

NIPT in the diagnosis of failed pregnancies

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et al.*



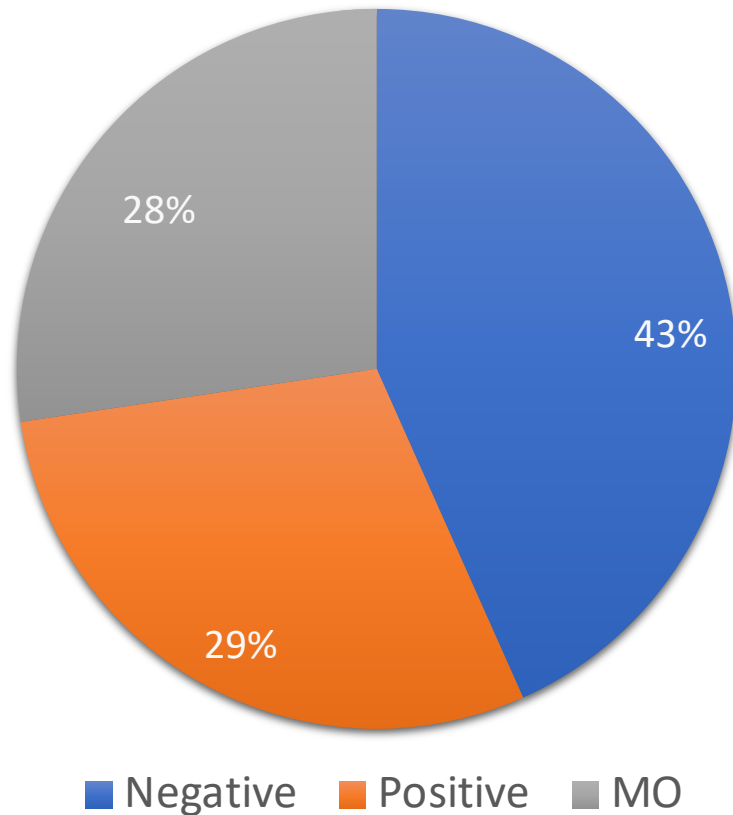
GENNET

Results of products of conception 2014-2024

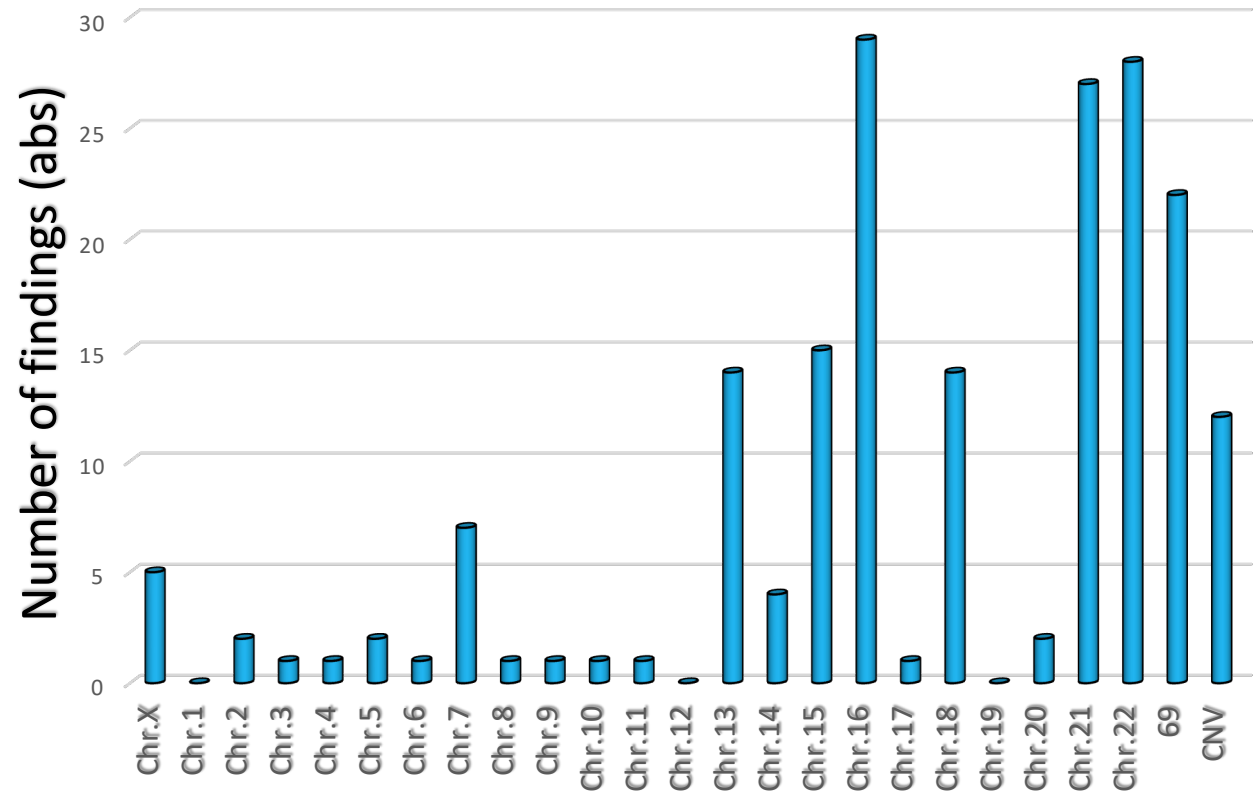
- SAB in the first trimester- chromosomal aberration in 45% - 70%

