ESHG

EUROPEAN SOCIETY OF HUMAN GENETICS

PPPC REPORT 2020-2022

Francesca Forzano, Chair

The Public and Professional Policy Committee (PPPC)

Outline

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

Activities Accomplished 1/4

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Recommendations on Opportunistic Genomic Screening
Lead Guido de Wert. (+Eurogentest Committee)

Published Eur J Hum Genet 2021 Mar;29(3):365-377. Epub 2020 Nov 22.
Dissemination activities: ASHG 2020 Building Bridges session

2020 APHRS/HRS Expert Consensus Statement on Investigation of Patients with Sudden Cardiac Arrest and of Their Families

Prof Martina Cornel contributed as invited ESHG/PPPC representative
Document has been endorsed by ESHG Board on October 8th 2020
Published Open Access on 19 October 2020 on Journal of Arrhythmia
https://doi.org/10.1002/joa3.12449

Activities Accomplished 2/4

Germline Gene Editing

- Public engagement online event had been organized in Germany after the ESHG meeting 2020. Partnership with the German Society of Human Genetics and a dutch team led by Sam Riedijk.
- Dutch, German and English translation of tools available
- Further online events under consideration

Consultation WHO Advisory Committee Human Genome Editing on Developing Global Standards for Governance and Oversight of Human Genome Editing - call for contribution (2020)

https://www.who.int/ethics/topics/human-genome-editing/consultation-2020/en/
PPPC has sent a reply drafted by **Heidi Howard**

Activities Accomplished 3/4

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Policy paper

ESHG warns against misuses of genetic tests and biobanks for discrimination purposes

Maurizio Genuardi, Francesca Forzano, Yves Moreau

Published EJHG 2021 Jun;29(6):894-896. Epub 2021 Jan 18
doi: 10.1038/s41431-020-00786-6

Activities Accomplished

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The use of polygenic risk scores in pre-implantation genetic testing: an unproven, unethical practice

Policy paper published EJHG (2022) 30:493–495, online 17.12.2021

Activities Accomplished

The use of polygenic risk scores in pre-implantation genetic testing: an unproven, unethical practice

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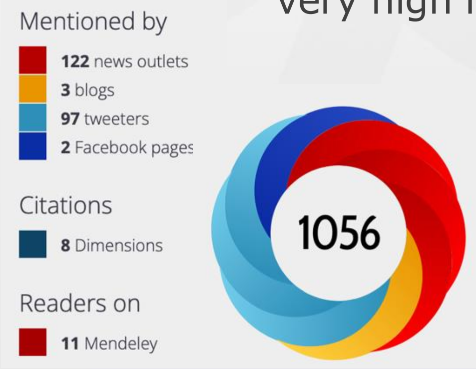
Science Media Center interview with FF, MG, MP

further interviews with science journalists

very high interest, ignited debate on PGT-P and PGT-A



Mentioned by

🟥 **122** news outlets
🟧 **3** blogs
🟦 **97** tweeters
🟦 **2** Facebook pages

Citations

🟦 **8** Dimensions

Readers on

🟥 **11** Mendeley

09.06.2022

ALL RESEARCH OUTPUTS	OUTPUTS FROM EUROPEAN JOURNAL OF HUMAN GENETICS	OUTPUTS OF SIMILAR AGE	OUTPUTS OF SIMILAR AGE FROM EUROPEAN JOURNAL OF HUMAN GENETICS
#10,318	#1	#528	#1
of 21,347,849 outputs	of 3,292 outputs	of 404,214 outputs	of 35 outputs

Demographic breakdown

Type	Count	As %
Members of the public	53	55%
Scientists	34	35%
Science communicators (journalists, bloggers, editors)	5	5%
Practitioners (doctors, other healthcare professionals)	5	5%

Activities Ongoing 1/4

GDPR applied to human genetics (clinics, research) - *delayed*

Lead: Emmanuelle Rial-Sebbag.
In collaboration with Eurogentest Committee (Els Dequeker)
External experts: Edward Dove, Deborah Mascalzoni, and Colin Mitchell from PHG Foundation (Cambridge)

Aim: analyze the impact of the GDPR on genetic practice and research, offer some pragmatic guidance on the do's and don'ts

We hope to submit the draft for membership review after summer 2022

Activities Ongoing 2/4

Cascade Testing: a strategy still underexploited across Europe?

delayed

Lead: Guido deWert

Collaborators PPPC alumnae Lisbeth Tranebjaerg, Hülya Kayserili, Florence Fellmann

Aim: analyse barriers for implementation in practice and provide recommendations.

