Ob/Gyn Utilization of the NIPT Expanded Testing Option

Sarah Mayes, BA (Advisor: Jennifer Czerwinski, MS, CGC)

Noninvasive prenatal testing (NIPT) enables the detection of common fetal aneuploidies such as Down syndrome, trisomy 18, trisomy 13, and sex chromosome abnormalities via analysis of cell-free fetal DNA circulating in maternal serum. Although the accuracy of NIPT for fetal aneuploidy is expected to be higher than that of currently available alternative maternal serum screening options, the implications of results are not as straightforward. In October 2013, the option to screen for additional trisomies and particular microdeletion syndromes, such as 22q11.2 deletion syndrome and 5p- syndrome, became clinically available. Due to this rapidly evolving prenatal screening technology, clinicians must make a conscious effort to keep abreast of the current options; however, the complexity of the testing methods, oftentimes unclear clinical utility of results and current lack of professional guidelines for its use renders this task challenging. To assess physicians’ awareness of, utilization of, and attitudes toward the expanded testing option of NIPT, 85 Houston, TX area Obstetrician/Gynecologists were surveyed. While all respondents indicated they were aware of NIPT in its traditional form, 75% were aware of the expanded testing option. Of these respondents, 17% report ever electing the expanded testing option when ordering NIPT. Thirty-nine percent of those surveyed indicated they would feel at least somewhat uncomfortable explaining the expanded testing option to a patient and, accordingly, 91% expressed that practitioners need more information regarding the screen. The responding Ob/Gyns indicated that this new screening option will be increasingly applicable to their future practice, with 28% indicating that they plan to incorporate the NIPT expanded testing option into their practice in the future. Based on these findings and the quickly evolving landscape of prenatal screening, education and reeducation of healthcare professionals is imperative to ensure responsible patient counseling, informed consent, and appropriate management following test results.

Advisory Committee:

- Jennifer Czerwinski, MS, CGC, Chair
- Syed S. Hashmi, MD, MPH, PhD
- Mark A. Turrentine, MD, FACOG
- Sandra Darilek, MS, CGC
- Lara Friel, MD, PhD