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American College of Medical Genetics and Genomics (ACMG) Releases Points to Consider Statement on the Safety and Efficacy of Polygenic Risk Score Assessment for Embryo Selection

Bethesda, MD – February 23, 2024 | Should we be using polygenic risk score assessment for embryo selection? Providing in-depth analysis, the Social, Ethical and Legal Issues Committee of the American College of Medical Genetics and Genomics (ACMG) has released a new Points to Consider statement to assist healthcare professionals and patients in understanding the safety and utility of preimplantation genetic testing for polygenic disorders (PGT-P) as a clinical service.

"Clinical Utility of Polygenic Risk Scores for Embryo Selection: A Points to Consider Statement of the American College of Medical Genetics and Genomics (ACMG)" was published in the College's flagship journal, *Genetics in Medicine.*

PGT-P is currently offered by a few commercial labs as a method for prospective parents to screen for common disorders, such as diabetes, cardiovascular disease and some cancers. However, few previously published studies have found this testing to be valid, and none have conducted a robust assessment of its clinical utility.

"While promotion of PGT-P has led to increased demand, this methodology remains unproven," said lead author Theresa A. Grebe, MD, FACMG. "To provide guidance to our patients about the safety and efficacy of this test, we undertook an in-depth analysis of PGT-P. We looked at all aspects of the testing, including prospective studies of outcome and considered various clinical settings in which this testing could be implemented. We determined that at this time there is insufficient evidence of clinical utility of PGT-P. Until further research is done, including an ethical examination, we do not recommend it be offered as a clinical service."

The new ACMG Points to Consider statement outlines the current state of research regarding polygenic risk scores (PRS), the benefits and limitations of PRS testing, aspects of in vitro fertilization that impact preimplantation genetic testing (PGT) in the setting of embryo selection and the challenges in applying PRS data developed in adults to embryo selection. Finally, the statement analyzes the utility of PGT-P in various clinical scenarios.

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The authors recommend that further research and longitudinal studies be conducted before PRS analysis for embryo screening can be responsibly offered. Currently, the risks of harm to the prospective parent or future child outweigh the benefits. The authors also suggest that a cross-section of stakeholders ultimately will need to address PGT-P's broader social, ethical and regulatory issues.

"The last two decades have seen an exponential increase in prenatal screening and testing. It is critical that any new testing modality has clinical utility, does not cause harm to patients and follows ethical principles," said ACMG President Susan Klugman, MD, FACMG, FACOG. "This new tool, preimplantation genetic testing for polygenic disease risk, requires in vitro fertilization and is proposed as an aid in embryo selection. This ACMG statement, based on a review of current data, points out many concerns with the use of PGT-P. New screenings and tests require further research at their introduction into clinical practice. At this time, PGT-P is not recommended for clinical use, as ongoing research and societal guidelines are needed."

About the American College of Medical Genetics and Genomics (ACMG) and ACMG Foundation

Founded in 1991, the American College of Medical Genetics and Genomics (ACMG) is a prominent authority in the field of medical genetics and genomics and the only nationally recognized medical professional organization solely dedicated to improving health through the practice of medical genetics and genomics. The only medical specialty society in the US that represents the full spectrum of medical genetics disciplines in a single organization, the ACMG provides education, resources and a voice for more than 2,500 clinical and laboratory geneticists, genetic counselors and other healthcare professionals. ACMG's mission is to improve health through the clinical and laboratory practice of medical genetics as well as through advocacy, education and clinical research, and to guide the safe and effective integration of genetics and genomics into all of medicine and healthcare, resulting in improved personal and public health. *Genetics in Medicine* and the new *Genetics in Medicine Open*, a gold open access journal, are the official ACMG journals. ACMG's website, www.acmg.net, offers resources including policy statements, practice guidelines, and educational programs. The ACMG Foundation for Genetic and Genomic Medicine works to advance ACMG educational and public health programs through charitable gifts from corporations, foundations and individuals.

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