INAHTA Briefs

Issue 2007/77

Title **Methods of Early Prenatal Diagnosis**

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Reference SBU Report 182, 2006. ISBN 978-91-85413-13-3.
Full text report in Swedish and summary and conclusions in English are available at www.sbu.se/published

Aim

To systematically review the literature with the intent of examining the scientific evidence for methods currently used, or about to be adopted, in prenatal diagnosis to detect fetal chromosomal and structural abnormalities.

Conclusions and results

Among others:

- A combined test of nuchal translucency measurement, ultrasonography, and maternal serum biochemistry (biochemical screening) in early pregnancy (10–14 gestational weeks), along with maternal age, is the clinically evaluated method to assess the probability of fetal Down syndrome that strikes the best balance between the percentage of detected cases and false-positive results (Evidence Grade 1).
- All methods (nuchal translucency measurement, maternal serum biochemistry in the second trimester, and the combined test) for assessing the probability of fetal Down syndrome examined by this report and evaluated in clinical practice strike a better balance between the percentage of detected cases and false-positive results than maternal age alone. Thus, the use of these methods requires fewer amniocenteses and chorionic villus samplings per detected cases of Down syndrome than maternal age alone (Evidence Grade 1).
- Most pregnant women want to be notified early and prefer screening in the first trimester (Evidence Grade 1).

Methods

The project analyzed medical, social, psychological, ethical, health economic, quality assurance, and safety aspects of early prenatal diagnosis. Literature searches were performed in electronic databases such as Cochrane Library and PubMed/MEDLINE. A search for health economic studies was also conducted in the National Health Service Economic Evaluation Database (NHS EED). Bibliographies were examined and members of the project team followed various areas of current research. Based on predetermined criteria, the identified literature was systematically selected and quality assessed.

Further research/reviews required

Among others:

- The clinical value of nasal bone ultrasonography, ultrasonographic soft markers, Doppler ultrasonography, 3D ultrasonography, or MRI as screening or prenatal diagnostic methods in the first or second trimester remains unclear.
- There is insufficient knowledge concerning which models are most suitable for informing pregnant women and their partners, or how information should be formulated to satisfy the needs of particular ethnic and cultural groups.

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ISSN 1654-501X