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Clinical utility of expanded genome wide NIPT/ceff free DNA in the general population

Thomas Westover MD Div MFM, Capital Health Trenton NJ

Introduction

Our USA MFM dept offers genome wide NIPT and single gene NIPT (and sono imaging each trimester) to approx 4k annual births. After IRB approval, we retrospectively reviewed 5 years of results to assess clinical utility and accuracy. Fetuses with structural anomalies or abnl NIPT underwent pre or postnatal testing with karyotype/snp array/NGS as indicated.

20,552 patients delivered live/stillbirths or TOP $\geq$ 12 wks gestation. 2.2% of fetuses were dxed with congenital anomalies either pre/postnatally via US, NIPT, physical exam, or state mandated newborn screening.

92% of the anomalous preg were dxed prenatally (8% postnatally). 88% of abnl fetuses had abnl US while 12% had normal US.

315 fetuses had abnl genome/single gene NIPT: 110 fetuses had confirmed CATS (nonmosaic 13/18/21), 6 fetuses had confirmed RATS (trisomy 8/9/14/15/22), 56 fetuses had confirmed SCA, 19 fetuses had confirmed abnl del/dup syndromes, 20 fetuses had confirmed single gene NIPT.

75% of structurally anomalous fetuses by US had normal NIPT. Conversely, 41% molecularly diagnosed fetuses (CATS, RATS, SCA, del/dup syndrome, single gene mutation) had normal US. Cohort size precluded exact estimation of positive predictive value; statistical estimation yielded wide confidence intervals

US yields the greatest clinical utility for the dx of clinically relevant fetal/newborn structural and genetic anomalies.

New genetic screening technologies offer dx opportunities that complement US and can yield diagnoses that are not detectable via US.

Ongoing research with large populations is needed to assess the relative contribution of each screening modality and determine sensitivity/specificity but it is intuitive that each screening platform offers unique windows into the presence or absence of fetal genetic disorders

