JOGNN

RESEARCH

Attitudes of European Geneticists Regarding Expanded Carrier Screening

Sandra Janssens, Davit Chokoshvili, Danya Vears, Anne De Paepe, and Pascal Borry Q5

Correspondence
Sandra Janssens, MD, Centre for Medical Genetics, University Hospital Ghent, De Pintelaan 185, 9000 Ghent, Belgium.
Sandra.jansses@ugent.be

Keywords
attitudes
carrier screening
ethics
qualitative research
recessive disorders

ABSTRACT

Objective: To explore attitudes of clinical and molecular geneticists about the implementation of multi-disease or expanded carrier screening (ECS) for monogenic recessive disorders.

Design: Qualitative; semistructured interviews.

Setting: In person or via Skype. Interviews were audiorecorded and transcribed verbatim.

Participants: European clinical and molecular geneticists with expertise in carrier screening ($N = 16$).

Methods: Inductive content analysis was used to identify common content categories in the data.

Results: Participants recognized important benefits of ECS, but they also identified major challenges, including limited benefit of ECS for most couples in the general population, lack of knowledge on carrier screening among nongenetic health care providers and the general public, potential negative implications of ECS for society, and limited economic resources. Participants favored an evidence-based approach to the implementation of population-wide ECS and were reluctant to actively offer ECS in the absence of demonstrable benefits. However, there was a consensus among the participants that ECS should be made available to couples who request the test. In addition, they believed ECS could be routinely offered to all people who use assisted reproduction.

Conclusion: Although a limited ECS offer is practical, it also raises concerns over equality in access to screening. A comprehensive risk–benefit analysis is needed to determine the desirability of systematic population-wide ECS.

JOGNN, ■, ■–■; 2016. http://dx.doi.org/10.1016/j.jogn.2016.08.012

Accepted August 2016

Sandra Janssens, MD, is a clinical geneticist in the Centre for Medical Genetics, University Hospital Ghent, De Pintelaan, Ghent, Belgium.

Davit Chokoshvili, MA, MSc, is a doctoral researcher in the Centre for Biomedical Ethics and Law, University of Leuven, Leuven, Belgium.

Danya Vears, PhD, is a postdoctoral researcher in the Centre for Biomedical Ethics and Law, University of Leuven, Leuven, Belgium.

(Continued)

The authors report no conflict of interest or relevant financial relationships.

The purpose of carrier screening is to identify couples at risk of conceiving a child with a monogenic recessive disorder. This risk is present when both reproductive partners carry a mutation associated with the same autosomal recessive disorder or when the woman is a carrier of an X-linked disorder (Wienke, Brown, Farmer, & Strange, 2014).

Because of the recessive pattern of inheritance, many carriers of these disorders have no family history suggestive of the condition. Once identified, at-risk couples have the option to act on this information and may alter their reproductive plans (Ropers, 2012).

In some countries and ethnic communities with a high birth prevalence of severe recessive disorders, carrier screening programs were introduced as early as the 1970s. Notable examples of the first screening programs include Tay-Sachs carrier screening in the Ashkenazi Jewish community (Kaback, 2000) and premarital screening of couples for beta-thalassemia in the Mediterranean region (Cousens, Gaff, Metcalfe, & Delatycki, 2010). Subsequently, carrier screening became available in some countries for conditions such as cystic fibrosis (CF), fragile X syndrome, and spinal muscular atrophy (Metcalfe, 2012).

Because of largely technical limitations, most tests for carrier screening have traditionally been used to target a limited set of pathogenic mutations associated with a single disorder or a small panel of monogenic disorders (Bajaj & Gross, 2014). However, recent advances in molecular diagnostics have resulted in the development of expanded carrier screening (ECS) panels capable of identifying hundreds of mutations implicated in a large number of recessive conditions (Bell et al., 2011; Kingsmore, 2012; Tanner et al., 2014). ECS products are currently available at a price comparable to that of carrier screening for single conditions (Higgins, Flanagan, Von Wald, & Hansen, 2015; Langlois,

FLA 5.4.0 DTD ■ JOGN145_proof ■ 19 November 2016 ■ 7:57 pm ■ ce

Health care providers with expertise in carrier screening are well-positioned to discuss the perceived advantages and disadvantages of a population expanded carrier screening program.

