



Presidential address

Feliciano J. Ramos, MD PhD

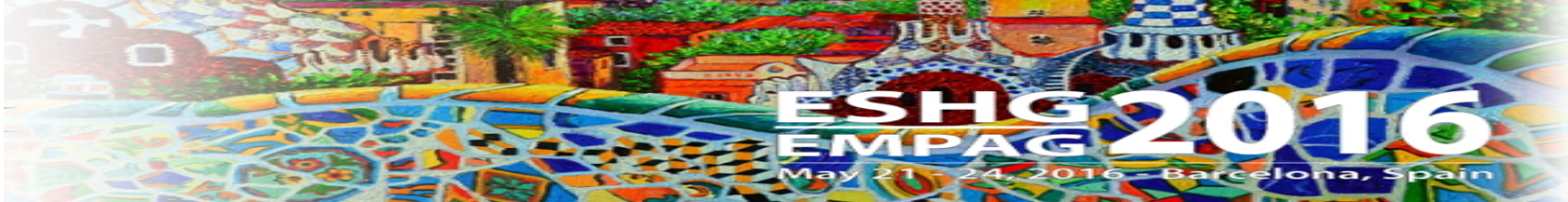
May 21, 2016



Disclosure Information

Feliciano J. Ramos, MD PhD

I have no financial relationships to disclose

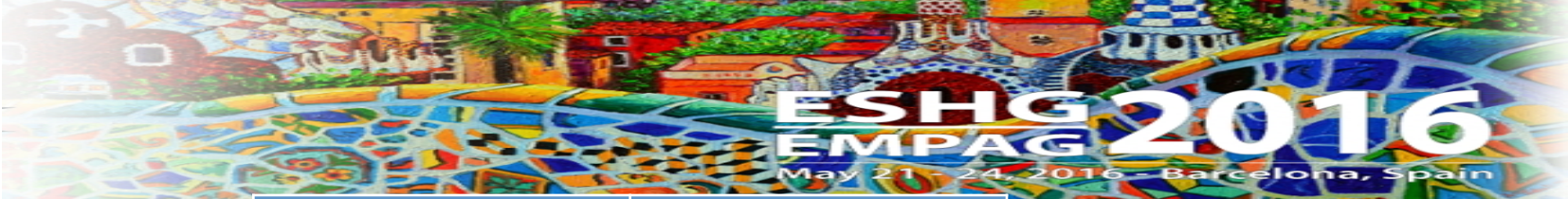


European Society of Human Genetics



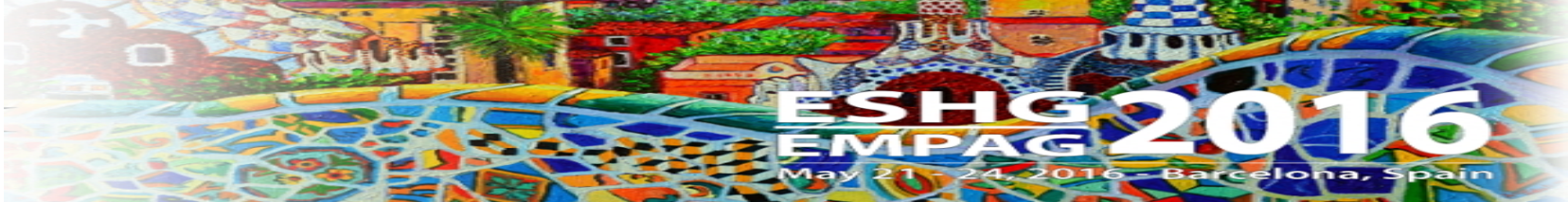
www.eshg.org

- 2.013 members
- 74 countries (36 non-EU) ▶
- Combined memberships:
(EU National Societies / ASHG / Collective)
- Five ESHG Committees
- Embraces EU-National HG Societies
(EU-NHGS' Presidents meet at the Annual Conference)



