The target of testing: reflections on the morality of prenatal diagnosis

Myra van Zwieten, PhD. Academic Medical Center/University of Amsterdam, the Netherlands.

New developments in molecular diagnostic techniques will soon radically change the scene in prenatal diagnosis.
Until now, *any* kind of chromosome abnormality can be detected in prenatal diagnosis, not only the one(s) for which the test is actually performed because of an existing high risk (i.e. trisomy 21, leading to Down's syndrome, in the majority of cases). Currently, standard procedure in chromosome analysis is to examine *all* 46 chromosomes, regardless the chromosome(s) that is suspected to be anomalous. This conventional method of full karyotype analysis occasionally leads to other testing results than the client might have expected.
New diagnostic tests might alter this situation as these new molecular tests will only detect a selected amount of specific chromosome abnormalities. Tests like quantitative fluorescence polymerase chain reactions (QF-PCR) or Multiple Ligation-dependent Probe Amplification (MLPA) have for instance proven to be able to detect trisomy 21, 18 and 13 only, without detecting other chromosome abnormalities.

Recently, discussion has started about whether the new molecular techniques should replace the conventional method of full karyotype analysis. Several kinds of arguments are referred to in this discussion. Supporters of this new scenario claim for example that this kind of 'targeted testing' would lead to less 'unexpected' findings, hence less difficult counseling issues. Opponents mention for instance that, because of the involved miscarriage risk of amniocentesis and chorionic villi sampling, clients should always be supplied with maximum information, regardless the original reason of having the test done. Both parties tend to take the patients' preferences into account as well, for instance by the idea of offering individual clients a 'menu' of testing possibilities.

However, as the future of prenatal diagnosis is at stake here, the discussion about targeted testing first and foremost needs to address the more fundamental question of 'What actually *is* the target of testing in prenatal diagnosis?'. Is prenatal diagnosis a medical diagnosis following a specific indication? Or is the objective of prenatal diagnosis to detect as many abnormalities as possible?
Based on empirical research (focus groups and in-depth interviews with prenatal diagnosis providers and clients), we will present a definition of prenatal diagnosis holding the different aspects that needs to be addressed in a reflective discussion on the morality of prenatal diagnosis.
Evidently, this ethical discussion involves the question of which testing results should and should not be processed in prenatal diagnosis. Since previous experiences have shown how deep the disagreement can be about 'drawing lines' in this setting, it remains to be questioned to what degree prenatal diagnosis providers would be willing to participate in this discussion.