

NIPT REPORT

Genetics - the SAFE test Laboratory
Jenner Wing - Basement - Room 01.242
St George's University Hospitals NHS Foundation Trust
Blackshaw Road
London
SW17 0QT

Prof. Basky Thilaganathan (Clinical Lead)
John Short (Scientific Lead)
Tel 0208 725 5864
Email: theSAFEtest@nhs.net
www.theSAFEtest.co.uk

PATIENT DETAILS:

Patient ID	Pt1234	Patient Date of Birth	01 May 1982
Patient Surname	Doe	Maternal Age (at test)	35 years
Patient Forename	Jane	Gestation Age (at test)	12 weeks 2 days
Clinician Name	N/A	Singleton/Twin Pregnancy?	Singleton
Hospital/Clinic Name	N/A	Date of Blood Draw	10 May 2018
Requester's Contact Details	N/A	Patient Location	N/A

TEST RESULTS:

TRISOMY	*BACKGROUND RISK (before the SAFE test)	SAFE TEST RISK SCORE	CLINICAL SUMMARY
Trisomy 21	1 in 38 ^{PT}	Greater than 95%	HIGH CHANCE — INVASIVE TEST RECOMMENDED
Trisomy 18	1 in 537 ^{PT}	Less than 1 in 1,000,000 (<0.0001%)	LOW CHANCE
Trisomy 13	1 in 537 ^{PT}	Less than 1 in 1,000,000 (<0.0001%)	LOW CHANCE

the SAFE test is indicated for screening NOT diagnosis — (results should be reviewed and discussed with the healthcare provider)

SUPPLEMENTARY INFORMATION FOR HEALTHCARE PROVIDERS:

Originating sample ID: **1234ExampleReport**
Sequencing run and sample validity checks passed: **YES**
Sample notes (if entered):
Report generated: 23 May 2018 11:18 (revision 1)
IONA® Software version: **TOA: 1.7.0.7833.746; DAA: 1.7.0.7833.572**

- The detection rate of the SAFE test for trisomies 21, 18 and 13 is >99%.
- The SAFE 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 SAFE test measures fetal fraction and the final risk score accounts for both the fetal fraction and background risk of the mother at the time of sampling (default). Additionally, the test may use the results of a Prior Risk test as the background risk. If this has been done, a superscript ^{PT} will appear by the background risk next to any, or all, of the trisomy results.
- The SAFE test is a screening test and an increased risk result should be discussed with the healthcare provider and confirmed by an appropriate diagnostic test following an invasive procedure (e.g. amniocentesis).
- The 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: Trisomy 21 >95%, Trisomy 18 >75%, Trisomy 13 >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 a SAFE test risk score greater than or equal to 1 in 150 (~0.67%) is considered high risk.
- Test methodology - whole genome shotgun sequencing.
- Test performed using peripheral whole blood.