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EUROPEAN SOCIETY OF HUMAN GENETICS

PEC REPORT 2025

Francesca Forzano, Chair

Outline

1. Activities, accomplished
2. Activities, ongoing
3. Activities, future

Activities Accomplished 1/3

Recommendations on Cascade Counselling and Testing Cascade

approved by the Board and submitted to publication to the EJHG

GDPR and what does it mean in practice for our genetic community.

Points to consider at the PEC review for final approval.

Survey on Opportunistic genomic screening.

Survey across European countries to understand the impact of our Recommendations on Opportunistic Genomic Screening. The results will be presented at the ESHG 2025 conference (poster) and subsequently published

Activities Accomplished 2/3

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Course: Covering The Gaps – The Course You Didn't Know You Needed

Co-Directors Francesca Forzano and Angus Clarke

Pilot on October 28-29 2024. Virtual, 2 days

Addressed to young scientists and clinicians

Topics: quality, regulation, research ethics, justice in diagnostics and research, data sharing and biobanks

Very positive feedbacks!

Second edition November 27-28 2025

Activities Accomplished 3/3

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Workshop on Rare Diseases, Genomics and Justice

Brocher Foundation. Collaboration between PEC – leader **Angus Clarke** - and a research team led by Ramona Moldovan funded by MRC UK.

The outcomes of the workshop will be published on a special issue of the Journal of Community Genetics, including a meeting summary and the key recommendations.

Activities Ongoing 1/4

Misuse of genetics for discrimination purposes – *delayed*
Lead: Yves Moreau
Advanced draft

Updated recommendations on testing in children.
Lead: Christophe Cordier

Activities Ongoing 2/4

Recommendations on performing and reporting NGS tests in prenatal diagnosis.

Lead: Sandi Deans

Joint activity PEC– Eurogentest - ISPD

Activities Ongoing 3/4

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Policy paper on how to manage the IF of HTT expansion?

Joint activity PEC– European Huntington Disease Network

Activities Ongoing 4/4

Under the radar:

USA: massive cuts to research fundings, sudden termination of the US Advisory Committee on Heritable Disorders in Newborns and Children, etc. Possible actions to support ASHG/ACMG statements and initiatives

Greece: programmatic agreement between Ministry of Health and 2 private American companies for genome screening of newborns 2025-2029 – no public consultation, no consent, companies would retain samples and data. Scientific Council of the ICH expressed serious reservations and has not granted permission

Joint Statement of the Hellenic Society of Medical Genetics and the Hellenic Neonatal Society released.

Activities, Future

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AI in genomics

Lead: Heidi Howard. Joint activity with GA4GH

Genetic testing and insurances.

Possible survey to understand what is currently happening across Europe in relation to use of genetic testing for modulating the insurance premium, access to genetic tests results from insurances, and offer of genetic tests from insurances.

Expanding Quality assessments.

Consideration for workshop.

New technologies(eg gene editing). Ethical implications of activities. Sustainability.

Members of the PEC 2024/2025

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Francesca Forzano (London, UK) - Chair
Carla van El (Amsterdam, The Netherlands) - Secretary General

Christophe Cordier (Lausanne, Switzerland)
Angus Clarke (Cardiff, United Kingdom)
Guido de Wert (Maastricht, The Netherlands)
Sabine Hentze (Heidelberg, Germany)
Heidi Howard (Uppsala, Sweden)
Milan Macek (Prague, Czech Republic)
Bela Melegh (Pecs, Hungary)
Alvaro Mendes (Porto, Portugal)
Yves Moreau (Leuven, Belgium)
Markus Perola (Helsinki, Finland)
Inga Prokopenko (Guildford, Surrey, UK)
Emmanuelle Rial-Sebbag (Toulouse, France)
Rosaline Favresse(Paris, France)
Vigdis Stefánsdottir (Reykjavik, Iceland)
Fiona Ulph (Manchester, UK)

Observers: Olga Antonova (Sofia, Bulgaria), Yalda Jashmidi (London, UK), Elena Avram (Bucharest, Romania); Rhys Dore (London, UK – ESHG-Y)