



## Guidelines for the Genetic Analysis of Solid Tissue Samples

### Available Genomic Tests

- 1) Microarray (SNP based microarray platform) - this is the principal test and will detect whole chromosome gains or losses, segmental imbalances throughout the genome to a high resolution, and other genomic conditions observed in pregnancy loss referrals (e.g. triploidy, molar pregnancies, uniparental isodisomy, degrees of maternal cell contamination).
- 2) QF-PCR - this is an alternative targeted test employed by the laboratory for the specific diagnosis of trisomy 21, 18 or 13, when such a diagnosis is highly likely.
- 3) Cell culture - viable cells can be propagated in culture to facilitate biochemical assays or long-term storage.

### Referral Eligibility (Pregnancy Loss Referrals)

- R22 - Fetus with a likely chromosomal abnormality.
- x R412/R27 - Fetal anomalies with a likely genetic cause (non-urgent).

Please note that we do not process:

- x Confirmation of an abnormal cytogenetic prenatal diagnosis.
- x First or second trimester unexplained 1<sup>st</sup> or 2<sup>nd</sup> miscarriages with no fetal malformations.

\* Please refer to the National Genomic Test Directory for rare and inherited disease [www.england.nhs.uk/publication/national-genomic-test-directories/](http://www.england.nhs.uk/publication/national-genomic-test-directories/) for full details of eligibility criteria, indication definitions, requesting specialities and testing methods.

### Summary of Test Provision

#### DNA extraction / storage

- x All processed samples will have DNA extracted and stored (this will include R412 indications which may be eligible for WGS (R27), WES or Large Panel testing in the future, following multidisciplinary review and clinical geneticist approval).
- x Samples received following sudden unexplained infant death, including coroner cases, will have DNA extracted and stored but will not be processed for testing unless testing has been specifically requested or a likely genetic cause indicated.

#### Microarray

- x 3<sup>rd</sup>

## Instructions for Sending Samples

### I. General requirements

- x Please **do not** place samples in fixative.
- x Please forward as soon as possible following collection, but if there is delay, store at 4°C. Do not freeze, expose to excess heat, or store in fixative.
- x Please ensure that packaging conforms to HSE packing Instructions P650 (see [www.hse.gov.uk/cdg/pdf/infect-subs.pdf](http://www.hse.gov.uk/cdg/pdf/infect-subs.pdf)).
- x Tests are principally DNA based.
- x For cell culture viable cells are required. Suspend samples in tissue culture media\* or sterile isotonic saline and send without delay.

### II. Pregnancy loss referrals

For ERPC (Evacuation of Retained Products of Conception) samples, separate the solid tissues and transfer to a dry, sterile, leak proof container. Please **do not** send in the procedure evacuation container.

POC	Solid tissues only	In dry, sterile, leakproof container
Placental biopsy (chorionic villi)	At least 2cm <sup>3</sup> taken adjacent to the cord insertion site to ensure it is the fetal side	In dry, sterile, leakproof container

:5\*/ :HEVLWH 6ROLG 7LVVXH 6HUULFH \*XLGH 9HUVLRQ -QGH[ :5\*/ '2& 3ULQWHG 2FW

