Office Guide to Prenatal Screening Options for Chromosome Differences

PRENATAL SCREENING
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Singleton Pregnancies

Questions to consider when offering prenatal genetic screening	enhanced First Trimester Screening (eFTS)	Second Trimester Screening (STS)[a]	Non-Invasive Prenatal Testing (NIPT)
When in pregnancy is it done?	11 weeks and 2 days to 13 weeks and 3 days	14 weeks and 0 days to 20 weeks and 6 days	9–10 weeks to the end of the pregnancy
Is it publicly funded?	Yes	Yes	Yes, if one of the eligibility criteria is met (see next page). Private-pay is an option if the test cannot be publicly funded.
What does it include?	Blood test and nuchal translucency ultrasound	Blood test	Blood test
What does it screen for?	• trisomy 21 • trisomy 18	• trisomy 21 • trisomy 18	• trisomy 21 • trisomy 18 • trisomy 13 • +/- sex chromosome differences[b,c,d]
What are its limitations?	Other genetic and physical differences are not screened for. However, extra information can be gained from the nuchal translucency and 18-22 week ultrasounds.	Other genetic and physical differences are not screened for. Extra information can be gained from the 18-22 week ultrasound.	Other genetic and physical differences are not screened for. If NIPT is performed, an 11-14 week ultrasound (with nuchal translucency evaluation where available) and an 18-22 week ultrasound should still be offered.[e]
What is the **detection rate for trisomy 21**?[f] (meaning, how many pregnancies where the baby really does have trisomy 21 will be flagged as "screen positive" / "high risk" by this test?)	88.0%	87.5%	More than 99%
What is the **false positive rate for trisomy 21**? (meaning, how many pregnancies will this test flag as "screen positive" / "high risk" but the baby does not really have trisomy 21?)	6.0%	8.4%	Less than 0.1%
What is the **detection rate for trisomy 18**? (meaning, how many pregnancies where the baby really does have trisomy 18 will be flagged as "screen positive" / "high risk" by this test?)	86.8%	S (47.1-86.8%)[h]	96.3%
What is the **false positive rate for trisomy 18**? (meaning, how many pregnancies will this test flag as "screen positive" / "high risk" but the baby does not really have trisomy 18?)	Less than 1%	Less than 1%	Less than 0.1%
What is the chance it will not give an informative result?	Less than 0.1%	Less than 0.1%	In an Ontario study, 4.8% of NIPTs did not give a result after the first attempt.[g]

a- Second Trimester Screening (STS) is formerly known as Maternal Serum Screening (MSS). This form of screening should be offered only when eFTS is not available.
b- In pregnancies where oocyte (egg) donors or gestational carriers (surrogates) are used, sex chromosome differences cannot be screened through Panorama™ NIPT (LifeLabs ®).
c- In addition to these conditions, Panorama™ NIPT (LifeLabs ®) may suggest the chance of triploidy, vanishing twins or previously unrecognized twin pregnancies. The test cannot distinguish between these possibilities, and further testing might be warranted
d- Screening for microdeletion syndromes may be available as a private-pay option. However, most practice guidelines do not currently support the use of NIPT to screen for microdeletion syndromes due to insufficient data on the technical and clinical validity of screening for these syndromes.
e- Audibert et al. No. 456 SOGC Guideline. J Obstet Gynaecol Can 2024;46(11):102694.
f- Detection and false positive rates were obtained from Ontario pregnancies with a due date between Sept. 2016 and Dec. 2023 (see Performance of Prenatal Screening Tests on the PSO website for more information).
g- Bellai-Dussault et al. Cytogenic outcomes following a failed cell-free DNA screen. Am J Obstet Gynaecol 2023;229(2):168e1-168,e8.
h- S=point estimate suppressed when confidence interval >20%.

Twin Pregnancies

All viable twin pregnancies are eligible for publicly funded NIPT. An 11-14 week (nuchal translucency) ultrasound and an 18-22 week ultrasound should still be offered (1).

The NIPT performance for the detection of trisomies 21, 18 and 13 in twins has been shown to be comparable to singletons (2, 3). However, there is a higher chance of NIPT failing to give a conclusive result than in singletons (2).

