Professional responsibility towards the application of new methods for prenatal testing

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New technical developments in molecular testing could result in dramatical changes in prenatal diagnosis. Techniques like Fluorescence In Situ Hybridisation (FISH) and Quantitative Fluorescent Polymerase Chain Reaction (QF-PCR) could replace the conventional full karyotyping method in which all chromosome abnormalities are detected, by testing only a selective set of chromosome abnormalities. Prenatal testing done for advanced maternal age could for instance include only the autosomal trisomies in the future, with the result that testing results other than trisomy 21, 18 or 13 would no longer be detected.
The application of targeted testing in prenatal diagnosis would be unique because this would mean restricting the range of an already existing test. An important reason for this is to get rid of the problems of the unexpected findings. Most problematic are the testing results for which the phenotypical consequences are unknown, e.g. mosaicism or apparently balanced structural chromosome rearrangements. In these cases only risk percentages can be presented to the parents. Application of the new molecular tests would actually mean that *unexpected* findings would become *unacceptable* problematic findings which should be excluded from the future target of prenatal diagnosis.
An important question is who will set this future target. Using the argument of patients' autonomy it is sometimes suggested that patients should be able to decide for themselves which testing results will be available for them. This argument could imply that professionals will not have any responsibility anymore with regard to the offer of prenatal testing, but in our view this is not the case. Based on the results of our empirical ethical research in the field of prenatal diagnosis we will elaborate on the professionals' responsibility in this matter.

In this presentation we will explain the dynamic process of establishing testing results by illustrating how testing data gradually evolve into meaningful testing data along a testing pathway. Unexpected findings are more difficult to establish because somewhere along this testing pathways the meaning of these results appears to be ambiguous. Our investigation have shown that professionals entrusted with the 'disambiguation process' of these results often try to assess the relevance of the testing data in view of the decision which the parents have to make about their pregnancy. This assessment will affect whether an ambiguous result will evolve into a negative or positive testing result, or remain ambiguous and communicated as such to the parents. Our observations have shed light on this assessment process including the arguments which play a role in this process, like the assessed meaning of the specific pregnancy and of the expected phenotypical defects.

Up to now professionals have to deal with unexpected findings in prenatal diagnosis on an individual and ad hoc base. The discussion about the application of targeted testing could be an opportunity to deal with the unexpected findings more systematically. For this reason the discussed question should be: "which testing results do we – as professionals – assess as relevant enough to communicate to parents as input for their decision about the continuation of their pregnancy?" It could be a great challenge for professionals seriously wishing to act upon their responsibility to systematically reflect on this question.