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PERSPECTIVE

From Mendel to Medical Genetics

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INTRODUCTION

Only a few years after the rediscovery of Mendel's laws of inheritance in 1900, the first human genetic disorders and variants were described.[1,2] Although these disorders were considered to be rare exceptions, the number of disorders to follow a Mendelian pattern of inheritance increased slowly. Victor McKusick, in his first volume of Mendelian Inheritance in Man published in 1966, listed 1486 entries, mainly phenotypes, whereas the current online catalogues contain over 8000 entries, with more than 5000 with a known molecular basis.[3–5]

The discovery of DNA as a carrier of the genetic code, its double helix structure and the rapidly developing possibility of the clinical use of chromosome- and DNA analyses made expertise in medical genetics (MG) valuable in health-care services and a subspecialty started to grow, mainly in gynaecology, neurology, paediatrics and laboratory medicine.

With the birth of the European Union (EU), a need for collaboration between established specialities emerged and the Union of European Medical Specialists (UEMS)[6] was founded in the same year that the Treaty of Rome[7] was signed. UEMS is an association of national medical professional organisations which focus on the harmonisation of training and education of medical doctors within and across all medical specialties. A number of specialities were soon mutually recognised in the member states as equivalent in training, leading to a national speciality licence being mutually recognised in all member states. At this time, MG was not recognised in any European country.

In this article, which includes the results of a recent survey, we describe the development of MG as a specialty in all European countries – not only within the EU itself – and the process undertaken in order to acquire its de iure recognition in the EU.

THE PROCESS TO EUROPEAN UNION RECOGNITION

The youthful status of our speciality is reflected in the fact that it has different names in different countries – MG, clinical genetics and human genetics being the most common, as they appear in the current version of European Directive 2005/36/EC on the recognition of professional qualifications[8] (Professional Qualifications Directive; PQD). In this paper, we will refer to MG.

The process of becoming an EU-recognised speciality started with a discussion in the European Society of Human Genetics (ESHG) Public and Professional Policy committee shortly after it was founded in 1997, but it was too late to have MG included in the 1999 revision of the PQD.

At the ESHG board meeting in Munich 2004, Jean-Jacques Cassiman brought up the issue again, as he had met with the Secretary General of UEMS. After a discussion in the ESHG board, Ulf Kristoffersson was appointed to lead an *ad hoc* committee together with Dian Donnai, who were later replaced by Helen Kingston and Didier Lacombe. Their task was to draft common European guidelines for medical training with a specialisation in MG. After 2 years, we were finished and the document was endorsed by the ESHG membership.

Important support came with the adoption of the Organisation for Economic Co-operation and Development (OECD) 'Guidelines for quality assurance in molecular genetic testing' (2007),[9] where many members of ESHG and the EuroGentest Network of Excellence EU project[10] were involved in the drafting. Article E5 of the Guidelines stipulates that 'Relevant government or professional authorities should recognise MG as a discipline comprising both a clinical and a laboratory specialty,' thus underlining the multidisciplinary character of genetic services and the need for official recognition of the medical and clinical laboratory professional branches involved in the provision of genetic services (see later). Further significant backing for the recognition of MG emerged when, in May 2008, the first international legally binding instrument concerning genetic testing for health purposes was adopted by the Committee of Ministers of the Council of Europe.[11]

In parallel, we established contact with UEMS and initiated the procedures necessary for us to become a member organisation representing MG. Only recognised specialities could become 'sections', but if at least two recognised specialities so wished, a Multidisciplinary Joint Committee (MJC) could be formed. Thus, with help of the sections of Paediatrics and Obstetrics and Gynaecology, an MJC for 'Clinical Genetics' was formed according to the procedures and statutes of UEMS. Ulf Kristoffersson was elected the Chair and Helen Kingston the Secretary. Being an MJC, we received a voice in UEMS and afterwards the UEMS council also adopted our ESHG-approved training guidelines as UEMS guidelines: 'Description of Clinical Genetics as a medical specialty in the EU: Aims and objectives for specialist training' (April 2009; amended 2017).[12]

