

## Noninvasive Prenatal Screening via Cell-Free DNA [Trisomy 13: Positive Cell Free DNA Screen]

**Etiologies of Positive Screen:** Fetal trisomy 13, fetal mosaic trisomy 13, confined placental mosaicism for trisomy 13, karyotypic abnormalities resulting in extra chromosome 13 material, vanishing twin.

**Clinical Considerations:** Trisomy 13 (Patau Syndrome) is a multi-system condition resulting in severe birth defects and intellectual disability. Intrauterine growth restriction and intrauterine fetal demise are common. Of those that survive pregnancy, long-term survival is rare.

**Screening Considerations:** Noninvasive prenatal screening (NIPS) via cell-free DNA is designed to detect fetal aneuploidy including trisomy 13. The detection rate is approximately 99% with a false positive rate of less than 1%. The positive predictive value (PPV) of NIPS for trisomy 13 is lower than for trisomy 21 and 18 and varies with maternal age: the PPV is approximately 10% at age 20 and 60% at age 40. Major reproductive decisions should not be made without clinical or laboratory confirmation. Prenatal ultrasound demonstrates an anomaly in more than 90% of fetuses. Diagnostic testing should be offered to confirm a screen positive test result. Expedited referrals to a genetics and/or maternal and fetal medicine (MFM) professional should be made to ensure timely, informed decision making and management.

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### ***YOU SHOULD TAKE THE FOLLOWING IMMEDIATE ACTIONS:***

- Inform patient of the test result and emphasize that, to confirm the diagnosis, a referral to MFM and/or a provider with prenatal genetics expertise will be offered for additional testing, including genetic counseling, fetal ultrasound, and diagnostic testing.
- Emphasize that the positive predictive value (PPV) of NIPS for trisomy 13 is lower than for other trisomies and depends on maternal age and ultrasound findings.
- Every patient has the right to accept or decline genetic screening or diagnosis.

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**Diagnostic Evaluation:** Karyotype or microarray analysis of chorionic villi or amniocytes is needed to confirm the diagnosis and distinguish between the various possible mechanisms listed above, which can inform clinical decision making and determine recurrence risk.

### **Patient Education Resources**

[Kidshealth.org](http://Kidshealth.org)

[Medline Plus](http://Medline Plus)

[PerinatalHospice.org](http://PerinatalHospice.org)

[Trisomy.org](http://Trisomy.org)

### **Provider Education Resources**

[ACMG- Noninvasive Prenatal Screening for Fetal Aneuploidy](#)

[ObG Project](#)

[UNC Positive Predictive Value Calculator](#)

[Perinatal Quality Positive Predictive Value Calculator](#)

### **Referral (local, state, regional and national):**

[Find Genetic Services](#)

[NSGC Find a Genetic Counselor](#)

[Society for Maternal Fetal Medicine Find an MFM](#)

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

**LOCAL RESOURCES:** Insert local web site links

State Resource site *(insert website information)*

Name

URL

Comments

Local Resource Site *(insert local and regional website information)*

Name

URL

Comments

APPENDIX: Resources with Full URL Addresses

*Additional Information:*

### Patient Education Resources

Kidshealth.org

- <https://kidshealth.org/>

Medline Plus

- <https://medlineplus.gov/genetics/condition/trisomy-13/>

PerinatalHospice.org

- <https://www.perinatalhospice.org/>

Trisomy.org

- <https://www.trisomy.org/>

### Provider Education Resources

ACMG- Noninvasive Prenatal Screening for Fetal Aneuploidy

- <https://www.acmg.net/PDFLibrary/Fetal-Aneuploidy-Noninvasive-Prenatal-Screening-Update.pdf>

ObG Project

- <https://www.obgproject.com/2016/07/21/trisomy-13-what-is-it/>

UNC Positive Predictive Value Calculator

- <https://www.med.unc.edu/mfm/nips-calc/>

Perinatal Quality Positive Predictive Value Calculator

- <https://www.perinatalquality.org/Vendors/NSGC/NIPT/>

### Referral (local, state, regional and national):

Find Genetic Services

- <https://clinics.acmg.net>

NSGC Find a Genetic Counselor

- <https://www.nsgc.org/page/find-a-genetic-counselor>

SMFM Find a Maternal and Fetal Medicine Specialist

- <https://www.smfm.org/members>

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