

Review of: "Non-Invasive Prenatal Testing (NIPT) for Aneuploidy in a Setting with a High Consanguineous Rate – A Retrospective Cohort Review of 1,153 Cases"

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Potential competing interests: No potential competing interests to declare.

A well written comprehensive paper, the first addressing NIPT in a setting with high consanguinity. It provides some reassuring evidence given the concerns previously expressed regarding NIPT testing as stated by Bajka et al. They identified fetal or maternal mosaicism or higher levels of homozygosity on the chromosomes tested (when the SNPs between mother and baby are too similar to yield informative results, possibly from consanguinity as being a potential reason for failure to obtain results post NIPT testing.

The authors choose high risk cut off as > 1 in 150? This is at variance with other reports where cut offs of 1 in 50 or 1 in 100 are used. Is there any specific reason for this?

The authors also acknowledge the major limitation of their studies- a small population with predominantly from an older wealthier population and emphasise the need for more data to confirm these initial findings

The paper is a useful addition to the current literature on NIPT and well worth a read.