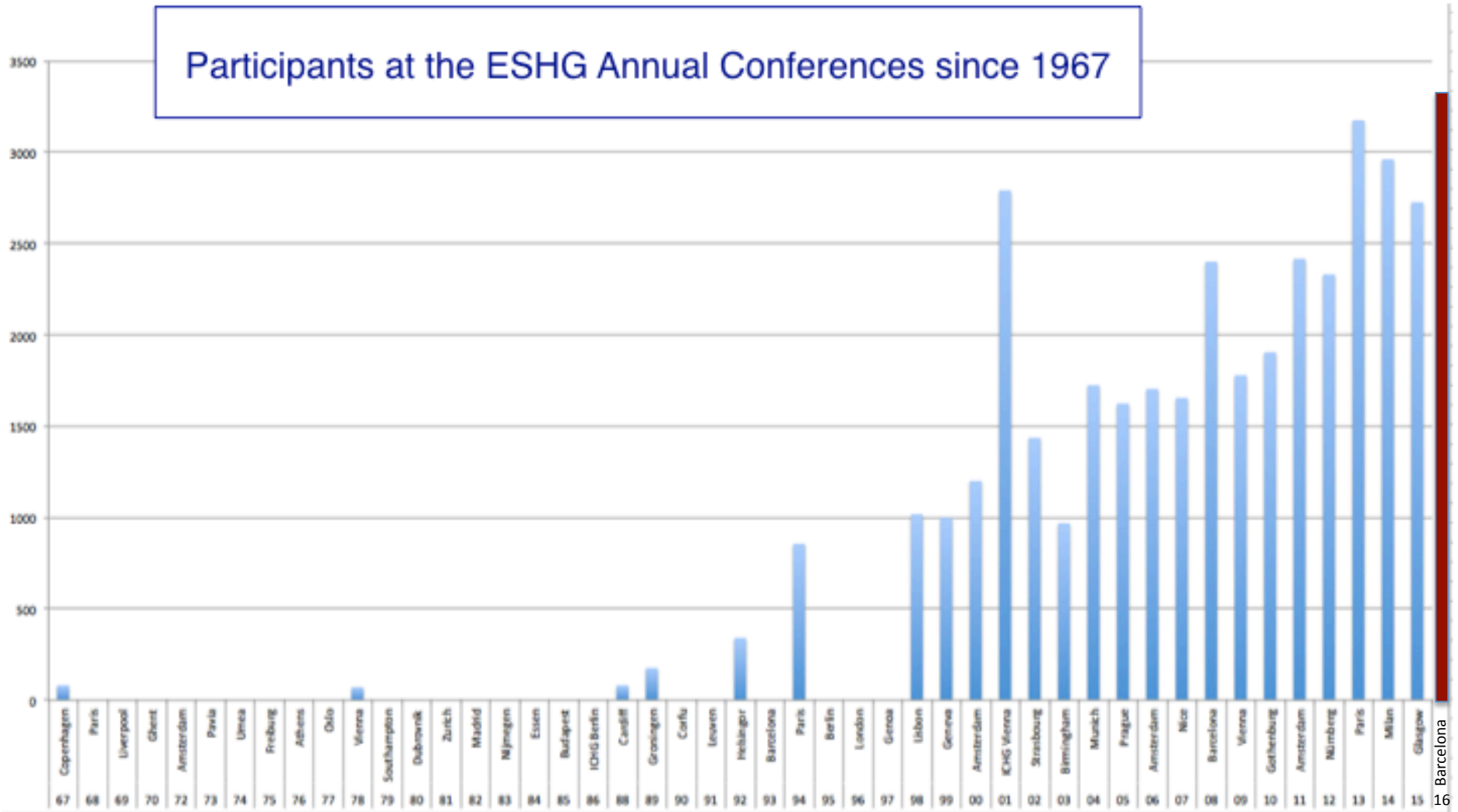
| COUNTRY | Nº members |
|----------------|------------|
| Spain | 287 |
| Romania | 167 |
| Netherlands | 125 |
| United Kingdom | 115 |
| United States | 111 |
| Germany | 100 |
| Italy | 83 |
| Belgium | 76 |
| Turkey | 73 |
| Switzerland | 65 |
| Poland | 58 |
| France | 54 |
| Czech Rep. | 52 |
| Austria | 34 |
| Russia | 34 |
| Finland | 33 |
| Norway | 33 |
| Australia | 32 |
| Canada | 31 |
| Japan | 29 |





