

Reporting facility address	Referrer contact details	PATIENT DETAILS			
		Patient ID		Maternal Age (at test)	
		Patient Surname		Gestation Age (at test)	
		Patient Forename		Patient Date of Birth	
		Clinician Name		Date of Blood Draw	
		Hospital/Clinic Name		Pregnancy Status:	
		IVF Pregnancy		Donor Age (at egg harvest)	

SCREENING TEST RESULTS			
TRISOMY	BACKGROUND RISK	The IONA [®] test RISK SCORE	CLINICAL SUMMARY
TRISOMY 21 (Down's Syndrome)	1 : 85	Less than 1 : 10,000 (<0.01%)	LOW RISK
TRISOMY 18 (Edwards' Syndrome)	1 : 213	Less than 1 : 10,000 (<0.01%)	LOW RISK
TRISOMY 13 (Patau's Syndrome)	1 : 667	Less than 1 : 10,000 (<0.01%)	LOW RISK

Autosomal Aneuploidies	NOT DETECTED
Sex chromosome results	XX
Fetal Sex	Female
Estimated Fetal Fraction	9%

Supplementary Information: Trisomy 13, 18 and 21 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 (cfDNA) extracted from maternal peripheral whole blood using whole genome shotgun next generation sequencing (NGS). The final IONA[®] adjusted risk score incorporates both the fetal fraction and background risk score at the time of sampling (maternal age by default). If the results of a First Trimester Combined Test are used, a superscript ^{CT} will appear by the background risk next to any, or all, of the trisomy results. A risk score greater than or equal to 1:150 (~0.67%) is considered high risk. This risk score is capped based on an estimated prevalence of biological factors such as placental mosaicism. 95% is the maximum risk score estimates displayed on the report. The IONA[®] test is a screening test and a high risk result should be discussed with a healthcare professional in the context of all available clinical findings, including advising for the need for genetic counselling or additional diagnostic testing (e.g. amniocentesis).

	Observed Sensitivity (2 sided 95% CI)	Observed Specificity (2 sided 95% CI)
T21*	>99.99% (92.3%, 100.0%)	99.5% (98.3%, 99.9%)
T18*	>99.99% (83.2%, 100.0%)	99.8% (98.8%, 100.0%)
T13*	>99.99% (69.2%, 100.0%)	>99.99% (99.2%, 100.0%)

Supplementary Information: Autosomal Aneuploidy (AA) The AA analysis detects full trisomy and full monosomy in the additional 19 human autosomal chromosomes. If Trisomy/Monosomy is detected, the result is displayed as 'Detected' and the chromosome listed. 'Not detected' is reported when no Trisomy/Monosomy is detected. This analysis is separate from the trisomy analysis and does not reflect on the quality of the result generated by the IONA[®] test. AA has been validated in singleton and monochorionic twin pregnancies.

Supplementary Information: Sex Chromosome Results The analysis measures proportions of the X and Y chromosomes present in cfDNA to infer on fetal sex with an accuracy of 99.3% and to infer on sex chromosome aneuploidies (SCAs). A valid result would be either reported as "female" or "male". In the presence of a sex chromosome aneuploidy, results are reported as: XO, XXY, XYY, XXX (XYY result indicates two or more fetal Y chromosomes). In the absence of an aneuploidy, the results are reported as XX or XY. Sex determination and sex chromosome aneuploidy testing has been validated in singleton and monochorionic twin pregnancies. This test is separate from the trisomy analysis and does not reflect on the quality of any other result generated by the IONA[®] test.

Test Limitations In rare cases, analysis of cfDNA derived from the fetoplacental unit may not correlate with the fetal genotype. The IONA[®] test may be affected by any chromosomal change which affects the genomic ratio. Thus, inaccurate test results or a failure to obtain test results may occur in rare circumstances which include but are not limited to biological factors affecting the mother including those arising during pregnancy; conditions affecting the pregnancy or fetus such as confined placental mosaicism (CPM), partial trisomy or translocations, intrauterine fetal demise or disappearing twin. This list is inclusive but not exhaustive. The IONA[®] does not screen for or rule out the possibility of other genetic conditions, balanced or unbalanced chromosomal abnormalities or mosaic anomalies being present other than those expressly identified in this document. The IONA[®] test is not intended to identify pregnancies at risk for open neural tube defects and is not a replacement for routine pregnancy monitoring.

Originating sample ID: N/A

Sample notes (if entered):

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

IONA[®] Software version: **TOA: 2.0.2.1912; DAA: 2.0.2.1769**