Prenatal Cell-Free DNA Screening ACT Sheet

[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: Prenatal Cell-Free DNA Screening 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 Prenatal Cell-Free DNA Screening 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.

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 Prenatal Cell-Free DNA Screening 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.

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
Medline Plus
PerinatalHospice.org
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

ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

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

ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

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