

Report of the Public & Professional Policy Committee



ESHG

Barcelona

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& public health genomics

On behalf of PPPC

Quality of Care

Community Genetics, Dept Clinical Genetics
EMGO Institute for Health and Care Research



The Professional and Public Policy Committee aims:

- To **identify and discuss the ethical, social and policy issues** related to human genetics and its application in research, clinical practice and laboratory genetic services.
- To be informed of various research projects, conferences and events, as well as policy initiatives and actions relating to those issues
- To **inform and stimulate the discussion** around these issues at meetings
- To address these issues and **provide guidance** through background documents, policy statements, recommendations or other publications
- To **participate in the public debate** around these issues
- To inform, interact with and provide advice to national and international policy makers

Activities 2015/16

- **expanded carrier screening** (consultation until March 15, 2015)
- detection of carrier status (AR diseases) in persons who do **not** have an a priori increased risk
- greater number of recessive conditions than previously possible
- aim is to facilitate informed reproductive decision-making by identifying couples at risk
- Discussion: selection of disorders, timing, information and counselling, and informed consent

POLICY

Responsible implementation of expanded carrier screening



Lidewij Henneman¹, Pascal Borry², Davit Chokoshvili^{2,3}, Martina C Cornel¹, Carla G van El¹, Francesca Forzano⁴, Alison Hall⁵, Heidi C Howard⁶, Sandra Janssens³, Hülya Kayserili⁷, Phillis Lakeman⁸, Anneke Lucassen⁹, Sylvia A Metcalfe¹⁰, Lovro Vidmar¹¹, Guido de Wert¹², Wybo J Dondorp¹² and Borut Peterlin^{*,11} on behalf of the European Society of Human Genetics (ESHG)

This document of the European Society of Human Genetics contains recommendations regarding responsible implementation of expanded carrier screening. Carrier screening is defined here as the detection of carrier status of recessive diseases in couples or persons who do not have an *a priori* increased risk of being a carrier based on their or their partners' personal or family history. Expanded carrier screening offers carrier screening for multiple autosomal and X-linked recessive disorders, facilitated by new genetic testing technologies, and allows testing of individuals regardless of ancestry or geographic origin. Carrier screening aims to identify couples who have an increased risk of having an affected child in order to facilitate informed reproductive decision making. In previous decades, carrier screening was typically performed for one or few relatively common recessive disorders associated with significant morbidity, reduced life-expectancy and often because of a considerable higher carrier frequency in a specific population for certain diseases. New genetic testing technologies enable the expansion of screening to multiple conditions, genes or sequence variants. Expanded carrier screening panels that have been introduced to date have been advertised and offered to health care professionals and the public on a commercial basis. This document discusses the challenges that expanded carrier screening might pose in the context of the lessons learnt from decades of population-based carrier screening and in the context of existing screening criteria. It aims to contribute to the public and professional discussion and to arrive at better clinical and laboratory practice guidelines.

Endorsement

- Discussed with the new Professional Guidelines sub-committee of ESHG-EuroGentest
- Officially endorsed ESHG recommendations follow a procedure of
 - agenda setting,
 - preparation in a multidisciplinary setting,
 - consultation of ESHG membership and external experts,
 - integration of suggestions and
 - voting by Board

Post mortem genetics

ch/fr/events/228/ethical-legal-an   brocher geneva cardiogenetic... Fondation Brocher | Ethical,...

Événements

- Événements à venir
- Événements passés
- Conditions
- Appel à projets

Université d'été ou d'hiver

Appels à projets



L'utilité de ce genre d'institutions est incontestable. Car le monde moderne est sans cesse confronté à des innovations, médicales ou autres, qui s'appliquent à l'homme ou à son environnement proche. Ce lieu est donc nécessaire pour préparer la matière intellectuelle qui sera ensuite transférée aux citoyens afin que ceux-ci puissent en tirer profit.

23 - 25 novembre 2016

Ethical, legal and practical aspects of post-mortem genetic analysis for sudden cardiac death in young adults

Organisateurs:

- Cornel Martina
- Rial-Sebbag Emmanuelle, INSERM, UMR 1027, Inserm/Paul Sabatier University Toulouse, PhD, Permanent Research fellow
- Fellmann Florence, CHUV Lausanne, Dr. Medical Geneticist

During this workshop, we will discuss the ethical, legal and practical issues of conducting post-mortem genetic testing with the ultimate aim of drafting European recommendations. We will focus on the case of sudden cardiac death (SCD) in young adults (less than 40 years of age) and aim to elaborate recommendations to aid in the inclusion of such testing in autopsy procedures in Europe. To date, attempts at incorporating genetic testing for sudden cardiac death in autopsy procedures has faced a number of challenges, including legal and ethical challenges, as well as very practical issues such as financial questions (who will pay for this?) and (mis)communication obstacles (pathologists and geneticists being unaware of the issues and unable to discuss and find applicable solutions).

We will gather an international multidisciplinary group of experts to discuss these challenges in order to find potential solutions. Clear and constructive recommendations on post-mortem genetic testing for SCD will not only help in the context of this disorder but may also prove to be useful for other medical disorders.

New activities

- Gene editing (incl CRISPR/CAS9)
 - Agenda setting, meeting Jan 2016 Zaandam
 - Recommendations with ESHRE, plan to develop draft for consultation by mid Octobre 2016
- Genomic Sequencing Results in Pediatric Practice. Exome sequencing in newborns with complex severe syndromes
 - With EBMG/UEMS
 - Bela Melegh (PPPC-member), Alessandra Renieri, Orsetta Zuffardi and Ulf Kristofferson

For national societies human genetics

- Discuss and disseminate Expanded Carrier Screening Recommendations?
- Suggestions to PPC for
 - activities/
 - topics/
 - experts to collaborate with