SAB QF-PCR

n= 2428



Chromosomal aberration



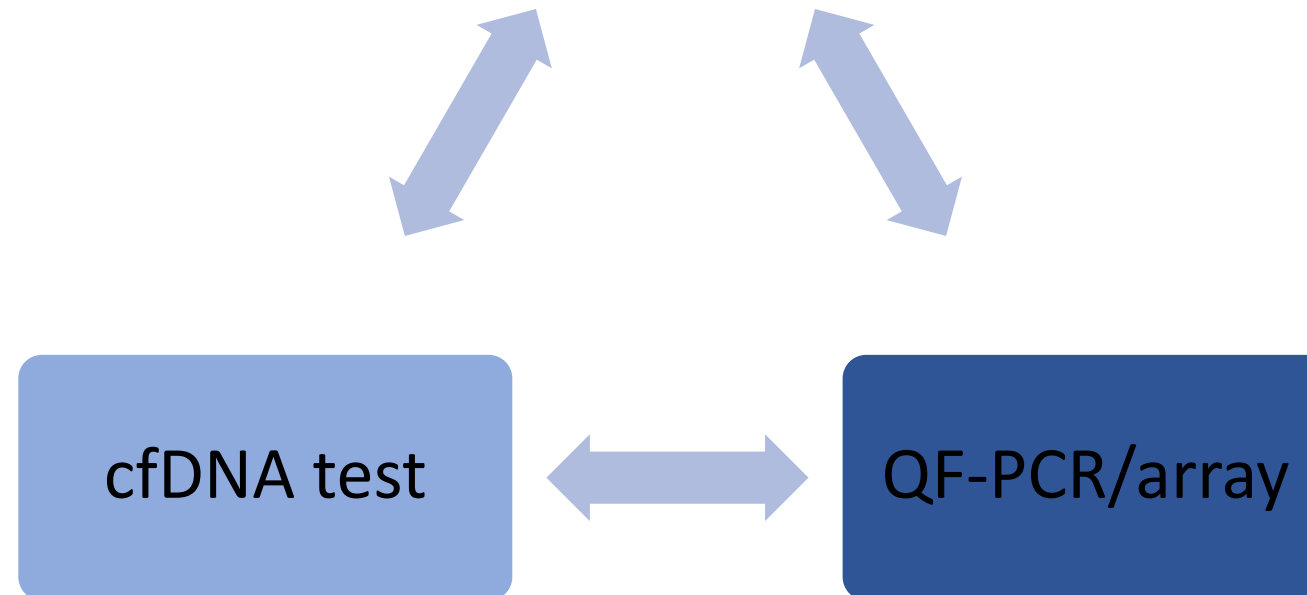
Could cfDNA testing be used to detect aneuploidies in early SABs



