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Leading Organizations in Prenatal Screening and Diagnosis Issue New Consensus Statement on Genome Sequencing for Fetal Diagnosis

January 9, 2018, Washington DC – Prenatal screening and testing has changed dramatically since the 1970s. The introduction of ultrasound technology and the more recent completion of DNA sequencing of the human genome have led to tremendous advances in prenatal diagnosis. With the availability of genomic sequencing, much more detailed genetic information related to the fetus can now be uncovered. As DNA technologies are developed and applied to prenatal testing, unique challenges and questions arise about their applicability to various populations. In response, three national organizations issued a joint position statement today entitled, “The use of diagnostic genome-wide sequencing for fetal diagnosis.”

The joint position statement was published today online in [Prenatal Diagnosis](#) and was issued jointly by the International Society for Prenatal Diagnosis (ISPD), the Perinatal Quality Foundation (PQF), and the Society for Maternal-Fetal Medicine (SMFM).

The more comprehensive information that is available with sequencing can improve the ability to uncover the cause of birth defects, but the practice is not without challenges and complexities that need to be considered. Outlined in greater detail in the newly released position statement, the consensus opinion for when the genome-wide testing should be considered are summarized:

- 1.) DNA sequencing is beginning to be used for the evaluation of fetuses with suspected genetic disorders for whom standard chromosomal testing has already been performed and is uninformative. In some cases, sequencing may be offered concurrently with standard testing when expert genetic opinion determines that that is unlikely to identify a cause for the presenting fetal phenotype.
- 2.) Routine use of genome-wide sequencing as a diagnostic test is not currently recommended due to insufficient validation data and knowledge about the benefits and pitfalls.

- 3.) Testing is currently best done in trios (both parents and the fetus). Parents require genetic experts to provide pre- and post-test counseling and for result disclosure; many specific counseling points are recommended as minimal for the informed consent process and to enhance the patient/parent understanding.
- 4.) The indications that might warrant consideration of prenatal genomic testing include: a fetus with ultrasound-identified anomalies that might suggest a genetic etiology, but the standard genetic testing has not identified the cause; cases in which expert genetic consensus suggests a high likelihood of a genetic etiology; couples with a previous affected fetus, stillbirth or child with no genetic diagnosis and the identification of a recurrent pattern of anomalies in a new pregnancy.
- 5.) A list of laboratory recommendations for consistent test quality, DNA variant interpretation, and return of results to parents are listed in the new position statement.

“In an effort initiated by the Position Statement Committee of ISPD, representatives from the three organizations, ISPD, PQF and SMFM, recognized the importance of a shared, consistent message to healthcare providers about the application of new technologies to prenatal and perinatal care,” said Ignatia Van den Veyver, MD, a maternal-fetal medicine specialist, ISPD President and co-author of the position statement. “This resulted in a productive, collaborative effort to develop this joint position statement. We envision this as the beginning of similar future collaborations that will benefit prenatal care providers and patients in the current rapidly evolving genomic era.”

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About ISPD

The International Society for Prenatal Diagnosis (ISPD) was founded in 1996 to address the need to advance the medical practice and science of prenatal diagnosis and therapy by bringing together a global multidisciplinary group of medical and scientific professionals with interests and expertise in a diverse array of clinical and research aspects of prenatal diagnosis and fetal care. While ISPD focuses on all areas relevant to this field, the Society has a unique focus on and expertise in reproductive and prenatal genetic screening and how this aspect of care integrates with other disciplines of prenatal diagnosis. ISPD’s vision is that evidence-based practice and culturally sensitive preconception and prenatal screening, diagnostics and therapy shall be available to all families. ISPD hosts an annual conference dedicated to discussing the latest in the field. For more information, please visit www.ispdhome.org.

About PQF

The Perinatal Quality Foundation is an independent non-profit foundation incorporated in 2004. The mission of the Perinatal Quality Foundation is to improve the quality of obstetrical medical services by providing state of the art educational programs, and evidence-based, statistically valid monitoring systems to evaluate current practices and facilitate the transition

of emerging technologies into clinical care. The strength of the PQF is its ability to bring together experts and leaders devoted to maternal and fetal health to reflect on, select, and implement programs to facilitate quality perinatal patient care.

About SMFM

The Society for Maternal-Fetal Medicine (est. 1977) is a non-profit membership organization representing the interests of obstetricians/gynecologists who have additional formal education in maternal-fetal medicine. The Society is devoted to reducing high-risk pregnancy complications by providing continuing education to its more than 2,000 members on the latest pregnancy assessment and treatment methods. It also serves as an advocate for improving public policy and expanding research funding and opportunities for maternal-fetal medicine. SMFM hosts an annual scientific meeting in which new ideas and research in the area of maternal-fetal medicine are unveiled and discussed. For more information, visit www.smfm.org.