European Society of Human Genetics

Participants at the ESHG Annual Conferences since 1967





EU-National Human Genetics Societies

- Started in 2006 (1st Meeting) – Initiative of Prof. *P. Franco Pignatti (ITA)*
- 39 countries represented, 49 HG Societies members
- Presidents of HG National Societies
- Voluntary membership. No fee.
- Principal aims:
 - 1) Coordination among EU National HG Societies
 - 2) Reach all european countries
 - 3) Seek NHGS endorsement of critical aspects of healthcare related to our field



EU recognition Specialty of Medical Genetics

L 59/4

EN

Official Journal of the European Union

4.3.2011

COMMISSION REGULATION (EU) No 213/2011

of 3 March 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

(Text with EEA relevance)

THE EUROPEAN COMMISSION,

training in more than two fifths of the Member States,
which justifies its inclusion into point 5.1.3 of Annex V
to Directive 2005/36/EC.

Having regard to the Treaty on the Functioning of the European
Union,

(5) In order to ensure a sufficiently high level of specialist
medical training, the minimum period of training
required for the medical specialty of medical oncology
to be automatically recognised should be five years.

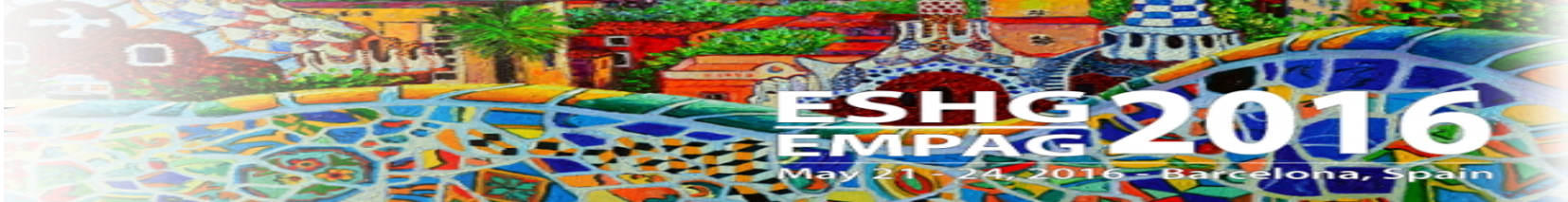
Having regard to Directive 2005/36/EC of the European
Parliament and of the Council of 7 September 2005 on the
recognition of professional qualifications ⁽¹⁾, and in particular
the second paragraph of Article 11 and the second paragraph
of Article 26,

(6) France has submitted a reasoned request to include in
point 5.1.3 of Annex V to Directive 2005/36/EC
specialist medical training in medical genetics.

Whereas:

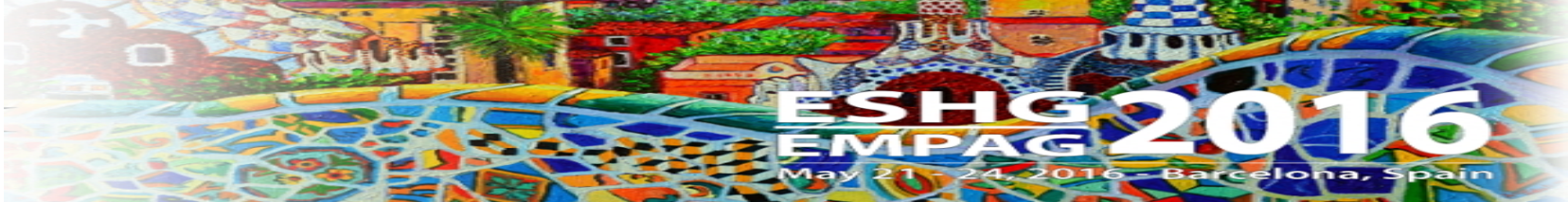
(7) **Medical genetics** is a specialty that responds to the rapid
development of knowledge in the field of genetics and its
implication in numerous specialised fields, such as
oncology, foetal medicine, paediatrics, chronic diseases.
Medical genetics plays a growing role in screening and

(1) Austria has requested the insertion in Annex II to
Directive 2005/36/EC of 10 training programmes