Benn, & Wilkins-Haug, 2015; McGowan, Cho, & Sharp, 2013). The capacity to screen for more disorders for a similar price and the ability to identify carriers regardless of ethnicity constitute major appeals of ECS (Cho, McGowan, Metcalfe, & Sharp, 2013; Lazarin et al., 2012; Ready, Haque, Srinivasan, & Marshall, 2012; Srinivasan et al., 2010). These advantages over traditional forms of carrier screening suggest ECS has the potential for wide implementation in reproductive health care (McGowan et al., 2013). Results of a survey conducted in the United States in 2012 suggested that ECS is already routinely offered by some obstetricians and gynecologists (Benn et al., 2014).

Widespread adoption of ECS will profoundly influence reproductive health care practices and is likely to be associated with significant practical and ethical challenges that will require special consideration. Valuable insights can be gained from exploring the opinions of genetic professionals who have extensive experience with diverse forms of genetic testing (Cho et al., 2013). Here, we report the results of an interview-based study with European clinical and molecular geneticists and present the issues that surround the implementation of ECS in reproductive medicine.

Methods

Because of the explorative nature of our research question, we conducted key informant interviews with clinical and molecular geneticists to investigate their views about the implementation of ECS in reproductive health care (Popay, Rogers, & Williams, 1998). Participants were eligible for inclusion if they were practicing clinical or molecular geneticists based in the European Economic Area and had demonstrable expert knowledge in carrier screening, such as authorship of relevant scientific publications or conference abstracts. Potential participants were identified by members of our research team and invited to participate via e-mail. Additional respondents were recruited by snowball sampling, where we asked our participants to identify colleagues with expertise in carrier screening.

Anne De Paepe, MD, PhD, is a professor in the Centre for Medical Genetics, University Hospital Ghent, and a University rector for Ghent University, Ghent, Belgium.

Pascal Borry, PhD, is a professor in the Centre for Biomedical Ethics and Law, University of Leuven, Leuven, Belgium.

Interviews were conducted using a semi-structured interview guide, which allowed for in-depth exploration of issues related to implementation of ECS (Liamputtong & Ezzy, 2005). Interviews took place between April and September 2014 and were audiorecorded and transcribed verbatim to enable coding and analysis.

Inductive content analysis was used to identify common content categories from the interviews, rather than coding using a predetermined coding scheme (Downe-Wamboldt, 1992; Graneheim & Lundman, 2004; Schamber, 2000). The data were coded into broad categories before sections of the data within these categories were compared and more specific content categories were developed. Coding was performed by DC using the qualitative data management software QSR Nvivo; data were reviewed by all members of the research team for validation. This study was approved by the institutional ethics committee of the University Hospital Ghent.

Results

The group of participants included 16 genetics professionals from eight member states of the European Economic Area. The group included 13 clinical geneticists, 2 molecular geneticists, and 1 medical geneticist with expertise in clinical and molecular genetics. At the time of the interviews, all participants were affiliated with an academic institution, and 12 geneticists (9 clinical geneticists, 2 molecular geneticists, 1 medical geneticist with expertise in clinical and molecular genetics) had more than 20 years of professional experience in clinical or diagnostic practice. Eleven participants were female, and five were male.

Thirteen interviews took place in person, and three were conducted via Skype. Three categories relevant to the implementation of large-scale ECS programs were identified from the data: *Potential benefits of ECS, Challenges of population-wide carrier screening using expanded panels,* and *Models for provision of ECS*. These categories and their subcategories are described below and are accompanied by illustrative quotes from the participants.

Category 1: *Potential Benefits of ECS*

All participants believed that systematically offering preconception ECS to prospective parents would result in significant potential benefits,

FLA 5.4.0 DTD ■ JOGN145_proof ■ 19 November 2016 ■ 7:57 pm ■ ce