

Title	First and second trimester prenatal screening for trisomies 13, 18, and 21 and open neural tube defects
Agency	Provincial Health Technology Assessment Program Institute of Health Economics 1200, 10405 Jasper Avenue, Edmonton, Alberta T5J 3N4, Canada Tel: +1 780 448 4881, Fax: +1 780 448 0018; http://www.ihe.ca/
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Aim

To review evidence on the efficacy, safety, cost-effectiveness, and cost impact of various first and second trimester screening methods for pregnant women who wish to obtain a risk assessment for open neural tube defects (NTD) or fetal trisomy 13, 18, and 21. A social and system demographic analysis and an analysis of ethical issues were also conducted.

Conclusions and results

Safety and efficacy

The majority of the evidence available from one systematic review and 72 screening accuracy studies concerned first trimester screens (nuchal translucency (NT), double serum, and combined) and second trimester serum screens (dual, triple, and quadruple serum). The results are summarized below.

- Limited evidence suggested that two-step screens provided the highest detection rate and lowest false positive rate.
- The first trimester combined screen (used in southern Alberta) and the full integrated and serum integrated (integrated screen without NT) screens met the minimum performance threshold for acceptable trisomy 21 first trimester screens set by the Society of Obstetricians and Gynecologists of Canada (SOGC).
- The quadruple serum screen (used in northern Alberta) met the minimum performance threshold set by the SOGC for second trimester trisomy 21 screens.
- There was strong evidence that second trimester ultrasound provided the most accurate information regarding open NTD.
- The impact of screening results on physician and maternal decision making and maternal or fetal outcomes is unknown.

Economic outcomes

Twelve cost-effectiveness studies of acceptable quality assessed 25 first and second trimester screening strategies, but it was unclear which screening algorithm was the most cost effective.

The results of the economic analysis are as follows.

- Dual and serum integrated prenatal screening (IPS) were the most cost-effective strategies, with serum IPS being more costly and more effective than dual.

- First trimester combined screening was associated with greater costs and greater effectiveness, compared with second trimester quadruple screening (when only considering the algorithms currently available in Alberta).
- Physician services accounted for approximately 70% of the costs associated with screening.

Recommendations

Empirical evidence strongly supported using the combined test for first trimester risk assessment for trisomy 21 and other aneuploidies; the quadruple serum test was an acceptable second trimester screen. Second trimester ultrasound was superior to second trimester serum screening for detecting open NTD.

Serum IPS provided the most value for money among the 13 strategies evaluated, but repeated measures without NT has the potential of being the most cost effective. Within the algorithms available in Alberta, combined screening is associated with additional benefit and additional costs, compared with quadruple serum screening.

Methods

Please refer to the full report for details of the methods.

Further research/reviews required

Few empirical studies assessed the performance of two-step screens and no empirical studies assessed the performance of contingent or repeated measures screens for fetal aneuploidy. In general, the accuracy studies suffered from several major methodological limitations. Three of the biases (differential verification and not adequately describing the index test or the study population) potentially affecting the studies are associated with a 1.5 to 2-fold overestimation of diagnostic test performance.

Written by

Institute of Health Economics, Canada