ESHG Committees

- **Education Committee (EC)**
 - Chair: *Prof. Han Brunner* (NL)
- **Public and Professional Policy Committee (PPPC)**
 - Chair: *Prof. Martina Cornell* (NL)
- **Annual Meeting Committee (AMC)**
 - Chair: *Prof. Andrew Read* (UK)
- **Scientific Programme Committee (SPC)**
 - Chairs: *Prof. Brunhilde Wirth* (GER) and *Dr. Joris Veltman* (NL)
- **ESHG-EuroGentest Committee**
 - Chair: *Dr. Hans Scheffer* (NL)



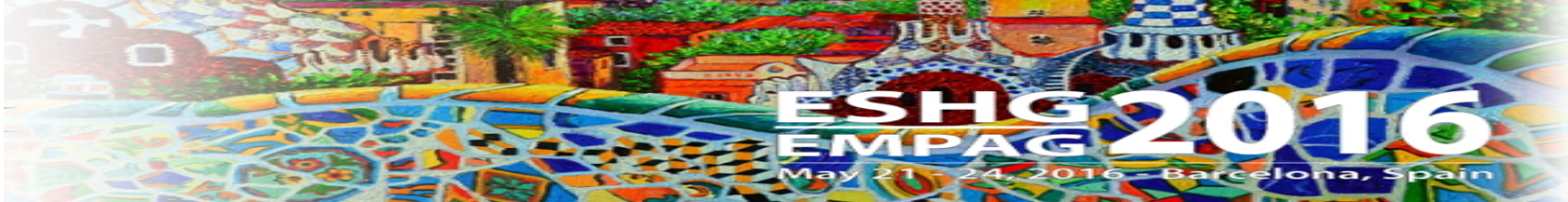
ESHG - Education Committee Activities

- Educational efforts for different audiences:
General public, students and professionals
 - Educational Sessions at the Annual Conference
 - Classic Courses: “Bertinoro” (ITA) and “Manchester Dysmorphology” (UK)
 - New courses in different thematic areas (i.e. hereditary cancer, cardiac genetics, statistical genetics, etc.)
 - DNA-day contest throughout Europe
 - School one-day event (organized by local host in collaboration with ESHG)
- Fellowships for students (developing EU countries)



ESHG – PPC main activities

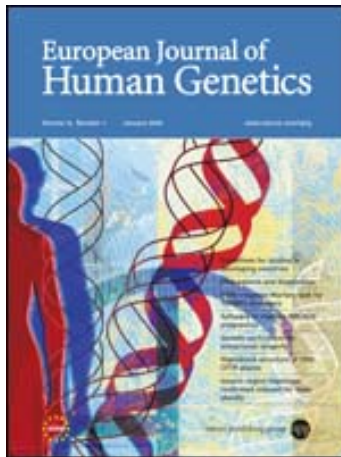
- Contribute to responsible translation of advancements in human genetics to society
 - Identify and discuss ethical, social and policy issues related to HG and its application in clinical/laboratory practice, and research
 - Provide professional guidance through baseline documents, policy statements and recommendations
 - Inform, advise and interact with policy-makers in issues related to HG
- Production of documents of interest related to HG
- Collaborate with other societies/organizations (ASHG, EMPAG, ESHRE, etc.)



ESHG – Scientific Program Committee (SPC)

- Select the most relevant new science in the field of HG to present during the Annual Conference
 - Design the Program
 - Nominate invited speakers
 - Review submitted abstracts -> Selection for oral/poster
 - Selection of travel grants
 - Selection of “Young Scientists Awards”
 - Select the person of the ESHG award
- Facilitate development of networking and contacts among geneticists (Workshops)

European Journal of Human Genetics - EJHG



- Official publication of ESHG
- Editor-in-Chief: *Prof. G. Van Ommen* (NL)
- 2014 Impact Factor: 4,349 – 1st Q
- Printed and on-line version
- 12 issues / year
- Publishes original research, short reports, reviews and special series (p.e. *Clinical Utility Gen Cards, Practical Genetics*).



nature.com | Publications A-Z Index | Browse by subject | ESHG

European Journal of Human Genetics

Volume 24, No 4
April 2014
ISSN: 1021-8413
EISSN: 1476-5438

2014 Impact Factor: 4,349*
70,289 Biotechnology & Molecular Biology
36/187 Genetics & Heredity
Editor-in-Chief: G-J B van Ommen

