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SOGC CLINICAL PRACTICE GUIDELINE

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No. 455, November 2024 (Replaces No. 192, April 2007, reaffirmed January 2016)

Clinical Consensus No. 455: Fetal Sex Determination and Disclosure

(En français : Déterminer et révéler le sexe fœtal)

The English document is the original version; translation may introduce small differences in the French version.

This Clinical Consensus Statement was prepared by the authors and overseen by the SOGC Diagnostic Imaging Committee. It was reviewed by the SOGC Diagnostic Imaging Committee and approved by the SOGC Guideline Management and Oversight Committee.

This Clinical Consensus Statement supersedes No. 192, published in April 2007, reaffirmed in January 2016.

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Keywords : diagnostic ultrasound; genitalia; fetal development; sex determination

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This document reflects emerging clinical and scientific advances as of the publication date and is subject to change. The information is not meant to dictate an exclusive course of treatment or procedure. Institutions are free to amend the recommendations. The SOGC suggests, however, that they adequately document any such amendments.

Informed consent: Patients have the right and responsibility to make informed decisions about their care, in partnership with their health care provider. To facilitate informed choice, patients should be provided with information and support that is evidence-based, culturally appropriate, and personalized. The values, beliefs, and individual needs of each patient in the context of their personal circumstances should be considered and the final decision about care and treatment options chosen by the patient should be respected.

Language and inclusivity: The SOGC recognizes the importance to be fully inclusive and when context is appropriate, gender-neutral language will be used. In other circumstances, we continue to use gendered language because of our mission to advance women's health. The SOGC recognizes and respects the rights of all people for whom the information in this document may apply, including but not limited to transgender, non-binary, and intersex people. The SOGC encourages health care providers to engage in respectful conversation with their patients about their gender identity and preferred gender pronouns and to apply these guidelines in a way that is sensitive to each person's needs.

Weeks Gestation Notation: The authors follow the World Health Organization's notation on gestational age: the first day of the last menstrual period is day 0 (of week 0); therefore, days 0 to 6 correspond to completed week 0, days 7 to 13 correspond to completed week 1, etc.

KEY MESSAGES

- Determination of fetal sex is an important component of obstetric ultrasound examination and should not be considered optional.
- Health care providers need to consider parental wishes regarding disclosure or nondisclosure of fetal sex when reporting ultrasound findings or reviewing the findings with parents.
- Health care providers need to be cognizant of increasing direct access of patients/parents to their ultrasound reports.

ABSTRACT

Objective: To provide guidance on ultrasound review of the fetal perineum as well as fetal sex determination and disclosure.

Target Population: All individuals with ongoing pregnancies.

Options: To include a review of the fetal perineum and determination of fetal sex as a component of the anatomic review during the routine second-trimester obstetric ultrasound and adhere to patient wishes regarding the disclosure of fetal sex.

Outcomes: Prenatal diagnosis of fetal genital and sex anomalies or variants, parental and pregnancy caregiver knowledge of fetal sex, and adherence to parental wishes regarding knowledge of fetal sex.

Benefits, Harms, and Costs: Benefits include the potential to improve perinatal outcomes through the diagnosis of fetal genital anomalies and respect for women's rightful autonomy over personal health information. Potential harms or costs include a possible error in fetal

sex determination, increased time for patients and health care providers in scheduling and performing the imaging, and the minimal risk of patients choosing to abort a pregnancy if the fetus is not the desired sex.

Evidence: Evidence built on the literature from the prior version of this statement through a review of international guidelines, Canadian legal rulings, and a literature search of PubMed and the Cochrane Database. English language research articles, review articles, and systematic reviews between January 1, 2003, and December 31, 2023, were included. Search terms included fetal ultrasonography, sex determination, and genitalia. The references of relevant articles were assessed, and applicable articles were included as well.

Intended Audience: All care providers for pregnant individuals in Canada.

Social Media Abstract: Fetal genitalia should be examined in pregnancy and the sex safely disclosed to the patient if they want this information.

CONSENSUS-BASED GOOD PRACTICE STATEMENTS:

- Diagnostic imaging providers and other health care providers should respect parental wishes regarding the disclosure of fetal sex.
- Diagnostic imaging providers should make every effort to determine fetal sex during an obstetric ultrasound examination from the second trimester onwards and should include this information in the ultrasound report.
- If fetal sex cannot be determined, re-examination or appropriate referral is recommended.
- 4. The obstetric ultrasound examination reports should contain a visible alert at the beginning of the report regarding the presence of information on fetal sex, thus giving parents the option of not reading any further.

