Non-invasive prenatal diagnosis can reliably detect trisomy 21

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Until a few years ago, invasive testing was the only way to diagnose trisomies, such as the Down syndrome, before birth. These invasive tests, for example amniocentesis, have a risk of causing miscarriage. Therefore, they are usually only used in so-called high-risk pregnancies, for example, when the woman is relatively old or when trisomies already occurred in earlier pregnancies.

In non-invasive prenatal diagnosis (NIPD), in contrast, the pregnant woman's blood is tested for the foetal trisomies 13, 18 and 21. The Federal Joint Committee (G-BA) commissioned the German Institute for Quality and Efficiency in Health Care (IQWiG) to analyse studies on the diagnostic characteristics of these tests and to outline hypothetical scenarios of a possible integration into prenatal care.

Many unknown variables

No robust estimations of the sensitivity and specificity of the tests on the rare trisomies 13 and 18 are possible. With over 99 per cent for trisomy 21, sensitivity and specificity of the tests are similar to those of invasive methods.

If pregnant women at an increased risk of foetal trisomy 21 were offered an NIPD, some of the miscarriages caused by invasive testing could probably be prevented. More detailed conclusions on the German health care situation are not possible: More precise modelling would require

knowing the proportions of women of different ages who have invasive or non-invasive testing or who do not make use of such testing—for example, because they want to have the child in any case. These numbers are not known for Germany, however.

More information: www.iqwig.de/en/projects-resul … regnancies.7776.html

Provided by Institute for Quality and Efficiency in Health Care

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