS has adopted the consensus document “Description of Clinical Genetics as a medical specialty in the EU: aims and objectives for specialist training” (2009/15; on April 25/2009). This document describes the profile, entry criteria, educational goals and most importantly the consensual time frame for specialist training in our specialty of 4 years. This document was already provided to the EC - DG Internal Market and Services, that is responsible for the amendment of Directive 2005/36. This Commission department follows upon recommendations of its so called “Group of Coordinators for Recognition of professional qualifications” (or “Recognition Committee”) that convenes about 4 times per year and represents views of respective national authorities on this subject. This group mostly comprises representatives of national ministries for e.g. European affaires, Health, Research or Education who vote on the recognition of a given specialty by a proportionate vote, i.e. representatives of large EU member states have the strongest leverage. Usually, requests for amendments of Directive 2005/36 for a certain specialty are issued either by individual member states representatives or more commonly by representatives of the current EU Council Presidency. In this respect full credit goes to prof. John Burn (Newcastle) and prof. Arnold Munnich (Paris) who visited the French Minister of Health for an informal breakfast in November 2008 and requested that French EU Presidency launches an official request to the Recognition Committee to include our specialty. Arnold and John were successful (!) and the DG Internal Market has received the official document “French request for inclusion of specialty of Medical Genetics under Annex V” for its March 26 / 2009 meeting. Among others the request stated: “Concerning the specialty of Medical Genetics, the French authorities wish to address the question of its existence and of its content in the other countries of the European Union in the Committee of Directive 2005/36/EC in view of its inclusion, if necessary, in the list of those specialties which can benefit from mutual recognition, insofar as at least 2/5 of the Member States would already recognise this specialty. In France this is a speciality sanctioned by a specialised diploma (diplôme d’études spécialisées – DES), issued by the universities. You will find in Annex a sheet recapitulating the activities concerned and the duration of training“.

Czech EU Council Presidency activities aimed at EU-wide recognition of clinical- / medical genetics

The Czech EU Council Presidency followed upon this initial request in that it was up to me to prepare, together with the Czech Ministry of Education representative in the Recognition Committee Ms. L. Slobodová, an overview regarding the status of our specialty in EU27. Members of the Committee are mostly interested in a consensus EU postgraduate curriculum (ref. UEMS), consensus on the duration of training (in our case 4 years – ref. UEMS), collection of legal dossiers regarding the recognition of this specialty in individual member states (we got them all in the “pdf” format in national languages), whether clinical- / medical genetics is a primary or secondary specialty (subspecialty), including other relevant details and contacts on representatives of national professional specialties under whom our field belongs. In this respect I have contacted all presidents and/or other representatives and got thanks to their prompt response all necessary data for the March 26 meeting in Brussels. I have received a lot of supportive documents from prof. Ulf Kristofferson and from our Spanish colleagues (Drs. Feliciano J. Ramos and Ismael Ejarque Doménech) who have been struggling to have clinical-/medical genetics recognised in Spain.

The Czech delegation has presented the Recognition Committee with an overview table concerning the status of our field in the European Union. Hereby, we comply with the provision that our specialty first must be recognised in 2/5 of the EU27. With the exceptions of Belgium, Estonia, Greece and Spain where recognition process is currently under way, Cyprus and Luxemburg where medical genetics is not recognised and no initiatives to change this situation have been launched, our specialty was otherwise “officially” recognised by all national authorities. Furthermore, with the exception of Hungary our specialty is a primary specialty, which substantially increases our chance for a EU-wide recognition. Interestingly, term “clinical genetics” is used in 9 cases, “medical genetics” in 10 instances, once either term “genetics”

(Malta) or “human genetics” (Germany) are used within official legal dossiers. We had to explain to the Recognition Committee that all these terms are synonyms for the same specialty, and the UEMS in the end was more in favour of using the term “clinical genetics” for the description of our specialty. Although these differences in “semantics” may sound negligible to us, it took some effort to explain them outside of our domain and to the members of the Recognition Committee.

With respect to the duration of training eight countries require 5 year long postgraduate training, while the majority settled on 4 years of postgraduate training in our specialty. Usually the “extra” year, i.e. beyond the 4 year “consensus” adopted by UEMS, comprises one year of internship in a medical field closely related to clinical-/medical genetics. The UEMS document clearly accounts for this by stating in its last paragraph dealing with the “Time frame for specialist training, inter alia: In the longer training period, up to one year could be in another speciality of importance for clinical/medical genetics“. Thus, the main issue that needs to be resolved at the moment is to receive “endorsements” from clinical-/medical genetics professional societies, where the curriculum is set for 5 years, in that they will accept professionals from countries where the curriculum is limited to 4 years.

