

Performance of the non-invasive prenatal test in vanishing twin pregnancies: a systematic review

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Objective

Worldwide non-invasive prenatal test (NIPT) is being offered as prenatal screening for chromosomal aberrations by analyzing cell free DNA in the maternal blood. However, the screening performance of the NIPT in vanishing twin (VT) pregnancies is relatively unknown. In this systematic review we studied all available literature regarding NIPT in case of VT.

Methods

A literature search of Medline, Embase, Cochrane Library and Google Scholar was performed on the 3th of May 2021. Studies were eligible if they described the test performance of the NIPT for trisomy 21, 18, 13, sex chromosomes and additional findings. Case-reports, posters and conference abstracts were excluded. The methodological quality of the studies was assessed with the quality assessment tool for diagnostic accuracy studies-2 (QUADAS-2).

Results

Five studies were eligible for inclusion, four were retrospective observational cohort studies and one was a prospective observational cohort study. The cohort sizes ranged from 15 to 767 VT pregnancies. Two studies reported on the performance for T21. In one study the NIPT tested positive for T21 in 19/767(2.5%) VT and T21 was confirmed in 3/19(15.8%) cases. In the other study the NIPT tested positive for T21 in 9/579(1.6%) VT and T21 was confirmed in 1/9(11.1%) cases. Three studies reported the screening performance for T18. NIPT tested positive for T18 in 4/767(0.5%), 2/579(0.3%) and in 1/15(6.6%) VT, of which 0, 0 and 1 were confirmed respectively. Two studies reported on the screening performance for T13. In one study the NIPT tested positive for T13 in 5/767(0.7%) and in the other study in 1/579(0.2%) of which none were confirmed. In addition, one study described additional findings through whole-genome NIPT. The NIPT tested positive in 23/767(2.9%) cases of which none were confirmed in the remaining fetus. Two studies solely described the number of false and true positives for the common trisomies combined. In the first study the NIPT tested positive for one of the common trisomies in 12/206(5.8%) of which 7/12(58%) were confirmed. In the second study, 11/87(12.6%) VT tested positive for one of the common trisomies and 4/11(36.4%) were confirmed. None of the studies reported false negative cases.

Conclusion

NIPT can be offered in case of a VT pregnancy due to no false negative cases. However, patients should be counseled regarding the higher risk of a false positive result. Further studies are necessary to determine the optimal timing of the NIPT in VT pregnancies.