Biochemical screening for fetal chromosomal abnormalities and neural tube defects - a technology assessment

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Authors' objectives
This report aims to assess, based on the current available evidence:

1. If it is possible to determine the effectiveness of biochemical screening during pregnancy

2. If biochemical screening during pregnancy can help reduce the high number of amniocentesis carried out in Germany

3. If biochemical screening during pregnancy be implemented in the Federal Republic of Germany

Authors' conclusions
- The biochemical screening during pregnancy is able to achieve a contribution to prenatal detection of chromosomal disorders and neural defects. The effectiveness of the combinations of markers appraised in this review is greater than that of screening for maternal age alone.

- The biochemical screening during pregnancy can reduce - subject to sensitivity - the frequency of invasive diagnostic procedures (above all amniocenteses) through exclusion of fetal disorders, e.g. by determining the individual risk before carrying out a planned amniocentesis and disclaiming further diagnostic procedures in case of unobtrusive (negative) screening results.

- The precise determination of gestational age is of great importance for the validity of the screening result. Indicated is a sonographic determination before executing the biochemical screening.

- The detection rate of fetal chromosomal abnormalities by biochemical screening is higher in older than in younger pregnant women. In older pregnant women anxiety caused by positive screening results is less, invasive diagnostic procedures to follow up diagnosis are taken up to a minor extent and there is more seldom termination of pregnancy affected by chromosomal abnormality or neural tube defect. An age limit of 35 years for differentiation between younger and older pregnant women is not obliged.

- The procedures for ensuring the diagnosis in positive screening results are combined with maternal and fetal risks. In many cases - especially in younger pregnant women - the risk of a procedure related loss of a not affected fetus by ensuring diagnosis is many times greater than the individual risk of carrying a fetus affected by chromosomal abnormality or neural tube defect.

- Biochemical screening is feasible in second or first trimester of pregnancy in different combinations of biochemical or biochemical and sonographic markers.

- Screening in second trimester [AFP, hCG and uE3 (and possibly supplied by Inhibin A)] is often examined as standard screening and seems to carried out with a an acceptable sensitivity in the ambulatory health care of the Federal Republic of Germany.
Screening in first trimester [fetal nuchal translucency, PAPP-A and free b-hCG (and possibly supplied by uE3)] features a higher detection rate and permits an earlier diagnosis than second trimester screening.

An evidence based recommendation for introducing a national biochemical screening program in Germany is actually not indicated because the second trimester screening doesn't represent an optimal solution for ethical reasons - late terminations - and in first trimester screening the measurement of fetal nuchal translucency is not feasible in outpatient health care with sufficient sensitivity.

A national screening program should enclose a first trimester screening with sonographical examination in specialised centres or equivalent qualified ambulatory units.

With regard to an economic evaluation - e.g. which screening strategy is more efficient than others in consideration of all relevant aspects there are still existing gaps in knowledge which should be uncovered by further analyses.

In the presence of new gentechnical methods the importance of biochemical screening during pregnancy in future will be diminished. Therefor the implementation of a national screening program for the Federal Republic of Germany upon the base of biochemical screening can only be a temporary solution.

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