Currently, we are preparing these documents for the next Recognition Committee which is scheduled for June 22 /2009. I will inform you about the most recent status of this initiative at the upcoming 5th Meeting of National Human Genetics Societies at ESHG 2009 in Vienna.

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UEMS 2009 / 15

Description of Clinical Genetics as a medical specialty in EU Aims and objectives for specialist training

Endorsed by: The European Society of Human Genetics board and membership (2007) The UEMS Multidisciplinary Joint Committee for Clinical Genetics (Jan 21, 2009) The UEMS Specialist Sections & European Boards (Feb 21, 2009)

Adopted by: The UEMS Council (April 25, 2009)

Specialty Profile

Clinical Genetics describes the medical elements of Genetics Services provided to individuals and families (and sometimes populations). Other components include

laboratory genetics (cytogenetics, molecular genetics, and biochemical genetics), genetic counselling and academic genetics. The core activities of a genetic service can be defined as ‘integrated clinical and laboratory services, provided for those with/concerned about a disorder with a significant genetic component (both inherited and sporadic). Due to the sharing of genes among family members, the whole family, not only the individual, represents the core patient in clinical/medical genetics.

This document relates to medically qualified individuals intending to train in the specialty of Clinical/Medical Genetics. It recognises that there may be overlaps with training programmes for other genetic professionals (scientists and counsellors) and that there may be opportunities for joint training for periods of the course.

Entry criteria

This may vary from country to country but would generally include a specified period of general medical training to include adult +/- paediatric medicine prior to commencing specialty training in Clinical Genetics, “internship”. Some countries may have a minimum period of training to be undertaken before specialisation.

Educational goals

Knowledge and Skills

- **Theoretical genetics/Basic Science** which may include
 - understanding cellular and molecular mechanisms that underpin human inheritance,
 - understanding patterns of inheritance and methods for risk assessment,
 - genetic epidemiology and biostatistics
- **Clinical/Medical knowledge and skills**
 - Pedigree construction.

- Diagnosis, investigation and genetic management of individuals with both common and rare inherited/ genetic diseases and their families.
- Risk assessment and role in genetic testing.
- Paediatric genetics including training in Dysmorphology (knowledge of common dysmorphic syndromes, their aetiology and the use of dysmorphology databases) and investigation of learning disability in children.
- Adult genetics to include knowledge of late onset disorders and disorders with a significant genetic component presenting in adult life (including predictive testing).
- Prenatal Genetics and knowledge about fetal development and teratogens
- Population genetics, including genetic screening programmes
- Special areas of genetics including
 - Inherited metabolic disorders
 - Neuro- and neuromuscular genetics
 - Cardiovascular genetics
 - Cancer genetics
 - Neurosensory genetics (visual and hearing conditions)
 - Pharmacogenetics
 - Other subspecialties of specific interest to the trainee
- **Genetic counselling and communication skills**
 - Training in genetic counselling for all types of genetic disease and situations encountered in clinical genetic practice. This includes counselling in relation to prenatal diagnosis and for late onset such as neurogenetic and cancer genetic disorders, including predictive testing. Where applicable, training in cocounselling with other professionals such as genetic counsellors.
 - Understanding ethical issues and importance of consent and confidentiality.
 - Development of good communication skills with patients, colleagues in genetic centres and other specialists and healthcare professionals, including understanding and handling of crisis reactions.
- **Laboratory skills**
 - Thorough knowledge of principles of laboratory techniques used in diagnostic testing
 - Interpretation of results from cytogenetic, molecular genetic and biochemical genetic analyses.
 - The time spent and the practical expertise gained in laboratory work may vary between countries, but sufficient to ensure highly specialised knowledge.