Prospective study GENNET 2020-2023

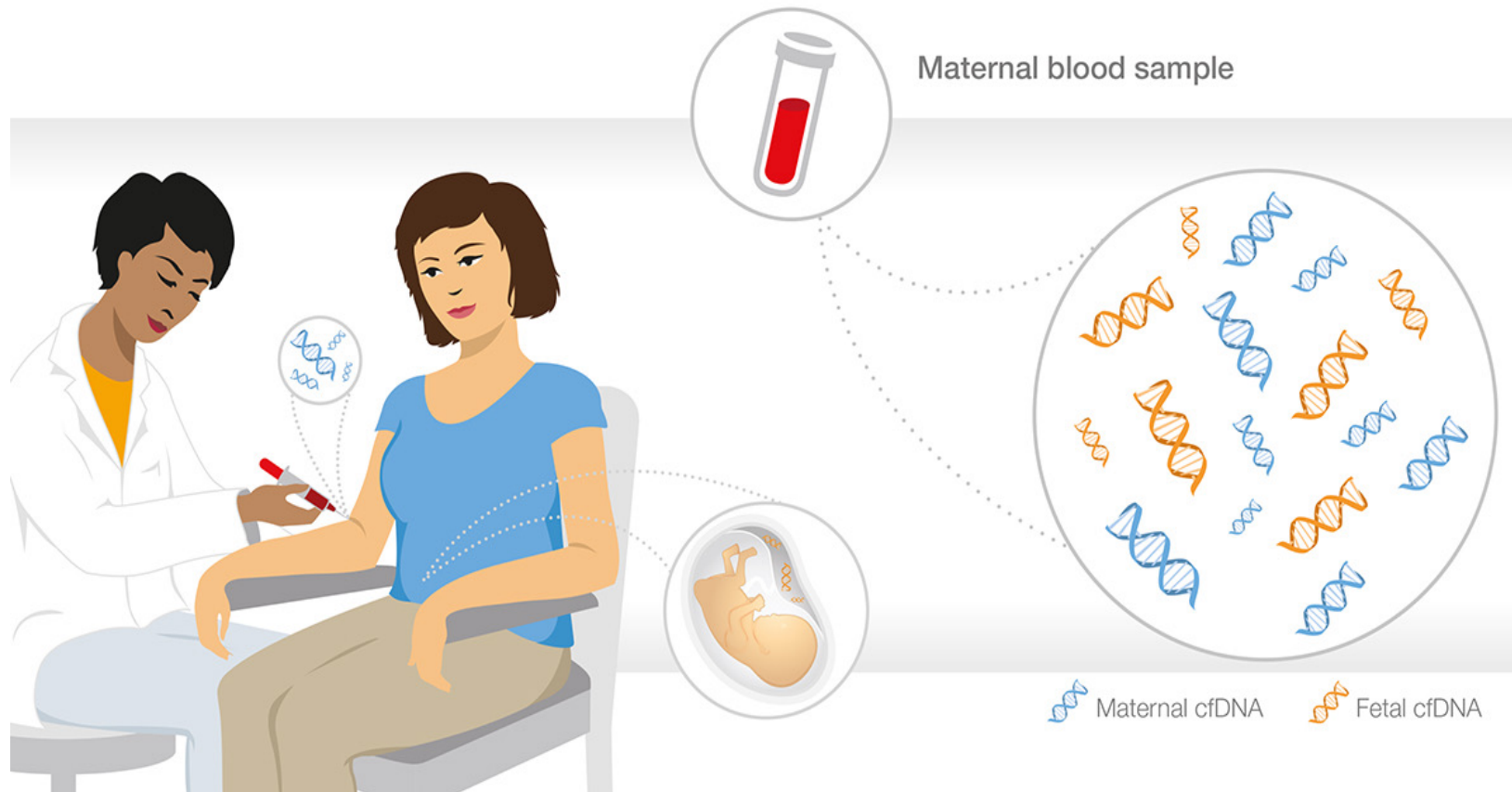
86 pregnancies

RCUI for missed abortion in 5.-12. week of pregnancy
(average 8 weeks)

