

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"

Thomas Liehr¹

¹ Friedrich-Schiller Universität Jena

Potential competing interests: No potential competing interests to declare.

This paper has following problems:

- Statement in Introduction on false positive (FP) rates of tests used before NIPT amniocentesis (AC) and CVS are at least misleading. And it is not true that those with FP results were those which were predominantly advised to take an invasive test. Also main point would be to tell about risk of invasive diagnostics here, which is below 0.5% nowadays.
- Whole paper does not mention that cfDNA is placenta derived.
- It is not mentioned that NIPT is only highly reliable for trisomy 21 detection and not for other CNVs. the 99% given in Introduction are only valid for T21.
- Introduction: it must read as 45,X and not 45,X0
- Introduction: - what is the idea behind that a CNV in SNP-based NGS-test (NIPT) could not be reliable? CNVs can be detected in each case, if homo- or heterozygous state.
- Introduction: authors state to have been made tests for T13, T17 and T21 - most likely the mean T18 instead of T17...
- Table 4 - data on age is incomplete for Non-Consang.
- I suggest you to read and cite these articles: PMID:35986330, PMID: 34220958, PMID: 36428876