During the European Human Genetics Conference in Vienna on 25 May 2009, the vast majority of those attending the 5th Meeting of the Presidents of the National Human Genetics Societies (NHGS)[13] signed a joint petition in support of the inclusion of MG in the PQD and endorsed the aforementioned UEMS consensus training curriculum. In addition, Jean-Jacques Cassiman contacted Frieda Brepoels, one of the Belgian members of the EU Parliament. She proposed a vote in favour of the recognition of MG in a Parliament Committee in March 2009. Unfortunately, there were not enough votes in favour for the motion to be carried.

Another important boost for the recognition of MG came from the successive French and Czech EU Council presidencies of the EU in July–December 2008 and January–June 2009, respectively. In November 2008, during the French term, John Burn and Arnold Munnich visited

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the French Minister of Health, Roselyne Bachelot, and asked for France to issue a formal request to the EC to start recognition proceedings. Indeed, the 'French Request for inclusion of the specialty of MG under Annex V' into PQD was later officially filed, following additional support from French Orphanet representatives (Ségolène Aymé) in March 2009. Concurrently, Milan Macek was the chief government advisor to the Czech Presidency. He worked closely with EURORDIS-Rare Diseases Europe, a non-profit alliance of over 700 European rare disease (RD) patient organisations (represented by Yann le Cam and his team) for the passage of the 'EU Council on Recommendation on an action in the field of RD (2009/C151/02).[14] After intensive work at the Council and lobbying within the 6-month window of opportunity, this key EU document was adopted in June 2009.

The provisions of the Council Recommendation created a strong momentum for the recognition of MG by setting out the relevance of training in the specialty for the diagnosis of RD, of which over 80% are genetic. Moreover, its Recital 15 provided us with justification for the cross-border mobility of MG (ie, 'expertise should travel rather than patients themselves'), it being the first line of diagnostic contact for the majority of these disorders. This clause was particularly relevant, since the PQD lists only those specialties where there is a justified need for cross-border provision of medical care and where there is a 'bottom up' consensus on a given postgraduate training curriculum by EU Member States for a particular medical specialty, that is, via the UEMS.

Following the French request to the EC, the ESHG worked with the NHGS representatives in providing the Recognition Committee (RC), an official EC body formed of member state representatives that has the power to authorise the EC to amend the PQD. At that time, EU presidents provided their national representatives at this committee with (a) endorsements of the UEMS consensus MG curriculum, including the harmonisation of respective national MG curricula with UEMS provisions and a minimal duration of postgraduate training of 4 years, (b) where applicable, legal dossiers stipulating national recognition of MG in their own countries and thus (c) 'evidence-based' support letters for the European recognition of MG.[15] These activities were coordinated by Milan Macek, who at that time served as the President of the ESHG, and were spearheaded by the Czech RC representative (Lucia Slobodová). By mid-2010, the RC was provided with the official evidence that MG is recognised as a medical specialty at the national level in 20 of the 27 EU member states, that is, as a primary specialty termed 8× 'clinical-', 10× 'medical-', 1× 'human-' and 1× genetics, while in Hungary MG was a subspecialty at that time. This overall number of national recognitions was greater than the qualified majority needed for a decisive vote by the RC (October 2010). Finally, on 3 March 2011, the EC adopted 'Regulation (EU) No 213/2011 amending Annexes II and V to Directive 2005/36/EC of the European Parliament and of the Council on the recognition of professional qualifications'.[16] This administrative act means that MG is now officially recognised as a European specialty. Subsequently, EU recognition of MG facilitated national recognitions in Spain (2014), Belgium and Croatia (both 2017), the transition of MG to a primary specialty status in Hungary (2012) and the creation of a new professional society in Iceland (2012).

