

Health Quality Ontario

Let's make our health system healthier

Noninvasive Prenatal Testing for Trisomies 21, 18, and 13, Sex Chromosome Aneuploidies, and Microdeletions: Health Quality Ontario Recommendation

DRAFT RECOMMENDATION

- Health Quality Ontario, under the guidance of the Ontario Health Technology Advisory Committee, recommends that Ontario continue publicly funding noninvasive prenatal testing for pregnancies at high risk for trisomies 21, 18, and 13, and sex chromosome aneuploidies
- Health Quality Ontario, under the guidance of the Ontario Health Technology Advisory Committee, recommends against publicly funding noninvasive prenatal testing for pregnancies at average risk for trisomies 21, 18, and 13, sex chromosome aneuploidies, and microdeletions

RATIONALE FOR THE RECOMMENDATION

The Ontario Health Technology Advisory Committee has reviewed the findings of the health technology assessment¹ and an associated report summarizing the qualitative literature on noninvasive prenatal testing.²

The Ontario Health Technology Advisory Committee noted that noninvasive prenatal testing demonstrated accuracy as well as clinical and personal utility in the average-risk pregnant population and offers benefits that are important to pregnant people and their families. It provides information about the pregnancy early in gestation, with high accuracy and with no associated risk of procedure-related pregnancy loss. Pregnant people and their families value this information because they can use it to plan for the birth and care of an affected child, to understand reasons for spontaneous pregnancy loss, or to seek a diagnosis and consider termination of the pregnancy.

However, noninvasive prenatal testing is expensive, and it is uncertain whether it represents good value for money in pregnancies at average risk for a chromosomal anomaly, because the prevalence of the conditions tested for is very low in this population.

Public Comment: TBD

Draft — do not cite. Report is a work in progress and could change following public consultation.

The committee felt that the current standard of care, in which noninvasive prenatal testing is publicly funded for high-risk pregnancies (and as a second-tier test after a positive result from traditional prenatal testing) offered good value for money and is in keeping with patient and societal values and preferences.

After considerable deliberation, the committee also came to a consensus that publicly funding noninvasive prenatal testing for average-risk pregnancies would not represent good value for money given the current prices of the tests.

The committee also discussed concerns about the lack of appropriate supports for families to help them make informed decisions about testing. The committee discussed the need for an appropriate implementation plan and counselling services to support the current and future use of noninvasive prenatal testing. Committee members affirmed that noninvasive prenatal testing should be implemented along with support for people living with the conditions it screens for, while also recognizing that these considerations are beyond the scope of the current health technology assessment.

Decision Determinants for Noninvasive Prenatal Testing for Trisomies 21, 18, and 13, Sex Chromosome Aneuploidies, and Microdeletions

Decision Criteria	Subcriteria	Decision Determinants Considerations
Overall clinical benefit How likely is the health technology/intervention to result in high, moderate, or low overall benefit?	Effectiveness How effective is the health technology/intervention likely to be (taking into account any variability)?	Noninvasive prenatal testing was accurate in detecting trisomies 21, 18, and 13 (GRADE: low to moderate), and decreased the number of diagnostic tests performed (GRADE: moderate).
	Safety How safe is the health technology/intervention likely to be?	Noninvasive prenatal testing is a noninvasive blood test and does not harm the fetus.
	Burden of illness What is the likely size of the burden of illness pertaining to this health technology/intervention?	The prevalence of trisomies 21, 18, or 13 ranges from approximately 1 in 800 to 1 in 16,000 newborns. The prevalence of sex chromosome aneuploidies ranges from approximately 1 in 500 to 1 in 2,500 newborns. The prevalence of microdeletions ranges from approximately 1 in 4,000 to 1 in 50,000 newborns.
	Need How large is the need for this health technology/intervention?	At the time of this writing this report, noninvasive prenatal testing is publicly funded for pregnancies at high risk for trisomies 21, 18, and 13, and sex chromosome aneuploidies. Pregnant people at average risk for chromosomal anomalies pay out of pocket if they wish to proceed with noninvasive prenatal testing.
Consistency with expected societal and ethical values^a How likely is adoption of the health technology/intervention to be congruent with societal and ethical values?	Societal values How likely is adoption of the health technology/intervention to be congruent with expected societal values?	Pregnant people and others with lived experience of noninvasive prenatal testing were supportive of making this technology more widely available. People interviewed as part of this health technology assessment perceived that noninvasive prenatal testing could provide important information about pregnancies, earlier and with more accuracy. Publicly funding this test for all average-risk pregnant people may be congruent with societal expectations to provide accurate information early in pregnancy to assist in decision-making. However, publicly funding noninvasive prenatal testing only for high-risk pregnancies may be congruent with societal values to use public funding efficiently.
	Ethical values How likely is adoption of the health technology/intervention to be congruent with expected ethical values?	The ethical principle of autonomy and self-determination supports a person's informed decision to use noninvasive prenatal testing and related technologies. However, interviewees expressed ethical concerns about informed-choice discussions with their health care providers, and about the

Decision Criteria	Subcriteria	Decision Determinants Considerations
		<p>timing, delivery, and quality of information provided during those discussions.</p> <p>The principle of equity supports providing a health technology that offers substantial improvement in health outcomes to any person who needs it, regardless of their ability to pay.</p>
<p>Value for money</p> <p>How efficient is the health technology/intervention likely to be?</p>	<p>Economic evaluation</p> <p>How efficient is the health technology/intervention likely to be?</p>	<p>Compared with traditional prenatal screening, second-tier noninvasive prenatal testing improved overall prenatal screening performance by detecting more affected fetuses and substantially reducing the number of diagnostic tests performed, with decreased costs.</p> <p>Compared with second-tier noninvasive prenatal testing, first-tier noninvasive prenatal testing could detect more affected fetuses but would increase total prenatal screening costs considerably. The incremental cost to detect one more affected fetus would be \$411,274.</p>
<p>Feasibility of adoption into health system</p> <p>How feasible is it to adopt the health technology/intervention into the Ontario health care system?</p>	<p>Economic feasibility</p> <p>How economically feasible is the health technology/intervention?</p> <p>Organizational feasibility</p> <p>How organizationally feasible is it to implement the health technology/intervention?</p>	<p>Noninvasive prenatal testing is currently used as a second-tier test in average-risk pregnancies in Ontario. If it were used as a first-tier test instead, it would lead to an annual budget increase of approximately \$35 million per year.</p> <p>Noninvasive prenatal testing is already available in Ontario and is publicly funded for high-risk pregnancies. Public funding for average-risk pregnancies would require additional health care resources to pay for the increased testing and counselling needed.</p>

Abbreviation: GRADE, Grading of Recommendations Assessment, Development and Evaluation.

^aThe anticipated or assumed common ethical and societal values held in regard to the target condition, target population, and/or treatment options. Unless there is evidence from scientific sources to corroborate the true nature of the ethical and societal values, the expected values are considered.

REFERENCES

(1) TBD

(2) TBD

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