Patient Perception of Residual Risk Post Negative Non-Invasive Prenatal Testing

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Recent technological advances have yielded a new method of prenatal screening, non-invasive prenatal testing (NIPT), which uses cell-free fetal DNA from the mother’s blood to assess for aneuploidy. NIPT has much higher detection rates and positive predictive values than previous methods however, NIPT is not diagnostic. Past studies have demonstrated that patients may underestimate the limitations of prenatal screening; however, patient perception of NIPT has not yet been assessed. Therefore, we conducted a prospective cohort study to assess patient understanding of the residual risk for aneuploidy after receiving a negative NIPT result. Ninety-four participants who had negative NIPT were surveyed. The majority of participants (61%) understood the residual risk post NIPT. Individuals with at least four years of college education were more likely to understand that NIPT does not eliminate the chance of trisomy 13/18 \((p=0.012)\) and sex chromosome abnormality \((p=0.039)\), and were more likely to understand which conditions NIPT tests for \((p=0.021)\), compared to those women with less formal education. There was a significant decline in general level of worry after a negative NIPT result for aneuploidy that NIPT tests for. There was also a significant decline in worry to have a pregnancy affected by any other genetic condition. The data demonstrate that despite the relatively recent implementation of NIPT into obstetric practice, the majority of women are aware of its limitations after receiving genetic counseling. However, clinicians may need to consider alternative ways to communicate the limitations of NIPT to those women with less formal education to ensure understanding.

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