After the EU recognition we applied, with the help of the Swedish Medical Association, to form a Section for Clinical Genetics[17] which was approved by the UEMS Council in 2013. Ulf Kristoffersson was elected the first president, and was followed by Bela Melegh in 2015. The three main tasks have been to update the training guidelines, to draft a syllabus for training and to develop a protocol for an European specialist exam planned to be offered for the first time in 2018. In 2016, the Section took the initiative of forming a 'MJC for Rare and Undiagnosed Disorders (MJC-RUD)', in order to be able to form a bridge between the UEMS and the newly established European Reference Networks (ERN) for RD[18] for collaboration on the harmonisation of MG training and education.

THE DEVELOPMENT OF MEDICAL GENETICS IN EUROPE

The 1997 survey 'Medical Genetics in Europe' provided evidence that 15 of the 24 EU countries participating in this exercise recognised MG.[19] In the spring of 2017, we performed an update of national legislative documents regulating MG in all member states of the Council of Europe,[20] 47 in all, adding Belarus as the only European country not being a member of this international organisation, and Israel, which is an 'Observer to the Parliamentary Assembly'. Five minor

Table 1 Current status of the medical genetics specialty in Europe: results of a 2017 survey

No of countries in the Council of Europe	*47*
Belarus added	1
Countries not included	4 (Monaco, Andorra, San Marino, Lichtenstein, Vatican)
No or incomplete data	2 (Azerbaijan and Luxembourg)
Countries included	42
No established specialty	2 (Greece and Cyprus)
No specialty but subspecialty	8
Currently primary specialty	32
Subspecialty before 1997	17
Primary specialty before 1997	15

Table 2 Agregated data on the year of recognition of medical genetics specialisation and/or subspecialisation

Year for specialty recognised	*Subspecialty recognised*	*Still subspecialty*	*Before specialty*
XXXX–1975	2	4	1
1976–1980	0	4	2
1981–1985	2	0	0
1986–1990	1	2	3
1991–1995	3	6	0
1996–2000	3	1	0
2001–2005	5	0	0
2006–2010	8	0	0
2011–2015	6	0	0
2016–2017	2	0	0
Sum	30	17	6
No data	2		

Table 3 Length of postgraduate training in the medical genetics specialty

Years of training for primary specialisation	
3 years	1
4 years	16
5 years	8
6 years	2
Missing data	5
Sum	32