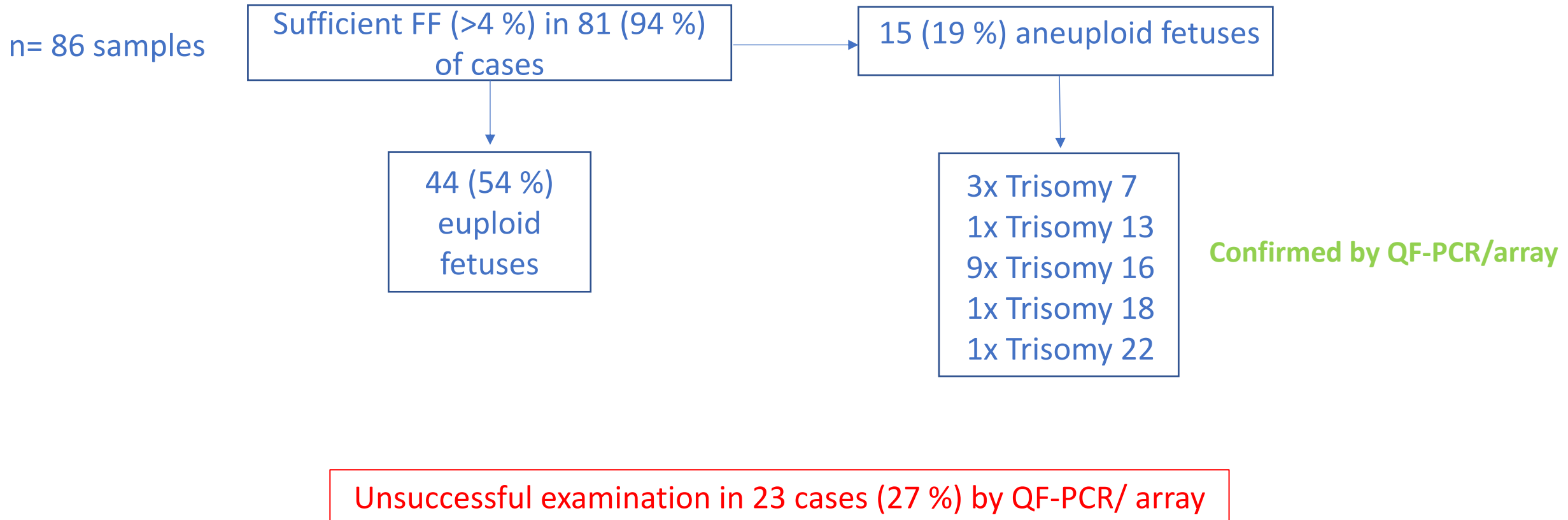


NIPT (Non-Invasive Prenatal Test)

- maternal blood sample was taken → cfDNA was isolated from the blood plasma
- Low-pass whole genome sequencing (NextSeq 550, Illumina)

