

FINAL RESULTS SUMMARY : TWINS

Result

Insufficient fetal DNA

Zygosity

Dizygotic

Fetal Sex

N/A

Fetal Fraction(s)

2.7%, 2.6%



FRATERNAL TWINS



A risk assessment for conditions screened could not be produced due to insufficient fetal DNA or borderline low fetal fraction in combination with other factors in this sample. Low fetal fraction could be attributed to normal variation, gestational age, maternal weight, and certain medications; also, this finding may be associated with some chromosome abnormalities. Review of the clinical history, ultrasound evaluation, and the option of diagnostic testing is recommended (ACOG Practice Bulletin, 226, 2020). Natera will accept a repeat sample. The likelihood of success with a redraw is summarized in the redraw success table found on page 2 of this report.

RESULT DETAILS: ANEUPLOIDIES

Condition Tested ¹	Result	Risk Before Test ²	Risk After Test
Trisomy 21	No Result	1/255	N/A
Trisomy 18	No Result	1/594	N/A
Trisomy 13	No Result	1/1,871	N/A

1. Reporting for Monosomy X, Trisomy 16, and microdeletion syndromes is not available for dizygotic twin pregnancies. Excludes cases with evidence of fetal and/or placental mosaicism. 2. Based on maternal age, gestational age, and/or general population, as applicable. References available upon request.

Testing Methodology: DNA isolated from maternal blood, which contains placental DNA, is amplified at specific loci using a targeted PCR assay and is sequenced using a high-throughput sequencer. Fetal fraction is determined using a proprietary algorithm incorporating data from single nucleotide polymorphism-based (SNP-based) next-generation sequencing (Pergament E et al. *Obstet Gynecol*. 2014 Aug;124(2 Pt 1):210-6). If there is sufficient fetal fraction, sequencing data is analyzed using a proprietary SNP-based algorithm to determine the fetal copy number for chromosomes 13, 18, 21, X and Y. If ordered, specific microdeletions will be evaluated using similar methodology (Wagner RJ et al. *Am J Obstet Gynecol*. 2005 Mar;212(3):332 e1-9). If the fetal fraction is insufficient, fetal signal enhancement and/or an additional algorithm to determine whether there is an increased risk for trisomy 18, and trisomy 13 may be utilized (McKenna et al. *Ultrasound Obstet Gynecol* 2019; 53:73-79). However, some samples will not produce a result due to failure to meet the necessary quality thresholds.

This test has been validated on women with a singleton, twin or egg donor pregnancy of at least nine weeks gestation. A result will not be available for higher order multiples and multiple gestation pregnancies with an egg donor or surrogate, or bone marrow transplant recipients. Complete test panel is not available for twin gestations and pregnancies achieved with an egg donor or surrogate. For twin pregnancies with a fetal fraction value below the threshold for analysis, a sum of the fetal fractions for both twins will be reported. As this assay is a screening test and not diagnostic, false positives and false negatives can occur. High risk test results need diagnostic confirmation by alternative testing methods. Low risk results do not fully exclude the diagnosis of any of the syndromes nor do they exclude the possibility of other chromosomal abnormalities or birth defects, which are not a part of this test. Potential sources of inaccurate results include, but are not limited to, mosaicism, low fetal fraction, limitations of current diagnostic techniques, or misidentification of samples. This test will not identify all deletions associated with each microdeletion syndrome. This test has been validated for deletions >0.5 Mb within the 22q11.2 A-C region. This test has been validated on full region deletions only for 1p36 deletion syndrome, Cri-du-chat syndrome, Prader-Willi syndrome and Angelman syndrome and may be unable to detect smaller deletions. Microdeletion risk scores may be dependent upon fetal fraction, as deletions on the maternally inherited copy are difficult to identify at lower fetal fractions. Test results should always be interpreted by a clinician in the context of clinical and familial data with the availability of genetic counseling when appropriate.

Disclaimer: This test was performed by Natera, Inc. 201 Industrial Rd, Suite 400, San Carlos, CA 94070 (CUSA ID 0001082790). The performance characteristics of this test were developed by Natera, Inc. This test has not been cleared or approved by the U.S. Food and Drug Administration (FDA). This laboratory is regulated under CLIA as qualified to perform high-complexity testing. © 2023 Natera, Inc. All Rights Reserved.

Reviewed By:  Wendy Wu, M.D., Ph.D., FACMG, Senior Laboratory Director

QA/Laboratory Director | Sherry Ross-Kuo, Ph.D., FACMG

IF THE ORDERING PROVIDER HAS QUESTIONS OR WISHES TO DISCUSS THE RESULTS, PLEASE CONTACT US AT 844-778-4700, option 2. Ask for the NIPT genetic counselor on call.

The Fraction Twins

J Ma



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