2014 Journal Citation Reports® Science Edition (Thomson Reuters, 2015)

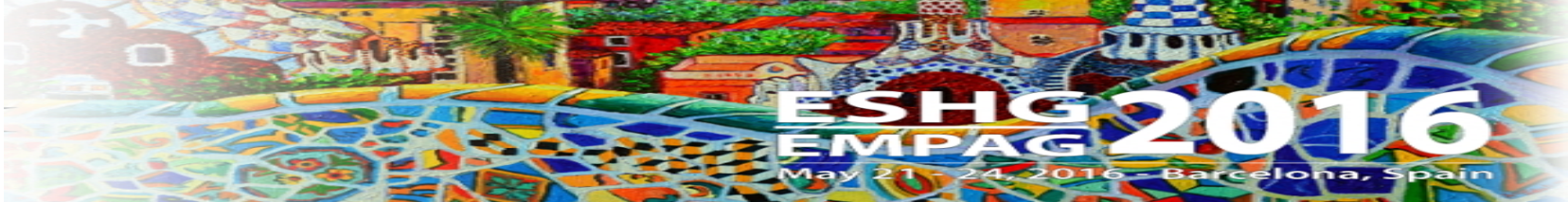
31 March 2016

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European Board of Medical Genetics - EBMG

- Professional body created in 2012
- Include 3 professional groups:
 - *Medical Geneticists*
 - *Clinical Laboratory Geneticists*
 - *Genetic Nurses/Counsellors*
- Registration process ongoing in all groups
- Elaborate syllabuses / training programs





The European Society of Human Genetics

is a non-profit organization. Its aims are to promote research in basic and applied human and medical genetics, to ensure high standards in clinical practice and to facilitate contacts between all persons who share these aims, particularly those working in Europe. The Society will encourage and seek to integrate research and its translation into clinical benefits and professional and public education in all areas of human genetics.



News

Guidelines for diagnostic next-generation sequencing

The EuroGentest Committee of the ESHG has developed Guidelines for diagnostic next-generation sequencing published in the European Journal of Human Genetics.

More information

EMA - 2 Calls for collaboration

The European Medicines Agency is launching a consultation to implement the

News

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- [ESHG Newsletters](#)
- [European DNA Day](#)

European Journal of Human Genetics

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Meetings - Courses

- [ESHG 2016](#)



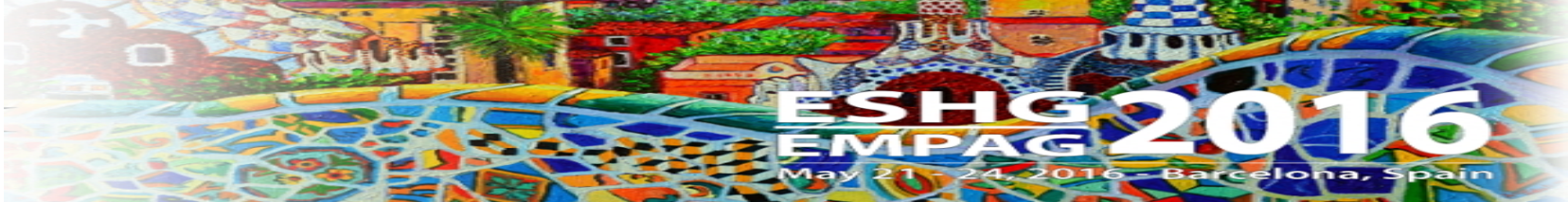
Commission Expert Group on Rare Diseases

Recommendation on

CROSS BORDER GENETIC TESTING OF RARE DISEASES IN THE EUROPEAN UNION

13 November 2015

1. Obtaining an **accurate and timely diagnosis is a priority** for all people with a potentially genetic RD; therefore, access to genetic testing -whether provided locally or on a cross-border basis - should be ensured, to facilitate such diagnoses, when there is a clear clinical indication.
2. The expert group underlines the **importance of assessing genetic testing**, on the basis that early diagnosis through clinically-guided genetic testing may avoid the need for further invasive and/or unnecessary exploratory and therapeutic procedures.
3. Whether genetic testing is provided on the national/regional level or on a cross-border basis, expertise should be **shared at the EU (or global) level**.
4. Appropriate **information on genetic testing laboratories should be made available to facilitate cross-border genetic testing** of rare diseases, particularly when pertaining to the quality of laboratories.



