

Reporting facility address:

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Referrer contact details:

Dr. Allister
Fetal Medicine Centre
123 Big Hill
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United Kingdom

PATIENT DETAILS:

Patient ID	02952	Maternal Age (at test)	34 years
Patient Surname	Waterman	Gestation Age (at test)	13 weeks 5 days
Patient Forename	Joselyn	Patient Date of Birth	06 Jul 1979
Clinician Name	Barbara Banks	Date of Blood Draw	19 May 2014
Hospital/Clinic Name	Fetal Medicine Centre	Pregnancy Status: Singleton/Twin	Singleton

TEST RESULTS:

TRISOMY	BACKGROUND RISK	The IONA [®] test RISK SCORE	CLINICAL SUMMARY
TRISOMY 21 (Down's Syndrome)	1 : 105 ^{CT}	> 95%	HIGH RISK INVASIVE TEST RECOMMENDED
TRISOMY 18 (Edwards' Syndrome)	1 : 797	< 1 : 1,000,000 (<0.0001%)	LOW RISK
TRISOMY 13 (Patau's Syndrome)	1 : 2487	< 1 : 1,000,000 (<0.0001%)	LOW RISK

Fetal Fraction	4%
Fetal Sex	Female

The IONA[®] test is indicated for screening NOT diagnosis — (results should be reviewed and discussed with your healthcare provider)

SUPPLEMENTARY INFORMATION:

- The detection rate of the IONA[®] test for trisomies 21, 18 and 13 is >99%.
- If fetal sex determination is requested, the accuracy is 99%. A "Sex Determination Failure" result may be reported if there is insufficient data to support the sex determination analysis. This is separate from the trisomy analysis and does not reflect on the quality of any other result generated by the IONA[®] test.
- The IONA[®] test estimates the risk of trisomies by determining the relative amounts of chromosomes 13, 18 and 21 in placentally-derived cell-free DNA extracted from the mother's plasma. The adjusted risk accounts for the background risk of the mother at the time of sampling (default). Additionally, the test may use the results of the First Trimester Combined Test as the background risk. If this has been done, a superscript ^{CT} will appear by the background risk next to any, or all, of the trisomy results.
- The IONA[®] test is a screening test and a high risk result should be discussed with the healthcare professional and confirmed by an appropriate diagnostic test (e.g. amniocentesis).
- The maternal age-adjusted risk score is capped. The cap is derived from an estimate of the prevalence of biological factors such as placental mosaicism. The result caps are: T21 >95%, T18 >75% and T13 >60%. These are the maximum risk estimates displayed on the report.
- In dichorionic twins, scientific publications suggest that the detection rate is reduced from greater than 99% to about 95%.
- A result with an IONA[®] test risk score greater than or equal to 1:150 (~0.67%) is considered high risk.

Originating sample ID: **S00002952**

Sample notes (if entered):

Sequencing run and sample validity checks passed: **Yes**

IONA[®] Software version: **TOA: 1.7.0.7833.746; DAA: 1.7.0.7833.572**