First-trimester prenatal screening for Down syndrome and other aneuploidies

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Record Status
This is a bibliographic record of a published health technology assessment from a member of INAHTA. No evaluation of the quality of this assessment has been made for the HTA database.

Citation

Authors' objectives
This report is a review of the published scientific literature on first-trimester prenatal screening for Down syndrome and other aneuploidies.

Authors' conclusions
Conclusions: The efficacy (under experimental conditions) of the different first-trimester prenatal screening modalities for Down syndrome and other aneuploidies is satisfactory, but it needs to be confirmed because of the methodological limitations of most of the studies. Despite the numerous studies involving more than 150,000 pregnancies, there are still some questions regarding effectiveness, especially that of nuchal translucency measurement in nonexperimental conditions.

Currently, it is impossible to state whether first-trimester or second-trimester screening is superior in terms of efficacy.

Different first-trimester prenatal screening modalities are already available in Quebec, both in the public and private sector.

First-trimester prenatal screening permits an earlier diagnosis than second-trimester screening. Consequently, pregnant women prefer this approach.

Implementing first-trimester screening will require changes to current prenatal care practice, mainly with regard to the week of pregnancy during which the pregnant woman's first medical visit takes place, the number of ultrasounds required and when, during the pregnancy, ultrasound is performed. Some of these changes are already being instituted in Quebec.

Prenatal Down syndrome screening should be included with all other prenatal screening activities and take into consideration the other diseases that these techniques might or might not be able to detect.

Recommendations: Based on the current state of knowledge, implementing wide-scale first-trimester screening in Quebec cannot be recommended. However, it is essential that current practices be guided in order to ensure the quality of the services provided. First-trimester screening should be restricted to university hospitals which have all the requirements for providing quality service and which agree to be evaluated. The primary objective of the evaluation would be to determine the effectiveness of the different modalities in the Quebec context. It should also make it possible to define the characteristics of the population and the service network and to determine the professionals' training needs regarding the techniques and genetic counselling, and the availability of appropriate equipment and the costs associated with screening in Quebec. It would also serve to determine the main aspects of developing and implementing quality control mechanisms, should the practice be expanded.

The conclusions of the 1999 CETS report, which examined second-trimester screening and diagnosis, still hold. Implementing second-trimester screening will make it possible to offer serum marker screening to all pregnant women who want it. It may also serve to set up genetic counselling services, which will be useful for all other types of prenatal screening and diagnosis. Eventually, it may become a complementary approach to or be replaced by first-trimester
screening. The results of research currently under way will make it possible to compare first-trimester screening and second-trimester screening and their usefulness when used alone or in combination.

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