Table 4

Country	Title (in English)	Name in national language	Primary specialty	Primary specialty established	Training (years)	Subspecialty (years)	Subspecialty name	Legal Dossier - official link for primary specialty	Contact	E-mail
Albania	Medical Genetics	Gjenetike Mjekesore	No			2	Medical Genetics	not provided	Anila Babameto-Laku	laku62@yahoo.com
Armenia	Medical Genetics	Բժշկական գենետիկա	Yes	2010	4	2000	Clinical genetics (1,5 y)	Certificate of Legal Entity State registration N03A 062161 0f Government of the Republic of Armenia; 04.07.2000.	Tamara Sarkisian	tamsar@sci.am
Austria	Medical Genetics	Medizinische Genetik	Yes	2006	6	1993	Humangenetik	Verordnung der Bundesministerin für Gesundheit über die Ausbildung zur Ärztin für Allgemeinmedizin/zum Arzt für Allgemeinmedizin und zur Fachärztin/zum Facharzt (Ärztinnen-/Ärzte-Ausbildungsordnung 2015 – ÄAO 2015) StF: BGBl. II Nr. 147/2015 vom 29.5.2015	Hans-Christoff Duba	Hans-Christoph.Duba@gespag.at
Azerbaijan	no contacts established									
Belarus	Clinical Genetics	Клиническая генетика	No			1987	Medical Genetics (2 years)	unavailable	Irina Kirillova	kirillova@hotmail.com
Belgium	Human Genetics	Génétique humaine, Menselijke erfelijkheid	Yes	2017	6	1988	Human genetics	Special BE law on genetics centres, "Royal Decree laying down the standards that the centers for human genetics must meet" which entered into force on January 1, 1988; publ. 25.12.1987 number 1987025417; page 19516.	Koen Devriendt	koenraad.devriendt@uzleuven.be
Bosnia and Hercegovina	Medical Genetics	Medicinska genetika	No			1986	Clinical Genetics (1986-2013); Medical genetics (from 2015)	Sub-specialization of Clinical genetics: Law on Health Care of the Republic of Bosnia and Herzegovina in 1986 (Official Gazette of the Republic of Bosnia and Herzegovina, years LII, no. 27, July 21, 1986; 1 year training). Medical Genetics was introduced on the basis of the Law on Medical Practice of the Federation of Bosnia and Herzegovina, published in the Official Gazette, No. 5, dated 19 July, 2013 and the Regulations on specialization / sub-specializations in the Federation of Bosnia and Herzegovina, Official Gazette No. 62, date 12 August, 2015, page 506-508 (18 month training).	Mensuda Hasanhodzic	hmensuda@gmail.com
Bulgaria	Medical Genetics	Медицинска генетика	Yes	2006	4	1976	Medical Genetics	Regulation 34 from 29/12/2006 Min Health published in State Gazette no 7. on 23/01/2007	Draga Toncheva	dragatoncheva@gmail.com
Croatia	Medical Genetics	Medicinska genetika	Yes	2017	5	1994	Medicinska genetika	pending by autumn 2017	Ingeborg Barišić	Ingeborg.Barisic@kdb.hr
Cyprus	Clinical Genetics	Κλινική γενετική						Regulation 185/2009 Coll. page 2695,	Violetta Anastasiadou	violetta.anastasiadou@cytanet.com.cy
Czechia	Medical Genetics	Lékařská genetika	Yes	2009	4	1969	Medical Genetics (2 years)	https://www.zakonyprolidi.cz/cs/2009-185	Milan Macek	milan.macek.jr@lfmotol.cuni.cz
Denmark	Clinical Genetics	Klinisk Genetik	Yes	1996	5			Bekendtgørelse om uddannelse af speciallæger. I medfør af §§ 32 og 34, stk. 5, i lov nr. 451 af 22. maj 2006 om autorisation af sundhedspersoner og om sundhedsfaglig virksomhed og under henvisning til § 3 i bekendtgørelse nr. 1248 af 24. oktober 2007 om speciallæger.	Uffe Birk Jensen	u.jensen@dadlnet.dk
Estonia	Medical Genetics	Meditsiinigeneetika	Yes	2009	4	1968	Medical Genetics	The change of legislation of Estonian Social Ministery from 28.11.2001, No 110 and changed on 27.07.2009 nr 74; https://www.riigiteataja.ee/akt/13211061.pdf	Katrin Õunap	katrin.ounap@kliinikum.ee
Finland	Medical Genetics	Perinnöllisyyslääketiede / Medizinsk Genetik	Yes	1981				Suomen Säädöskokoelma N:O 678 Asetus erikoislääkärin tutkinnosta) signed by the President of Finland on 4 September 1998	Helena Kaariainen	helena.kaariainen@thl.fi
France	Medical Genetics	Génétique médicale	Yes	1995	4			J.O. Numéro 31 du 5 Fevrier 1995 page 1992 LOIS LOI no 95-116 du 4 février 1995 portant diverses dispositions d'ordre social (1) NOR : SPS9400133L	Didier Lacombe	didier.lacombe@chu-bordeau.fr
Georgia	Medical Genetics	სამედიცინო გენეტიკა	Yes	2007	3			http://old.moh.gov.ge/inde.php?lang_id=GEO&sec_id=298&info_id=2357	Oleg Kvilidze	kvlividze@gmail.com

Table 4 (continued)

