Disease — Cell-Free Fetal DNA Sex Determination

Contact details

Regional Genetics Service
Level 5/6, Barclay House
37 Queen Square
London, WC1N 3BH
T +44 (0) 20 7762 6888
F +44 (0) 20 7813 8578

Samples required

- **Pregnant Women**
  2x 10mls venous blood in plastic EDTA bottles or glass Streck tubes*
- **Testing must be arranged in advance**, through your Local Clinical Genetics Department or Fetal Medicine Unit
- A completed DNA request card should accompany all samples **with an appropriate telephone number and a secure fax number.**

Patient details

To facilitate accurate testing and reporting please provide patient demographic details (full name, date of birth, address and ethnic origin), details of any relevant family history and full contact details for the referring clinician

Introduction

Free fetal DNA may be detected in maternal plasma from early in gestation and used for determination of fetal gender. The sex of the fetus is determined by the presence of Y-specific sequence for a male fetus and the absence of Y specific material in the cell free DNA extract in the case of a female fetus.
The analytical sensitivity and specificity of the Real Time PCR assay was measured in 189 pregnancies (394 tests) over a period of 2 years from April 2007 to March 2009. When audited against pregnancy outcome there were 145 cases with a known outcome and in these cases the test demonstrated 100% (95% CI 97.5-99.9) concordance with no false positives or false negatives.
This is achieved by testing two separate maternal samples for the presence of SRY and by stipulating that the fetus is at least 7 weeks gestation at the time of sampling.

Referrals

All referrals should be made via a Clinical Genetics Department or Fetal Medicine Unit, please contact the laboratory in advance of sending a sample.
Samples are accepted from patients from 7 weeks gestation (confirmed by scan) at which time there should be a sufficient concentration of free fetal DNA in the circulation.
*From 7 to 9 weeks gestation, 2 x 10ml samples are required, taken one week apart. At 9+ weeks gestation 2 x 10ml samples may be taken at the same time. Samples should be sent to arrive in the laboratory within 24 hours of sampling (24-48 hours for Streck tubes) if possible. Advance notice is required so samples can be processed rapidly upon receipt. Information on the outcomes of the pregnancy will be requested as part of a national ethically approved audit. Information sheets for parents and the audit are available on our laboratory website.

Service offered

We offer this service to pregnancies at risk of X-linked disorders or congenital adrenal hyperplasia. It is not available for non-medical indications. This test may not be applicable in multiple pregnancies including those with a possible vanishing twin.
A male fetus is detected by the presence of SRY-specific sequence. The assay cannot distinguish between a lack of SRY indicative of a female fetus and a failure to extract sufficient free fetal DNA for analysis. A second sample ideally at later date but dependent on the gestation age is therefore required to repeat the analysis. Consistent absence of SRY in the presence of the control marker is taken as evidence that the fetus is female.

Technical

Maternal EDTA blood is separated as rapidly as possible after collection. Cell free DNA is then extracted from the plasma. Molecular analysis is performed using real time PCR and Taqman assays for the SRY marker and a CCR5 control marker. Results of the duplicate analysis will be released following analysis of the second sample.

Target reporting time

The results of the Y-specific probe analysis should be available within 3 days of the laboratory receiving the second sample.