



Title Maternal Ultrasound and Serum Screening in the Detection

of Structural and Chromosomal Abnormalities

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**Reference** Report 27/2005. ISBN 951-33-1796-X. ISSN 1239-6273.

Full report available at: www.stakes.fi/finohta

## Aim

To systematically collect the best evidence and knowledge on the sensitivity, specificity, and possible side effects of various screening methods; to describe the ethical and psychological effects of screening for fetal abnormalities; and to analyze cost effectiveness.

## Conclusions and results

Major congenital anomalies are detected in 2 to 3 out of every 100 births, of which about one third are affected by several serious anomalies or a syndrome. The effects of an anomaly on the course of pregnancy or an infant's prognosis vary widely. Screening for fetal abnormalities has major ethical and psychological consequences. Present screening practices in Finland vary from one municipality to another and do not ensure equality. We hope that the research evidence and expert knowledge presented in this report will provide a basis for fruitful health policy discussions on the targets and implementation of fetal screening. A uniform national screening system and improvements in the quality of screening are set as targets. The report provides necessary data and information.

#### Recommendations

A system to evaluate the quality and effect of screening for fetal abnormalities must be established. A national expert group is needed to control the quality of screening (especially education of healthcare professionals in counseling and ultrasound methods). Parents should be allowed to make a conscious, voluntary decision concerning their participation. Counseling on screening should be competent, consistent, and adequately available at each screening stage.

# Methods

A systematic review was performed on the various methods for screening structural and chromosomal abnormalities. The report examines the different screening models in the context of Finnish practice, and compares their implementation and results with a situation where

screening is not performed. Modeling is used to analyze the shares of abnormalities identified through screening and the costs of screening. Furthermore, the ethical and social dimensions of screening were analyzed with an expert group. The report also addresses important factors affecting the quality management of screening organizations.

# Further research/reviews required

Methods to screen for structural and chromosomal abnormalities are under continuous development. To be successful, maternal screening should set clear targets for detecting fetal abnormalities. The targets may change as new treatment options emerge.