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LETTER TO THE EDITOR

Reproductive Endocrinology Infertility (REI) Specialists' Utilization and Attitudes Toward Expanded Carrier Screening (ECS) for Third-Party Oocyte Donors

Tanya L. Glenn[1] · Nigel Pereira[2] · Jody Madeira[3] · Rose A. Maxwell[1] · J. Preston Parry[4] · Heidi Mertes[5] · Guido Pennings[5] · Steven R. Lindheim[1,6,7]

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Dear Editor,

Through this communication, we wish to highlight that genetic testing for inherited conditions is an important and controversial part of preconception planning. Recent technological advancements in high-throughput genotyping and sequencing approaches have allowed providers to screen for many conditions at a reasonable cost [1, 2]. This technique, known as expanded preconception carrier screening (EPCS), provides information regarding more conditions than currently recommended screening guidelines [1, 2].

Multiple professional organizations acknowledge the role of EPCS, including the American College of Medical Genetics and Genomics, American Congress of Obstetricians and Gynecologists, National Society of Genetic Counselors, Perinatal Quality Foundation, and Society for Maternal–Fetal Medicine [3]. However, traditional screening enabled the detection of diseases with a well-defined phenotype with childhood onset. In contrast, several of the diseases EPCS detects have onset later in life creating numerous ethical dilemmas. Therefore, EPCS can present a significant challenge for patient management, particularly in third-party reproduction [4, 5].

Since EPCS has become available, there has been growing demand for more thorough genetic screening of gamete

Tanya L. Glenn was a resident in the Department of Obstetrics & Gynecology at Wright State University in Dayton, OH. Nigel Pereira is an Assistant Professor of Reproductive Medicine and Obstetrics and Gynecology at Weill Cornell University in New York, NY. Jody Madeira is a Professor of Law and Louis F Niezer Faculty Fellow, Co-Director in the Center for Law, Society & Culture at the Maura School of Law at Indiana University in Bloomington, IN. Rose A. Maxwell is an Assistant Professor & Director of Research in the Department of Obstetrics & Gynecology at Wright State University in Dayton, OH. J. Preston Parry is chief physician at Positive Steps Fertility in Madison, MS. Heidi Mertes is a Professor of Ethics in the Department of Philosophy and Moral Sciences at Ghent University and a postdoctoral research fellow of the Research Foundation - Flanders in Ghent, Belgium. Guido Pennings is a Professor of Ethics and Bioethics in the Department of Philosophy and Moral Sciences at Ghent University and Director of Bioethics Institute Ghent in Ghent, Belgium. Steven R. Lindheim is a Clinical Professor at Wright State University in Dayton, OH and chief physician in the Department of Reproductive Medicine Renji Hospital Shanghai Jiaotong University School of Medicine in Shanghai, China.

These findings were presented at the European Society of Human Reproduction and Embryology (ESHRE) 34th Annual Meeting 2018, Barcelona, Spain; as well as the Armed Forces District Meeting 2018, Honolulu, Hawaii.

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✉ Steven R. Lindheim
doclalalindheim@gmail.com

[1] Wright State University Boonshoft School of Medicine, 3640 Colonel Glenn Hwy, Dayton, OH 45435, USA

[2] Weill Cornell Medicine-Reproductive Medicine, 1305 York Avenue, 6th Floor, New York, NY 10021, USA

[3] Maurer School of Law Indiana University, 211 S Indiana Ave, Bloomington, IN 47405, USA

[4] Positive Steps Fertility, 149 Fountains Blvd, Madison, MS 39110, USA

[5] Department of Philosophy and Moral Science, Bioethics Institute Ghent, Ghent University, Ghent, Belgium

[6] Department of Reproductive Medicine Renji Hospital, Shanghai Jiaotong University School of Medicine, Shanghai, China

[7] Department of Obstetrics and Gynecology, Wright State University, 128 Apple Street, Suite 3800 Weber CHE, Dayton, OH 45409, USA

