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| <b>Title</b>     | First and second trimester prenatal screening update  |
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| <b>Reference</b> | August 2014 (English); ISBN 978-1-926929-36-1 (online): <a href="http://www.ihe.ca/advanced-search/first-and-second-trimester-prenatal-screening-update">http://www.ihe.ca/advanced-search/first-and-second-trimester-prenatal-screening-update</a>   |

## Aim

To examine the safety, screening accuracy, therapeutic efficacy, patient outcomes, and cost-effectiveness of screening algorithms that incorporate the first trimester quadruple serum screen with or without nuchal translucency ultrasound (1T-Quad±NT) followed by testing of cell-free fetal DNA in maternal plasma (cff-DNA), commonly referred to as non-invasive prenatal testing (NIPT), for detecting fetal trisomies.

## Conclusions and results

### *Accuracy and effectiveness*

No empirical studies were identified that evaluated the accuracy and effectiveness of the 1T-Quad±NT + NIPT algorithm for prenatal screening of fetal trisomies. However, two modeling studies examined the performance of 1T-Quad, one of which analyzed 1T-Quad alone or contingently with NT or NIPT. The results of these two studies suggested that 1T-Quad±NT may be an appropriate and useful alternative to the first trimester combined screen.

NIPT on its own has been the focus of much recent research. Good evidence exists for the screening accuracy of NIPT using the massive parallel sequencing techniques (either shotgun or targeted approaches) as a second-tier screen for women whose pregnancies are considered high-risk for fetal trisomy based on first or second trimester screening results.

### *Economic outcomes*

A cost-effectiveness and budget impact model was developed using data from published evidence, an Alberta Health Services advisory group report, and Alberta administrative databases. When effectiveness is defined as the number of correctly diagnosed pregnancies, serum integrated prenatal screening (SIPS) + NIPT provides the best value for money. For options that provide results in the first trimester of pregnancy without using SIPS, 1T-Quad + NT + NIPT and 1T-Quad (with a detection rate of 0.85) + NIPT (when NT services are unavailable) are the most cost-effective screening algorithms. Establishing a systematic, province-wide screening program with increased coverage of pregnancies will result in net budget increases to physician, outpatient, and laboratory services.

## Recommendations

Current evidence supports the use of NIPT as a second-tier prenatal risk assessment for women whose pregnancies have been rated as high-risk. The 1T-Quad±NT may be a reasonable screening option for jurisdictions with limited resources for NT; however, uncertainty remains about its actual performance.

When effectiveness is defined as the number of correctly diagnosed pregnancies, SIPS + NIPT is the screening option that provides the best value for money. If the analysis focuses on options that provide information in the first trimester, 1T-Quad (with a detection rate of 0.85) + NIPT is the most cost-effective algorithm when NT services are unavailable. If NT provides additional benefit beyond that of detecting trisomy 21, then the most cost-effective option when NT services are available is 1T-Quad + NT + NIPT.

## Methods

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

## Further research/reviews required

The effects of 1T-Quad±NT or NIPT on patient and physician decision-making or maternal and fetal health are unknown. A Genome Canada-funded study is currently evaluating the NIPT and 1T-Quad±NT algorithm for prenatal screening of fetal trisomies. The results of this trial will likely provide robust evidence about the accuracy and effectiveness of this algorithm, in addition to answering questions about potential social and ethical issues.

## Written by

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