Centers of Expertise

European Reference Networks



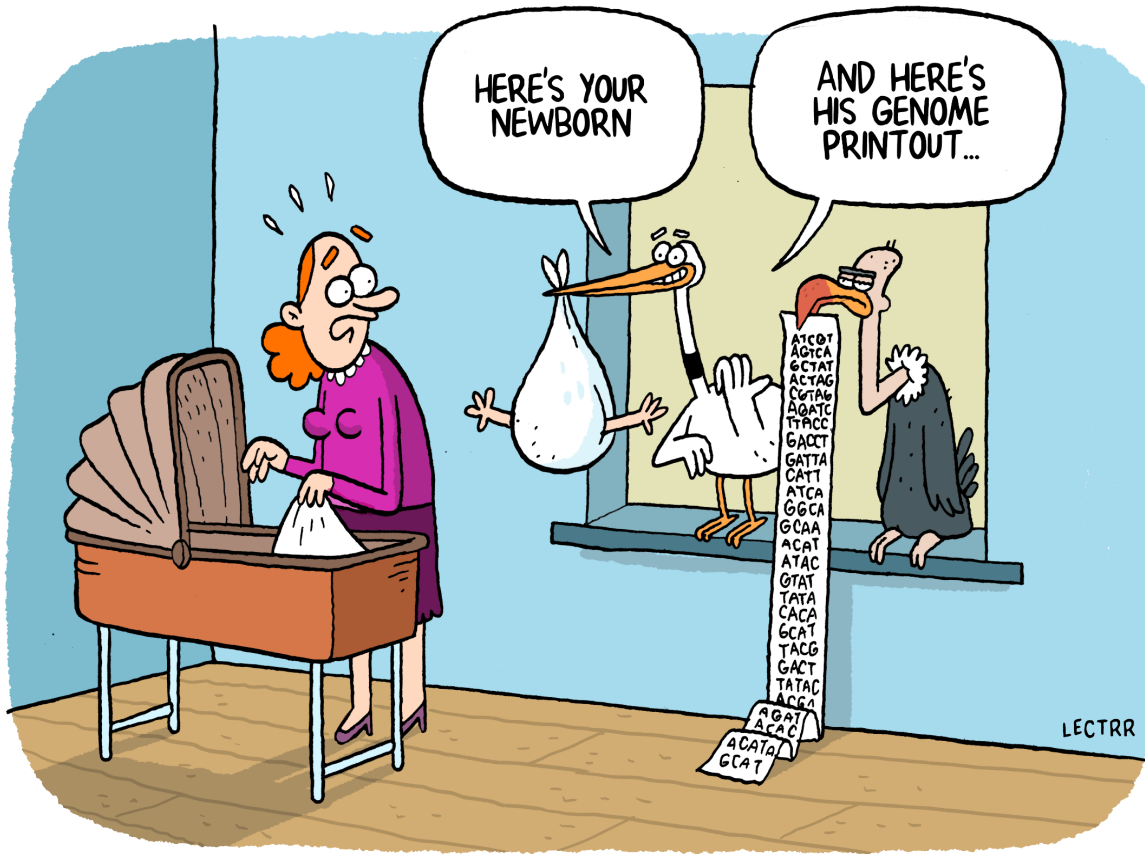
Genetics in Europe: Future challenges for ESHG

- Official recognition of Specialty in ALL european countries (lacking in Greece, Belgium, Cyprus, other...)
- Homogeneization of training programs in HG
- European exam (EBMG)
- Practical implementation of “Cross-border Genetics” between Health Care systems of member states
- Educational activities/courses (physical/on-line webinars):
Excellence, available to the most needed
- Legal/Ethical issues (germline cell genetics, NIPDT ...)

✓ *Other challenges still to come...*

Human Genetics and cartoons

Main Entrance Hall



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Thank you !

Gracias!