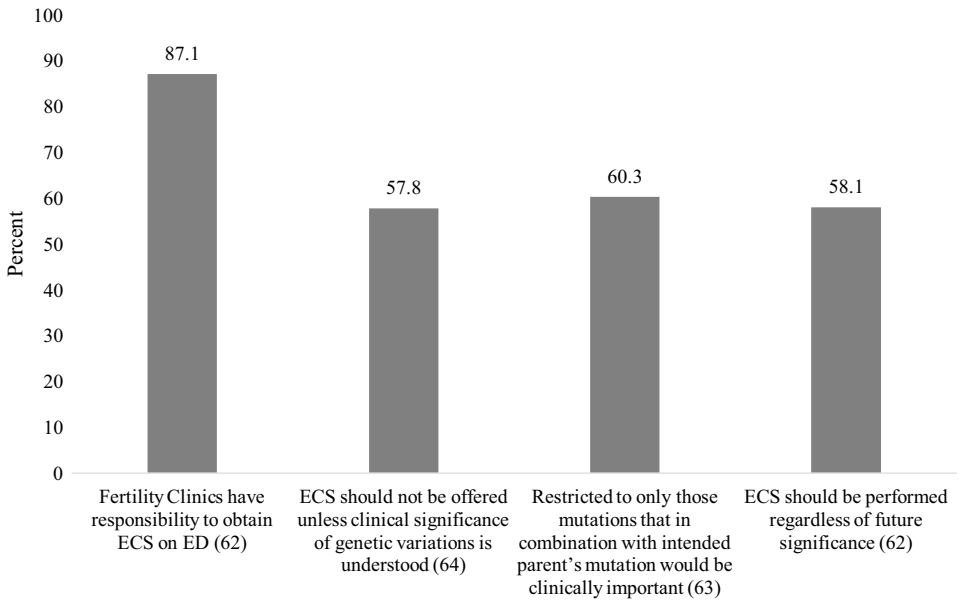

Fig. 1 Principle-based questions

donors, reinforced by reports on the genetic anomalies these individuals may have passed. This has caused an ethical dilemma in determining whom and what to test, as well as who can refuse testing. Moreover, efforts to address these challenges have led to a wide variety of practices. Given these variations in clinical practice and beliefs, we conducted a prospective IRB-approved quantitative survey from February to December 2017 to characterize current EPCS practices related to third-party reproduction to all Society for Reproductive Endocrine Infertility members.

In total, 83 physicians responded (an 11.1% response rate), 94.5% of whom offered oocyte donation. Of these, 42.2% always performed EPCS for oocyte donation, whereas 59% routinely performed EPCS in infertile patients. Following a positive oocyte donor carrier screen, physicians performed counseling 45.6% of the time, with geneticists and nurses offering counseling 34.2% and 12.7% of the time, respectively. Eighty-three percent reported a willingness to use gamete donors who test positive as carriers of autosomal recessive disorders, provided the intended parent(s) test negative for that specific trait.

With respect to principle-based questions, 57.8% felt that EPCS should not be offered unless the clinical significance of genetic variations is understood, and 60.3% felt that EPCS should be restricted to those mutations that are clinically significant when combined with the same parental mutation. The vast majority of respondents (98.4%) believed that results should be shared with oocyte donors, and 54.7% felt that donors could decline genetic testing. Sixty-nine percent of respondents disagreed that using EPCS in third-party reproduction encourages a genetically selected population, yet 3.6% felt that EPCS crossed ethical boundaries (Fig. 1).

With respect to demographics, respondents in practice < 5 years were more likely to feel that ECS should not be offered until the clinical significance of genetic variations is understood, and that it should be restricted to only clinically meaningful mutations (adjusted OR 11.3, 95% CI 0.1.1–115.5). No differences were noted in respondents' gender or age.

EPCS is becoming a standard practice for third-party reproduction; however, there are clearly varying beliefs and practices among REIs and its utilization. This suggests the need to standardize guidelines for EPCS use and guidelines for inclusion and exclusion of gamete donors with a positive carrier status. This need is particularly urgent as genetic testing technology continues to advance, and a wider array of genetic aberrations are discovered.

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Compliance with Ethical Standards

Conflict of interest All authors participated in the final approval of this manuscript.

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Informed Consent This was an optional survey-based study, and no identifying information was disclosed.

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