

Targeted testing in prenatal diagnosis: who decides about the target?

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Prenatal diagnosis may detect other chromosome abnormalities than the one specifically searched for. Predominantly, the increased risk for trisomy 21, leading to Down's syndrome, is the reason for pregnant women to have amniocentesis or chorionic villi sampling done. However, due to the applied technique of full karyotyping where all 46 chromosomes of the fetus are examined, other chromosome anomalies can also be found. Testing results of mild or unclear clinical significance, like 45,X (Turner's syndrome) or 47,XXY (Klinefelter's syndrome), are known to lead to difficult counseling issues and dilemmas for clients needing to decide about termination of pregnancy. The use of new molecular techniques like Quantitative Fluorescent Polymerase Chain Reaction (QF-PCR) in a scenario of 'targeted testing' would enable prenatal diagnosis to focus on high risk abnormality(s) only, and exclude these additional results.

We analysed the possible impact of targeted testing by interviewing providers and clients about their experiences with additional testing results. We found that implementation of targeted testing would require an explicit answer to the morally laden question of which testing results should be excluded from prenatal diagnosis. Moreover, it is not yet clear who should decide about this. The providers of prenatal diagnosis or their clients? Other stakeholders, like general practitioners or policy makers, might also be involved, because the question which results to include is linked to the more fundamental question of what actually *is* the target, i.e. the goal to be achieved, in prenatal diagnosis.