

# Prenatal Screening for Fetal Aneuploidy in Singleton Pregnancies

This clinical practice guideline has been prepared by the Genetics Committee of the Society of Obstetricians and Gynaecologists of Canada (SOGC) and the Prenatal Diagnosis Committee of the Canadian College of Medical Geneticists (CCMG). It was approved by both the Executive and Council of the Society of Obstetricians and Gynaecologists of Canada and the Board of Directors of the Canadian College of Medical Geneticists.

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**Key Words:** Aneuploidy, Down syndrome, trisomy, prenatal screening, genetic health risk, genetic health surveillance, prenatal diagnosis

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Disclosure statements have been received from all members of the committees.

## Abstract

**Objective:** To develop a Canadian consensus document on maternal screening for fetal aneuploidy (e.g., Down syndrome and trisomy 18) in singleton pregnancies.

**Options:** Pregnancy screening for fetal aneuploidy started in the mid 1960s, using maternal age as the screening test. New developments in maternal serum and ultrasound screening have made it possible to offer all pregnant patients a non-invasive screening test to assess their risk of having a fetus with aneuploidy to determine whether invasive prenatal diagnostic testing is necessary. This document reviews the options available for non-invasive screening and makes recommendations for Canadian patients and health care workers.

**Outcomes:** To offer non-invasive screening for fetal aneuploidy (trisomy 13, 18, 21) to all pregnant women. Invasive prenatal diagnosis would be offered to women who screen above a set risk cut-off level on non-invasive screening or to pregnant women whose personal, obstetrical, or family history places them at increased risk. Currently available non-invasive screening options include maternal age *combined* with one of the following: (1) first trimester screening (nuchal translucency, maternal age, and maternal serum biochemical markers), (2) second trimester serum screening (maternal age and maternal serum biochemical

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markers), or (3) 2-step integrated screening, which includes first and second trimester serum screening with or without nuchal translucency (integrated prenatal screen, serum integrated prenatal screening, contingent, and sequential). These options are reviewed, and recommendations are made.

**Evidence:** Studies published between 1982 and 2009 were retrieved through searches of PubMed or Medline and CINAHL and the Cochrane Library, using appropriate controlled vocabulary and key words (aneuploidy, Down syndrome, trisomy, prenatal screening, genetic health risk, genetic health surveillance, prenatal diagnosis). Results were restricted to systematic reviews, randomized controlled trials, and relevant observational studies. There were no language restrictions. Searches were updated on a regular basis and incorporated in the guideline to August 2010. Grey (unpublished) literature was identified through searching the websites of health technology assessment and health technology assessment-related agencies, clinical practice guideline collections, clinical trial registries, and national and international medical specialty societies. The previous Society of Obstetricians and Gynaecologists of Canada guidelines regarding prenatal screening were also reviewed in developing this clinical practice guideline.

**Values:** The quality of evidence was rated using the criteria described in the Report of the Canadian Task Force on Preventive Health Care.

**Benefits, harms, and costs:** This guideline is intended to reduce the number of prenatal invasive procedures done when maternal age is the only indication. This will have the benefit of reducing the numbers of normal pregnancies lost because of complications of invasive procedures. Any screening test has an inherent false-positive rate, which may result in undue anxiety. It is not possible at this time to undertake a detailed cost-benefit analysis of the implementation of this guideline, since this would require health surveillance and research and health resources not presently available; however, these factors need to be evaluated in a prospective approach by provincial and territorial initiatives.

### Recommendations

1. All pregnant women in Canada, regardless of age, should be offered, through an informed counselling process, the option of a prenatal screening test for the most common clinically significant fetal

aneuploidies in addition to a second trimester ultrasound for dating, assessment of fetal anatomy, and detection of multiples. (I-A)

2. Counselling must be non-directive and must respect a woman's right to accept or decline any or all of the testing or options offered at any point in the process. (III-A)
3. Maternal age alone is a poor minimum standard for prenatal screening for aneuploidy, and it should not be used as a basis for recommending invasive testing when non-invasive prenatal screening for aneuploidy is available. (II-2A)
4. Invasive prenatal diagnosis for cytogenetic analysis should not be performed without multiple marker screening results except for women who are at increased risk of fetal aneuploidy (a) because of ultrasound findings, (b) because the pregnancy was conceived by in vitro fertilization with intracytoplasmic sperm injection, or (c) because the woman or her partner has a history of a previous child or fetus with a chromosomal abnormality or is a carrier of a chromosome rearrangement that increases the risk of having a fetus with a chromosomal abnormality. (II-2E)
5. At minimum, any prenatal screen offered to Canadian women who present for care in the first trimester should have a detection rate of 75% with no more than a 3% false-positive rate. The performance of the screen should be substantiated by annual audit. (III-B)
6. The minimum standard for women presenting in the second trimester should be a screen that has a detection rate of 75% with no more than a 5% false-positive rate. The performance of the screen should be substantiated by annual audit. (III-B)
7. First trimester nuchal translucency should be interpreted for risk assessment only when measured by sonographers or sonologists trained and accredited for this service and when there is ongoing quality assurance (II-2A), and it should not be offered as a screen without biochemical markers in singleton pregnancies. (I-E)
8. Evaluation of the fetal nasal bone in the first trimester should not be incorporated as a screen unless it is performed by sonographers or sonologists trained and accredited for this service and there is ongoing quality assurance. (II-2E)
9. For women who undertake first trimester screening, second trimester serum alpha fetoprotein screening and/or ultrasound examination is recommended to screen for open neural tube defects. (II-1A)
10. Timely referral and access is critical for women and should be facilitated to ensure women are able to undergo the type of screening test they have chosen as first trimester screening. The first steps of integrated screening (with or without nuchal translucency), contingent, or sequential screening are performed in an early and relatively narrow time window. (II-1A)
11. Ultrasound dating should be performed if menstrual or conception dating is unreliable. For any abnormal serum screen calculated on the basis of menstrual dating, an ultrasound should be done to confirm gestational age. (II-1A)
12. The presence or absence of soft markers or anomalies in the 18- to 20-week ultrasound can be used to modify the a priori risk of aneuploidy established by age or prior screening. (II-2B)
13. Information such as gestational dating, maternal weight, ethnicity, insulin-dependent diabetes mellitus, and use of assisted reproduction technologies should be provided to the laboratory to improve accuracy of testing. (II-2A)
14. Health care providers should be aware of the screening modalities available in their province or territory. (III-B)
15. A reliable system needs to be in place ensuring timely reporting of results. (III-C)

### ABBREVIATIONS

AFP	alpha fetoprotein
CVS	chorionic villus sampling
DR	detection rate
FPR	false-positive rate
FTS	first trimester screening
hCG	human chorionic gonadotropin
IPS	integrated prenatal screening
MMS	multiple marker screening
MoM	multiples of the median
MSAFP	maternal serum alpha fetoprotein
NT	nuchal translucency
ONTD	open neural tube defect
PAPP-A	pregnancy-associated plasma protein-A
PR	positive rate
SLOS	Smith-Lemli-Opitz Syndrome
uE3	unconjugated estriol

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