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.

Thank you for asking me to review this paper written by a team in Qatar concerning the accuracy of NIPT in a specific population .

The study is a retrospective one and it was well designed .The data collected was complete. We have always this idea about the limitation and the accuracy of NIPT in a consanguinous couples and this work highlights this problem and give us a answer about this .

I have two comments :

The first one is about i beleive an error in writting : the table 5 where they write that “ shows the details of the high-risk tests that were confirmed by karyotyping. There were 10 cases of trisomy 21, 6 cases of trisomy 13, and 4 cases of trisomy 18……” This correspond i beleive to table 6 and not to table 5

The second one is about the content : if it's possible just to highlight that in consanguinous couples the risk of genetiec disease in the fetus is increased because of the recessive transmission and not the risk of aneuploidy . So NIPT is a good test to rule out aneuploidy like trsiomy 13,18 and 21 but not a good test to rule out recessive diseases specific to consanguinity . And in these couples good counseling is to offer them ultrasound and genetic consultation .

This work highlights the utility and performance of NIPT in consanguinous and it is well designed .

Thank you fot your trust

Regards