Country	Title (in English)	Name in national language	Primary specialty	Primary specialty established	Training (years)	Subspecialty (years)	Subspecialty name	Legal Dossier - official link for primary specialty	Contact	E-mail
Germany	Human Genetics	Humangenetik	Yes	1992	5	1978	Zusatzbezeichnung Medizinische Genetik	Bundesärztekammer, Arbeitsgemeinschaft der Deutschen Ärztekammern) in the document "Weiterbildung, Stand 2006" on page 60.	Christine Scholz	organisation@gfhev.de
Greece	Clinical Genetics	Κλινική γενετική						Not established	Lina Florentin	lflorentin@leto.gr
Hungary	Clinical Genetics	Klinikai genetika	Yes	2012	4	1993	Humángenetika (Human Genetics (1993-1999), Klinikai Genetika (Clinical Genetics (1999-2011) Training: 2 years	22/2012. (I.14.) EMMI Decree on receiving high level secialist certification in healthcare http://njt.hu/cgi_bin/njt_doc.cgi?docid=154386.333 678	Bela Melegh	bela.melegh@aok.pte.hu
Iceland	Medical Genetics	Erfðalæknisfræði	Yes	2015	5	2	Pediatric Genetics	www.mannis.is	Vigdis Stefánsdóttir	vigga@islandia.is
Ireland	Clinical Genetics	Géineolaíocht Cliniciúil	Yes	2012	4			https://www.medicalcouncil.ie/education/Specialty-Options/	Sally Ann Lynch	sally.lynch@ucd.ie
Israel	Medical Genetics	[illegible]	No			1986	Medical Genetics (2 1/2)	The Physicians' Regulations (Approval of Specialist Title and Eaminations), 1973 is the relevant law authorizing each of the recognized specialties in Israel, including medical genetics	Lina Basel-Salmon	basel@post.tau.ac.il
Italy	Medical Genetics	Genetica medica	Yes	1970	4		no	http://attiministeriali.miur.it/anno-2016/settembre/di-16092016.asp	Alessandra Renieri	alessandra.renieri@unisi.it
Latvia	Medical Genetics	Medicīnas ģenētika	Yes	2000	5	1994	Medical Genetics (Medicīniskā ģenētika)	The Ministry of Welfare Order No. 127 in 11.04.2000. (Labklājības ministrijas Rīkojums Nr.127 2000.gada 11.aprīlī), published in "Latvijas Vēstnesis" 18.04.2000. 136/139 (2047/2050). https://www.vestnesis.lv/ta/id/4714	Aigars Dzalbs	aigars.dzalbs@inbo.lv
Lithnuania	Clinical Genetics	Gydytojas genetikas	Yes	2004		1991	Clinical genetics (gydytojas genetikas) 2y	PATVIRTINTA Lietuvos Respublikos sveikatos apsaugos ministro 2004 m. birželio 28 d. įsakymu Nr.V-469	Vaidutis Kučinskas	Vaidutis.kucinskas@santa.lt
Luembourg	Medical Genetics	Médecine génétique			4			incomplete data		
Macedonia	Clinical Genetics	Клиничка генетика	Yes	2015	4	1991	Clinical Genetics	Врз основа на член 138 став (2), член 140 став (10), член 142 став (7), член 144 став (4) и член 332 став (9) од Законот за здравствената заштита „Службен весник на Република Македонија" бр. 43/12, 145/12, 87/13, 164/13, 39/14, 43/14, 132/14, 88/14,10/2015 и 61/15), министерот за здравство донесе П Р А В И Л Н И К ЗА СПЕЦИЈАЛИЗАЦИИТЕ И СУПСПЕЦИЈАЛИЗАЦИИТЕ НА ЗДРАВСТВЕНИТЕ РАБОТНИЦИ СО ВИСОКО ОБРАЗОВАНИЕ ОД ОБЛАСТА НА МЕДИЦИНАТА http://www.medf.ukim.edu.mk/upload/dok/3386_706005275.pdf	Dijana Plaseska- Karanfilska	dijana@manu.edu.mk
Malta	Clinical / Medical Genetics	Genetika Klinika/Medika	Yes	2005	5			Kummissjoin Nazzjonali ghal Edukazzjoni Avvanzata u Oghla. National Commission for Higher and Further Education (NCHFE)	Isabella Borg	isabella.borg@um.edu.mt
Moldova	Human Genetics	Genetică Umană	No			1978	Human Genetics (2 years in Russia)	http://biologiemoleculara.usmf.md/genetica-umana	Victoria Sacara	victoriasacara@hotmail.com
Montenegro	Clinical genetics	Klinička genetika	No				Clinical genetics (2 years)	PRAVILNIK O SPECIJALIZACIJAMA; Službeni list Crne	Olivera Miljanovic	oliveram@ac.me
The Netherlands	Clinical Genetics	Klinische genetica	Yes	1987	4	1972	Anthropogenetics	Goedkeuring kaderbesluit Centraal College Medische Specialismen Besluit van de Minister van Volksgezondheid, Welzijn en Sport van 1 d ecember 2004, nr. IBE/BO-2535849, houdende de goedkeuring van het kaderbesluit en de 27 specifieke besluiten van het Centraal College Medische Specialismen De Minister van Volksgezondheid, Welzijn en Sport, Gelet op: – artikel 14, tweede lid, onder f, van de Wet BIG; – artikel 14, tweede lid, van de Regeling inzake de opleiding en registratie van specialisten	Frederik Hes	f.j.hes@lumc.nl
Norway	Medical Genetics	Medisinsk Genetikk	Yes	1973	4			https://helsedirektoratet.no/autorisasjon-utdanning-og-godkjenning/spesialistgodkjenning/lege/medisinsk-genetikk	Gunnar Houge	gunnarhouge@gmail.com