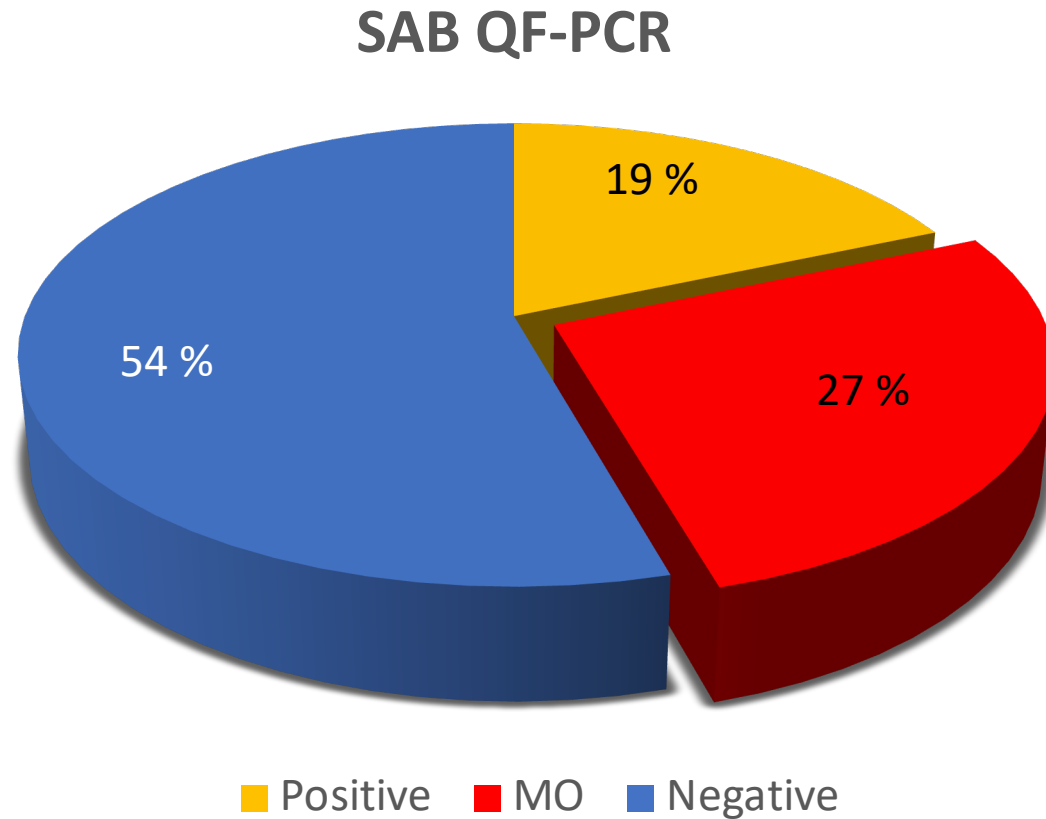


Results of the study



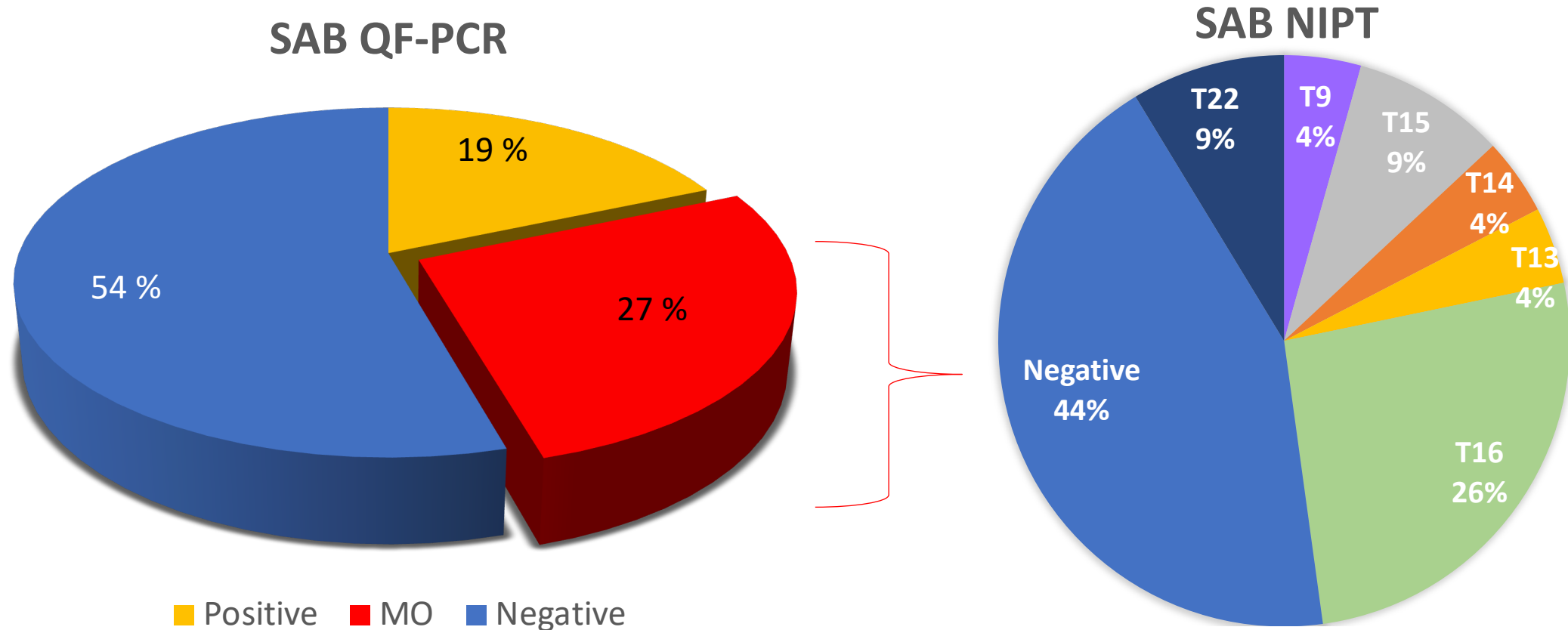
Results of the study

- Maternal contamination of tissue samples was found in 27% of cases, that could not be further evaluated with QF-PCR/ array



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In these cases, cfDNA testing was able to give the results.

Conclusion

- Non-invasive Testing of Products of Conception can be used for detection of aneuploidies in nonviable pregnancies early in the first trimester- sufficient FF for the result in 3/4 of cases
- Suitable there, where conventional methods (QF-PCR/array) cannot be used - material from the abortion cannot be collected or maternal contamination is proven.
- Importance of Chromosomal Aberration Detection in Reproductive Losses:
 - Elucidation of the cause of reproductive failure
 - Examination of parents and determination of risk for further pregnancy
 - Plan of preconception and prenatal care in the next pregnancy
 - Couples in the IVF program-> indication for PGT-A (PGT-SR)

Thank you

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