Aneuploidy Screening and Invasive Testing in Western North Carolina in the Era of Cell-free Fetal DNA Testing

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Noninvasive prenatal testing (NIPT) offers accurate screening for fetal aneuploidy, primarily trisomies 21, 18, and 13. Invasive diagnostic testing (IT) carries a risk of fetal loss but remains the standard for definitive diagnosis. We describe our experience with NIPT and subsequent IT in a regional referral center in western North Carolina. Decisions about NIPT, IT and the results were assessed prospectively for patients presenting for prenatal genetic counseling from November 2012 through November 2013. We compared NIPT and IT rates between women presenting in their first trimester [W1 ≤ 12wks: n = 74 (18.9)] versus later trimesters (W2-3 > 12wks: n = 318 (81.1%)] using Chi square or Fisher's exact test. Data are presented: n(%). Women presenting in the first trimester chose NIPT significantly more often than women later in pregnancy [W1 = 59 (79.7) vs. W2-3 = 140 (44); $p < 0.001$], and they chose IT significantly more often [9(12.2) vs. 11(3.5); $p = 0.005$). Indications for genetic counseling and reasons for declining NIPT were also significantly different. Our experience was different for women presenting for genetic counseling early versus later in pregnancy. There remains a subgroup of women who desire definitive and comprehensive information available only by invasive testing. These results provide more relevant and specific information when counseling patients.

Key Words: Prenatal genetic counseling; Non-invasive prenatal testing; Invasive prenatal testing