Non-Invasive Prenatal Testing for Fetal Aneuploidy

Patient:
DOB: Age: Gender:
Patient Identifiers:

Visit Number (FIN):

Client:

Physician:

ARUP Test Code: 2007537

Collection Date: 12/05/2016
Received in lab: 12/06/2016
Completion Date: 12/12/2016

Patient Information Used in Risk Calculations

Maternal Age at Delivery:	years	Maternal Weight:	134 lbs
Estimated Due Date:	09 Jun 2017	Report Fetal Sex?	Yes
Gestational Age at Draw:	13 weeks 3 days		

Results Summary

HIGH RISK Monosomy X

Fetal Fraction: **6.4%**

Fetal Sex: **Female**

This pregnancy is classified as HIGH RISK for monosomy X (Turner syndrome) by this screen. This result should be confirmed by a diagnostic test. On average, 50% of pregnancies classified as HIGH RISK are found to have monosomy X based on a published study of 17,885 women (PMID 25111587).

This is a screening test, and is NOT diagnostic for the conditions listed in this report. Both false positive and false negative results may occur. Genetic counseling and confirmatory fetal diagnostic testing is recommended. Irrevocable action such as pregnancy termination should not be taken based on the results of this test alone.

Conditions Screened

Trisomy 21:	Low risk
Trisomy 18:	Low risk
Trisomy 13:	Low risk
Monosomy X:	**HIGH RISK**
Triploidy/Vanishing twin:	Low risk

TEST INFORMATION: Non-Invasive Prenatal Testing for Fetal Aneuploidy (Powered by Constellation)

METHODOLOGY: DNA isolated from the maternal blood, which contains placental DNA, is amplified at >13,300 loci using a targeted PCR assay and sequenced using a high-throughput sequencer. Sequence data is analyzed using Natera's Constellation software to estimate the fetal copy number for chromosomes 13, 18, 21, X, and Y, thereby identifying whole chromosome abnormalities at those chromosomes as well as fetal sex. Barring QC failures and fetal fractions below the performance limits of the algorithm, the minimum confidence threshold is 0.98 for a High Risk call, and 0.85 for a Low Risk call. The majority of specimens will have a confidence of >0.99 across all regions tested. If a sample fails to meet the quality threshold, no result will be reported for one or more chromosomes.

DISCLAIMER: Results exclude cases with evidence of fetal and/or placental mosaicism. Risk scores are calculated based on maternal age, gestational age and test results. Findings of unknown significance and possible non-paternity will not be reported. As this assay is a screening test and not diagnostic, false positive and false negatives can occur. Positive test results need diagnostic confirmation by alternative testing methods. Negative results do not fully exclude the diagnosis of any of the above syndromes. False positive and false negative results may be due to placental, fetal or maternal mosaicism, small imbalances, point mutations, gene inactivation, haploblocks, or other genetic/epigenetic mechanisms. Other potential sources of error include, but are not limited to, DNA sample contamination or degradation, limitations of current diagnostic techniques, misidentification of samples, or other factors that may interfere with correct interpretation of the analysis. This test has the potential to uncover consanguinity in the family. This test is not intended to identify pregnancies at risk for open neural tube defects. This test was developed and its performance characteristics determined by ARUP Laboratories. The U.S. Food and Drug Administration has not approved or cleared this test; however, FDA clearance or approval is not currently required for clinical use. The results are not intended to be used as the sole means for clinical diagnosis or patient management decisions.