INTRODUCTION

In Canada, diagnostic ultrasound units are inconsistent in their policies on the determination and disclosure of fetal sex to patients. Fetal sex determination by ultrasound has been recommended as part of the complete fetal anatomic review with the caveat that determination may be restricted by local practice or by difficulty in accurately determining the actual fetal sex. However, once fetal sex is determined, a patient's request for disclosure should be respected. This can be achieved by communicating directly with the patient but should always be documented in the report to the referring health professional. ^{1–3}

Good Practice Statement 1

FETAL SEX DETERMINATION AND DISCLOSURE

The Supreme Court of Canada (McInerney v. MacDonald [1992]) concluded that a patient is entitled to examine and copy from his or her medical record all information the physician considered in administering advice or treatment. The physician must justify denying access to the record on the basis that doing so would not be in the patient's best interest. Based on this ruling, it is legally difficult to defend nondisclosure. Disclosure of fetal sex upon request respects a woman's rightful autonomy over her personal health information.

Those who oppose fetal sex determination and disclosure have concerns about the risk of error, the time involved in determining fetal sex, and that the information may lead women to abort pregnancies when the fetus is not the wanted sex.

The risk of error in fetal sex determination is estimated to be less than 2% in the second and third trimesters, but prospective parents should be made aware of this possibility with disclosure.⁵ Fetal sex determination with first-trimester ultrasound is less reliable and, even in a focused research setting, was not possible in more than 10% of cases and was compounded with erroneous prediction occurring more than 10% of the time.⁶ This degree of error is not acceptable, and reliance on first-trimester ultrasound for determination of fetal sex is not recommended. If early identification of fetal sex is desired because of an increased risk of X-linked conditions or to

ABBREVIATIONS

cfDNA Cell-free DNA

NIPS Non-invasive prenatal screening

tailor the management of potential fetal congenital adrenal hyperplasia, non-invasive prenatal screening (NIPS) with cell-free DNA (cfDNA) is highly accurate and available after 10 weeks gestation.

Genital anomalies occur relatively frequently and are of clinical importance. For example, hypospadias occurs in 1 out of 250 live male infants born worldwide, 8,9 while Canadian data show that 1 in 156 males born in Nova Scotia have hypospadias. 10 Antenatal diagnosis of hypospadias can be difficult but is of clinical relevance as this anomaly is also associated with other genetic, anatomic, and growth abnormalities. As a more severe example, perineal anomalies such as bladder exstrophy often present with ambiguous genitalia, and antenatal diagnosis is therefore beneficial. These cases warrant review by multiple disciplines to optimize outcomes for affected pregnancies. Also, with the increasing use of NIPS to screen for common autosomal trisomies, pregnant women often choose to also screen for sex chromosome aneuploidy as an early way to determine fetal sex. Sex discordancy between NIPS genotype and ultrasound phenotype is possible, allowing for the possible identification of various conditions affecting normal sex differentiation, such as those seen with congenital adrenal hyperplasia in female fetuses.¹¹ It is for reasons such as these that examination of the fetal perineum should be considered part of the fetal anatomic review, and every effort should be made to have a complete assessment. Any concerns should also prompt referral to select specialties, usually starting with maternal-fetal medicine. Although there is no evidence that fetal sex determination extends the exam time, duration of examination is no longer of relevance given the clinical importance of accurately assessing this area of fetal anatomy.

Good Practice Statements 2 and 3

A small number of pregnant women may still consider abortion when the fetus is an unwanted sex; however, this matter is best addressed by the health professionals who are providing care for these women. Given that fetal sex can now be readily determined through maternal serum cfDNA as early as 10 weeks gestation, 12 it has become much less likely that fetal sex determination during the second-trimester ultrasound would significantly contribute to such abortions.

Although diagnostic imaging units may wish to maintain a policy of nondisclosure of fetal sex, this information should always be included in their reports. This would allow the referring physicians or midwives to not only

SOGC CLINICAL PRACTICE GUIDELINE

disclose the fetal sex at their patients' request but also, on rare occasions, use this information to assess genetic risks. However, both diagnostic imaging units and care providers should also recognize the potential that patients might access reports and inadvertently find out the fetal sex when their desire had been to not know this information. As a result, the SOGC recommends that a bolded alert always be placed at the start of the report indicating that fetal sex is included in the results. Patients will then know not to read further, if applicable.

Good Practice Statement 4

CONCLUSION

In summary, the SOGC recommends that fetal genitalia be assessed as a part of the routine second-trimester obstetric ultrasound⁹ and that this examination now be considered an essential element in the complete anatomic review. Fetal sex should be determined, and a patient's request for disclosure should be respected. This could be communicated directly to the patient but should always be part of the report to the referring health professional. A bolded alert should always be placed at the start of the report indicating that fetal sex is included. If fetal sex cannot be determined, re-examination or appropriate referral is recommended.

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