Table 4 (continued)

Country	Title (in English)	Name in national language	Primary specialty	Primary specialty established	Training (years)	Subspecialty (years)	Subspecialty name	Legal Dossier - official link for primary specialty	Contact	E-mail
Poland	Clinical Genetics	Genetyka kliniczna	Yes	2003	5			Dziennik Ustaw Nr 213 — 14226 — Poz. 1779 ROZPORZĄDZENIE MINISTRA ZDROWIA z dnia 20 października 2005 r.w sprawie specjalizacji lekarzy i lekarzy dentystów	Olga Haus	haus@cm.umk.pl
Portugal	Medical Genetics	Genética Médica	Yes	2001	5	1979	Competência em Genética	DIÁRIO DA REPÚBLICA — I SÉRIE-B N.o 52 — 2 de Março de 2001 Portaria n.o 148/2001 de 2 de Março	Heloísa Santos	heloisa.santos@mail.telepac.pt
Romania	Medical Genetics	Genetica Medicala	Yes	2008	4			Ministerul SănătăÑii Publice Ordin nr. 1509/2008 din 02/09/2008	Maria Puiu	maria_puiu@umft.ro
Russian Federation	Genetics	Генетика	No			1988	Генетика 1988 (2 years)	Приказ Минздрава России от 26.02.2015 N 77н "Об установлении соответствия должностей медицинских работников и фармацевтических работников. The specialty is included in the nomenclature of medical specialties: "Doctor-geneticist" and "Doctor-Laboratory geneticist" http://mzdrav.rk.gov.ru/rus/file/pub/pub_246553.pdf	Evgeny Ginter	ekginter@mail.ru
Serbia	Clinical Genetics	Клиничка генетика	No			1970	Клиничка генетика (1 year)	http://www.zdravlje.gov.rs/downloads/2013/December/Decembar2013PravilnikSpec.pdf	Ivana Novakovic	novivana@eunet.rs
Slovakia	Medical Genetics	Lekárska genetika	Yes	2006	4	1972	Lekárska genetika	Act 322/2006 Coll, page 1994, section. 112	Ludevit Kadasi	kadasi@fns.uniba.sk
Slovenia	Clinical Genetics	Klinična genetika	Yes	2001	5			Pravilnik o vrstah, vsebini in poteku specializacij zdravnikov Št. 0070-2/2009, https://www.uradni-list.si/glasilo-uradni-list-rs/vsebina/2009-01-0866/pravilnik-o-vrstah-vsebini-in-poteku-specializacij-zdravnikov	Karin Writzl	karinwritzl@gmail.com
Spain	Clinical Genetics	Genética Clínica	Yes	2014	4			RD 639/2014 - Boletín Oficial del Estado (BOE) Nº 190, del 06/08/2014, pág 63130-63167.	Feliciano Ramos	framos@unizar.es
Sweden	Clinical Genetics	Klinisk genetik	Yes	1992		1977	Klinisk genetik	http://www.socialstyrelsen.se/ansokaomlegitimationochintyg/bevis,specialistkompetens/Documents/stmal-kliniskgen.pdf	Ulf Kristoffersson	ulf.kristoffersson@med.lu.se
Switzerland	Medical Genetics	Medizinische Genetik, Génétique Médicale, Genetica Medicale	Yes	1999	5			http://www.fmh.ch/bildung-siwf/fachgebiete/facharzttitel-und-schwerpunkte/medizinische-genetik.html	Peter Miny	Peter.Miny@unibas.ch
Turkey	Medical Genetics	Tıbbi Genetik	Yes	2003	4	1966	Medical Genetics	Regulation YÖK 15665/22.07.2003	Munis Dundar	dundar@erciyes.edu.tr
Ukraine	Medical Genetics	Генетика медична	No			1986	Medical Genetics (2 years)	List of primary specialty. Ministry of Health Protection Ukraine, act N333 (z0747-05) 06.07.2005, N76 (z0256-06) 21.02.2006	Halyna Makukh	makukh.h@ihp.lviv.ua
United Kingdom	Clinical Genetics	Clinical Genetics	Yes	1984	4			https://www.england.nhs.uk/commissioning/wp-content/uploads/sites/12/2014/04/e01-med-gen.pdf	Jill Clayton-Smith	Jill.Clayton-smith@cmft.nhs.uk
Not included (small countries)										
Holy Sea (Vatican)										
Monaco										
San Marino										
Andorra										
Liechtenstein										