Lab-specific factors to consider when offering NIPT for twin pregnancies:

Type of twin pregnancy	Can NIPT be done through Panorama™ NIPT (LifeLabs®)?	Can NIPT be done through Harmony™ Prenatal Test (Dynacare®)?
Conceived with pregnant individual's own oocytes (eggs)	• Yes (trisomies 21/18/13)[a] • The result is provided for the pregnancy, not each twin • Identifies whether twins are likely monozygotic (identical) or dizygotic (fraternal)[b] • Fetal sex is provided for each twin	• Yes (trisomies 21/18/13)[c] • The result is provided for the pregnancy, not each twin • Fetal sex is not provided for each twin. The presence of a Y chromosome indicates at least one fetus is male.
Conceived with an oocyte (egg) donor	No (only for singletons)	Yes (trisomies 21/18/13)
Gestational carrier (surrogate) is involved	No (only for singletons)	Yes (trisomies 21/18/13)

a- Only monozygotic twins can be screened for sex chromosome differences with Panorama™ NIPT.
b- Zygosity testing through NIPT can complement, but not replace, ultrasound evaluation of chorionicity (Benn et al. Non-invasive prenatal testing in the management of twin pregnancies. Prenat Diagn 2021;41:1233-40).
c- Twins cannot be screened for sex chromosome differences with Harmony™ Prenatal Test.

Vanishing Twin Pregnancies

A "vanishing" twin scenario is when a pregnancy begins as a twin pregnancy and one twin miscarries, leading to a singleton pregnancy.

The optimal prenatal genetic screening modality for vanishing twin pregnancies is a nuchal translucency ultrasound followed by Second Trimester Screening (STS). The STS must be done at least 8 weeks from the time of demise of the vanishing twin. If this ultrasound is not available, STS can be done on its own.

Other forms of prenatal genetic screening (e.g. eFTS or NIPT) are not recommended in this scenario due to the risk of inaccurate results (4,5).

Eligibility Criteria for Publicly Funded NIPT

Publicly funded NIPT can be ordered by health-care practitioners through two provincial laboratories: Dynacare® (Harmony™ Prenatal Test) and LifeLabs® (Panorama™ NIPT) if at least one of the following criteria is met:

Category I

Please select all that apply:

- ☐ A multiple marker screening test (e.g. eFTS/STS) positive for aneuploidy.
- ☐ The age of the pregnant individual will be 40 years or older at the expected date of delivery. In the context of in vitro fertilization, this is defined as the oocyte (egg) age at retrieval being 39 or older (whether self-provided or donor oocyte).
- ☐ Previous pregnancy or child with trisomy 21, 18, or 13.
- ☐ Twins with ultrasound demonstration of fetal heart activity in both fetuses.
- ☐ Increased nuchal translucency (NT) ≥3.5mm*.

  ** NT ≥3.5 mm can be associated with genetic conditions and structural differences not detected by NIPT. Prompt referral to a Genetics or Maternal Fetal Medicine (MFM) specialist is indicated regardless of NIPT order or result.*

Category II

The following situations require specialist consultation to determine whether NIPT is warranted and to provide appropriate pre- and post-test counselling.
Select all that apply (must be completed by a Genetics or MFM specialist):

- ☐ Fetal congenital anomalies suggestive of trisomy 21, 18 or 13 identified on ultrasound. Specify_____.
- ☐ Cystic hygroma
- ☐ Isolated soft marker with moderate likelihood ratio for trisomy 21 (must check one or more):
  - ☐ Increased nuchal fold (≥6mm)
  - ☐ Absent/hypoplastic nasal bone
- ☐ Multiple soft markers with low likelihood ratio for trisomy 21 (must check two or more):
  - ☐ Aberrant right subclavian artery
  - ☐ Pyelectasis
  - ☐ Clinodactyly
  - ☐ Hyperechogenic bowel
  - ☐ Intracardiac echogenic focus / foci
  - ☐ Short femur
  - ☐ Short humerus
  - ☐ Ventriculomegaly
  - ☐ Other, specify: _____
- ☐ NIPT for sex chromosome determination (at least one of the following):
  - ☐ Risk of a sex-linked condition.
  - ☐ Ultrasound findings suggestive of a sex chromosome aneuploidy.
  - ☐ Ultrasound findings suggestive of a difference/disorder of sex development.

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***Updated 3/2026**: Visit our website regularly to obtain the most up-to-date version of this living document.*

References
1. Audibert et al. No. 456 SOGC Guideline. J Obstet Gynaecol Can 2024;46(11):102694.
2. Palomaki et al. International Society for Prenatal Diagnosis Position Statement. Prenat Diagn. 2020;1-11.
3. Dungan et al. Noninvasive prenatal screening (NIPS) for fetal chromosome abnormalities in a general-risk population: An evidence-based clinical guideline of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2023 Feb:25(2):100336.
4. Screening for fetal chromosomal abnormalities. ACOG Practice Bulletin No. 226. American College of Obstetricians and Gynecologists. Obstet Gynecol 2020;136:e48-69.
5. Hopkins and Dugoff. Screening for aneuploidy in twins. Am J Obstet Gynecol MFM. 2022 Mar;4(2S):100499.