

Genomic Diagnostics Laboratory (GDL)

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REFERRAL CRITERIA, REQUIREMENTS FOR CYTOGENETIC ANALYSIS, AND TISSUE DISPOSAL

POLICY FOR SOLID TISSUE SAMPLES FOLLOWING LOSS OR TERMINATION OF PREGNANCY

Referral Criteria

Samples are accepted for testing from the following referral categories:

- Structural fetal abnormality; please clearly describe the abnormalities on the referral form. Please specify any suspected syndromes.
- A third or subsequent consecutive miscarriage. Please include the dates of each miscarriage on the referral form.
- Samples requested by the GDL for follow up investigations. Please note that routine confirmation of prenatal results are no longer performed by this laboratory.
- Neonatal death or IUD when the gestational age is 24 weeks or greater (TESTED BY QF-PCR ONLY).
- Samples for culture prior to export for further testing.

Cytogenetic testing will not be carried out on samples referred for isolated neural tube defects without additional malformations or recurrences in the family.

If you require further information please contact the laboratory directly by telephone and ask to speak to the Duty Scientist.

Sample Requirements

Fresh tissue samples are required (less than seven days old for post mortem samples). The most appropriate sample types are:

- A full depth skin sample (0.5cm³)
- A sample of umbilical cord tissue approximately 2-3 cm in length
- A sample of placental tissue approximately 1-2 cm³ in size which includes placental villi.

If possible, please send a sample of cardiac or cord blood in EDTA (0.5ml collected in a paediatric tube).

Please Note

- **Whole Fetuses cannot be accepted by the laboratory under any circumstances.** The referring centre will be contacted to arrange collection.
- Tissue samples that are more than 7 days post mortem or appear necrotic on arrival in the laboratory may not be suitable for testing. Please state the date of delivery on the referral form.
- Samples that have been preserved in formalin are unsuitable for cytogenetic studies. Samples that have been frozen are unsuitable for cell culture but are suitable for molecular cytogenetic analysis.
- **Please do not** include cord clips, dressings or any other medical device with the sample.

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- **Please do not** send the entire products of conception from an early pregnancy loss. Please examine the tissue at delivery and only send sufficient material for testing (approx 1cm³).
- Products of conception **will only be accepted from clinically confirmed pregnancies** (i.e. those that have been confirmed by both high levels of [hCG](#) and ultrasound confirmation of a gestational sac or fetal heartbeat). This information **must** be provided on the referral form. Samples that do not meet these criteria cannot be processed.

Transport Requirements

- a. Solid Tissue samples should be sent either dry or in sterile saline (if stored overnight) in a sterile **leak proof** container.
- b. All samples **MUST** be accompanied by a fully completed Genetics Test Request form, on which the consent statement has been signed by the referring clinician (copies can be downloaded from the Mangen website www.mangen.co.uk). Please do not send paperwork for other departments with samples for genetic analysis.
- c. The sample container and referral form should be sealed separately into a biohazard bag. **All packaging should conform to UN3373 standards.**

Tissue Disposal Policy

Please **ensure that the sample size does not exceed that detailed in the sample requirements section** above; in this event, all material will be exhausted by testing.

This laboratory does not have facilities for the handling or disposal of excess fetal material. In the event of samples containing excess fetal material being sent to the laboratory, the referring centre will be contacted to arrange the return or collection of the sample.

In cases where the sample has not been collected within fourteen days of contact, and the wishes of the mother for disposal of the tissue have not been communicated to the laboratory, fetal material of less than ten weeks gestation will be cremated locally as part of a monthly communal cremation. Crematorium fees (currently approximately £65) will be invoiced to the referring centre.

Where there are special requirements for the handling or disposal of samples, please clearly specify the details on the referral form and provide any relevant documentation.

Essential Referral Information

All samples for genetic analysis must include the following information in order to be processed:

- Referring Clinician and Referral Centre clearly indicated.
- Full patient details including Maternal NHS number and Maternal Date of Birth.
- Full clinical details including the reason for request (please detail abnormalities), gestational age, date of delivery and date of sampling.
- The site of origin of the tissue sample sent.

OTHER TISSUE SAMPLES REFERRALS

- If a skin biopsy from a living patient requires testing for tissue specific mosaicism, or tissue requires cell culture prior to export for molecular or biochemical testing, please telephone the laboratory to request fresh transport medium. Please also comply with transport requirements b. and c. above.
- Solid tumour samples for cytogenetic testing should be sent to the Oncology Cytogenetics Laboratory at the CHRISTIE NHS FOUNDATION TRUST (0161 446 3165).

PLEASE CONTACT THE LABORATORY TO DISCUSS ANY ASPECT OF SAMPLING OR REFERRAL IF REQUIRED

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