Other aspects of the Training Programme

- **Maintaining Good Medical Practice**
 - Develop a commitment to lifelong learning through continuing professional development and attend relevant courses and conferences.
 - Participate in Audit and Clinical Governance
 - Adhere to established consent and confidentiality procedures
 - Understand ethical and legal issues
- **IT skills**
 - Use of information technology including online resources and databases
- **Management training**
 - Knowledge about general healthcare policy, goals and priorities
 - Understanding the organisation of genetic services
 - Opportunities to participate in departmental activities related to organizational planning, financial management, and monitoring and maintaining quality standards
 - Development of multidisciplinary team working and leadership skills
- **Teaching**
 - Develop teaching skills by participating in the education and training of various categories of staff
 - Involvement with patient groups and patient education
- **Supplementary Education and Training**
 - Subspecialty training: some trainees will elect to develop expertise in a subspecialty area such as cancer genetics, dysmorphology or neurogenetics.

Quality Assurance

- Competency-based curricula should form the basis of a training programme.
- A written agreed curriculum for the training period should be set up as a contract between the trainee and the supervisor if not otherwise determined by national regulations.

- Trainees should maintain a Training Log including details of clinical and laboratory experience, educational activities, research and publications.
- A mechanism should be in place for continuous assessment of trainees against agreed quality standards. Some countries will have a nationally prescribed system for assessment and certification.
- Specialist examination may be compulsory in some countries

Research

- Medical genetics has a rapidly changing knowledge base and during specialty training the clinical/medical geneticist should be encouraged to participate in research. Some trainees will wish to take time out from the clinical training programme to undertake an intensive period of research leading to a higher academic degree. On completion of training some academic clinical/medical geneticists will continue to lead research programmes whilst many others will collaborate with laboratory based colleagues in the genetics team.

Time frame for specialist training

- The training period should minimum 4 years full time work; part time work would extend the training period.
- An educational training programme will be agreed for each trainee according to the specialty specific curriculum • In the longer training period, up to one year could be in another speciality of importance for clinical/medical/medical genetics.
- The time spent in laboratory work may vary between countries according to national curricula.
- A period of research resulting in a PhD/other higher exam may, if appropriate, replace training for a variable period of time according to national guidelines. However, in absence of national guidelines, it is not recommended that this time period is longer than 1/3 of the total training period

Editorial Report for EJHG over 2008



Dr. Gert-Jan van Ommen
Editor in Chief

- An important hurdle was made in 2007. Our factor impact increased by 0.306 points, to 4.003. Its ranking in the category of Genetics and Heredity also improved by 2 points, from 40 to 42.

- Submissions increased by 12.5% in 2008, while the acceptance rate has remained stable for the past three years, at 35%. EJHG published 10% more articles in 2008 than in 2007. However, in the first months of 2009 we have already seen a further rise in submissions, most likely driven by the IF increase. To cope with this we will have to raise our acceptance bar - which of course should further increase our IF.

- EJHG authorship is still predominantly European, with 70% of accepted articles. However, US/Canadian and Asian authorship keeps increasing, to 17% and 5% in 2008, while the rest of the world contributes.

- Decision times remained low for EJHG, with a median first decision of 20 working days and the median final decision time of 18 working days after submission of the last revision. We note that these are median figures and we are aware that specific manuscripts have had much longer processing times. This is mainly due to the fact that with the increasing amount of genetics journals, it has become more difficult to solicit reviewers and actually have them return their reviews on time. We will do all that is in our powers to address this,

amongst others by a 30% extension of the editorial time commitment.

- Due to the increased submissions, time to print publication increased throughout 2008, from 3.8 months in January to 5.4 months in December, which we aim to reduce by the increased editorial office input.

- EJHG performed especially well online in 2008. Seven out of twelve months we published more than 60% of articles as Advanced Online Publication within 25 working days. The average total web page views increased by 59%, home page views by 11%, abstracts by 39% and full-text articles by 33%. The most frequently accessed article lists are quite varied, although they feature mostly recent articles. Practical genetics tend to perform well, especially in terms of PDF downloads. Four of the most cited articles in 2008 were also featured on the most accessed lists. EJHG content accessed from PubMed averaged at almost 10,000 times per month.

EJHG Top Cited Papers 2007-2008

As every year, EJHG has a junior authors' high-citation award, to hand out at the Vienna meeting. The 1st prize includes a € 500 award and places 1-3 receive one year free ESHG membership + online EJHG, and free registration for the Barcelona meeting. This year's winner is Dr. Silverberg et al. with 26 citations in all of 2007 and Jan-March of 2008, for his paper "Refined genomic localization and ethnic differences observed for the IBD5 association with Crohn's disease", which appeared in EJHG 15 no. 3 (2007). Second and third prizes go to Dr. Bronner et al. for "Progranulin mutations