The draft has been restructured. We aim to submit it for membership review shortly.

Activities Ongoing 3/4

Opportunistic Genomic Screening, survey

Lead: Heidi Howard

Scope: assessing impact of ESHG Recommendations in practice
To be launched at ESHG2022

Activities Ongoing 4/4

Misuse of genetics for discrimination purposes

Lead: Yves Moreau
External collaborators: Barbara Prainsack, Sam Riedijk (board member), Denise Syndercombe-Court, Gudrun Rappold, Veronika Lipphardt

Scope and aim:

- drafting some general **principles** for ethical conduct, illustrated by specific examples how practices involving genomic data can lead to unethical and undesired outcomes (misuse in terms of discrimination & exploitation)
- share and help discuss the lessons we can learn about these by formulating **recommendations** for institutions and individual conduct

Joint activities

- **Eurogentest.** One joint document ongoing (GDPR).
  Future joint documents: Reporting in prenatal setting; IVD.
- **Education Committee.** One joint document ongoing: guidelines on best practice on the use of language when addressing patients with RD, to promote a more respectful and inclusive attitude.
- **IFHGS**. We have submitted a session proposal to IFHGS Congress 2023 for a joint session with the other societies on privacy, consent and data sharing. Accepted.
- **ESHRE**. Expanded Carrier Screening survey. Ongoing. Closed 11.04.2022 (8 weeks); 338 replies received, 299 valid.

Collaboration with EURORDIS

Key principles for newborn screening

PPPC representative Francesca Forzano invited to contribute to the Position statement

Screen4Care

EURORDIS chairs the Patient Advisory Board of this project and has welcomed 2 PPPC representatives (Vigdis Stefansdottir, Fiona Ulph)

https://www.screen4care.eu/people-living-with-rare-diseases/involvement

Edith Sky Gross (Barcelona, Spain), EURORDIS, has joined PPPC as a **new member** as patients and public representative

Activities, Future 1/2

Just Genomics : Avoiding harm and inequities arising from genomics
Lead: Angus Clarke, Carla van El, Heidi Howard.

We have submitted an application to COST Action, which has been unsuccessful. Alternative funding options under consideration.

Reporting in prenatal setting (suggested by exec)

Quality and Guideline committees (Eurogentest) lead, project not yet started.

Could be integrated in a previous document on reporting genomic tests or a separate one.

Consideration for a viewpoint on NBS by WGS.

Consideration for a viewpoint/position statement on feedback of results from biobanks.

Activities, Future 2/2

Proposal for a new ESHG course

Proponents: Francesca Forzano and Angus Clarke

Title: *Things you need to know but nobody told you*.

Format: virtual, over 2 days. Sponsorship by ESHG requested – submitted to EC for their review.

Purpose:

- ➢ Provide participants with information that is not generally taught in formal academic programmes
- ➢ Encourage participants to reflect on the importance of the '*how*', including effective communication of research results/ new technologies/ treatments etc. to peers and lay public, and the importance to take into account the potential public perception of ethical and related 'political' issues in genetics
- ➢ Provide a framework on how to address ethical challenges in clinical or research settings.

Members of the PPPC 2021/2022

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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)
Dragica Radojkovic (Belgrade, Serbia)
Emmanuelle Rial-Sebbag (Toulouse, France)
Edith Sky Gross (Barcelona, Spain)
Vigdis Stefánsdottir (Reykjavik, Iceland)
Fiona Ulph (Manchester, UK)

Observers: Olga Antonova (Sofia, Bulgaria), Yalda Jashmidi (London, UK)

https://www.eshg.org/index.php?id=pppc