No. 261 (Replaces No. 187, February 2007)

This guideline was peer reviewed by the SOGC's Genetics Committee in January 2017 and has been reaffirmed for continued use until further notice. Please note updated content can be reviewed in the Clinical Practice Guideline: No. 348-Joint SOGC-CCMG Guideline: Update on Prenatal Screening for Fetal Aneuploidy, Fetal Anomalies, and Adverse Pregnancy Outcomes [September 2017].

No. 261-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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Table 1. Key to evidence statements and grading of recommendations, using the ranking of the Canadian Task Force on Preventive Health Care

Quality of evidence assessment[a]	Classification of recommendations[b]
I: Evidence obtained from at least one properly randomized controlled trial.	A. There is good evidence to recommend the clinical preventive action.
II-1: Evidence from well-designed controlled trials without randomization.	B. There is fair evidence to recommend the clinical preventive action.
II-2: Evidence from well−designed cohort (prospective or retrospective) or case−control studies, preferably from more than one centre or research group.	C. The existing evidence is conflicting and does not allow to make a recommendation for or against use of the clinical preventive action; however, other factors may influence decision-making.
II-3: Evidence obtained from comparisons between times or places with or without the intervention . Dramatic results in uncontrolled experiments (such as the results of treatment with penicillin in the 1940s) could also be included in this category.	D. There is fair evidence to recommend against the clinical preventive action.
III: Opinions of respected authorities, based on clinical experience, descriptive studies, or reports of expert committees.	E. There is good evidence to recommend against the clinical preventive action.
	L. There is insufficient evidence (in quantity or quality) to make a recommendation; however, other factors may influence decision-making.

[a]The quality of evidence reported in these guidelines has been adapted from The Evaluation of Evidence criteria described in the Canadian Task Force on Preventive Health Care.[1]

[b]Recommendations included in these guidelines have been adapted from the Classification of Recommendations criteria described in the The Canadian Task Force on Preventive Health Care.[1]

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 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

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