Rapid invasive prenatal testing for common aneuploidies

Contact details
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Samples required
- 10-20 ml amniotic fluid
- 10-20mg chorionic villi (after cleaning)
- It is advisable to contact the laboratory prior to sending a sample.

Patient details
Referrals must be made by specialist consultants.
Please provide patient demographic details (full name, date of birth, address and ethnic origin), gestational age at the time of sampling, results of any pre-screening performed and the gestation age it was performed at, details of any relevant family history and full contact details for the referring clinician.

Introduction
Almost all prenatal samples referred to the North East Thames Regional Genetics Service will have rapid invasive prenatal testing in order to test for the commonest aneuploidy conditions (i.e. Down syndrome, Edward’s syndrome, Patau syndrome and Turner syndrome).

A small aliquot of the prenatal sample will be forwarded to Viapath, Genetics Department, 5th Floor Tower Wing, Guy’s Hospital, London, SE1 9RT to be tested by QF-PCR. They will issue QF-PCR results directly to the referring hospital.

Referrals
- Pregnant women with a positive prior screening risk but no scan abnormalities N.B. Women with scan abnormalities will be offered rapid prenatal testing, but will also be offered microarray testing
- Pregnant women with a known family history of a genetic/metabolic/biochemical condition referred by a specialist consultant

Service offered
Forwarding of an aliquot of the prenatal sample to the pan-London testing laboratory at Viapath, Genetics Department, 5th Floor Tower Wing, Guy’s Hospital, London, SE1 9RT.

Based on the reason for referral and the outcome of the QF-PCR test, the following workstreams are applied:
- Normal QF-PCR result and no scan anomalies – no further testing is performed.
- Normal QF-PCR result and ultrasound scan anomalies – microarray testing will be performed by the North East Thames Regional Genetics Service.
- Abnormal QF-PCR results with or without abnormal ultrasound scan anomalies – the QF-PCR result will be confirmed by karyotyping performed by the North East Thames Regional Genetics Service.

Technical
Rapid prenatal testing will be performed using Guy’s in-house QF-PCR test for chromosomes 13, 18 and 21, and where appropriate for the sex chromosomes, X and Y.

Target reporting time
Within 3 days from sample receipt.