member states were not included in this survey (Andorra, Lichtenstein, Monaco, San Marino and the Vatican) as they usually utilise the provisions and/or genetic services of their neighbouring countries. From one country, Azerbaijan, no information was available (Table 1) and for Luxembourg information was drawn from the data listed in the PQD. In Table 2 aggregated data on the year of recognition of specialisation and/or subspecialisation is presented, and in Table 3 the length of training is summarised (data drawn from the Table 4).

DISCUSSION

At present all but two European countries, Greece and Cyprus, have recognised the MG speciality; Belgium and Croatia as late as this year. Seventeen countries recognised MG as a subspecialty before 2000, and nine of them later changed the status to a stand-alone, that is, primary, speciality. This recognition went slowly until the turn of the millennium, when the scientific progress in human molecular genetics made the discipline an important partner in the development of health care and further evolved with the concept of personalised (stratified or precision) medicine. Full recognition was adopted in 21 countries, that is, about half of the Council of Europe member states, after 2000 (see above).

Training requirements for specialisation varies between the countries ranges from 3 to 6 years, the most common duration being 4 years which is also the minimum length stipulated by PQD. The content of training varies between countries, especially regarding the amount of laboratory competence needed and requirement of clinical electives in other related medical specialties (eg, gynaecology, neurology and/or paediatrics). In spite of the different languages and the varying tasks of a specialist in MG in different European countries, we have now the possibility of working in many different settings and environments, an opportunity that we hope many young doctors will take advantage of.

MG also aims to collaborate closely with the two other professional branches involved in genetic services, clinical laboratory geneticists and genetic nurses and counsellors, under the auspices of the European Board of Medical Genetics (EBMG).[21] This independent board was established in 2012 to serve the needs of patients through establishing standards of practice in all professional branches providing genetic services, and to ultimately issue professional certifications.

Finally, recognition of MG will also aid implementation of Articles 54 and 55 of Directive 2011/24/EU of the European Parliament and of the Council on the application of patients' rights in cross-border healthcare,[22] which provide special provisions for RD and was seminal for the development of ERNs, where MG is embedded as a core specialty in the majority of their cross-border, interdisciplinary research and diagnostic activities.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

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