Indications for cytogenetic testing after pregnancy loss

Cytogenetic investigation of pregnancy loss is now routinely performed by a combination of QF-PCR and MLPA (multiplex ligation-dependent probe amplification) in this laboratory. This combination of techniques has a higher success rate than karyotyping.

Karyotyping will still be performed where this can provide relevant information that investigation by QF-PCR/MLPA may not provide e.g. in the investigation of pregnancy loss from known balanced translocation carriers.

Parental follow-up will be required in a proportion of cases either to exclude a familial rearrangement, a Robertsonian translocation or a possible polymorphic variant.

Fetal/placental tissue

- Any fetus, stillbirth or neonatal death with congenital abnormality suggestive of a chromosome anomaly or with neural tube defect or with IUGR
- Unexplained stillbirth or neonatal death (>24 weeks)
- Unexplained IUD/spontaneous abortion (>16 weeks)

Please note: Cytogenetic investigations are not normally indicated for spontaneous miscarriages at less than 16 weeks gestation (in line with Commissioners’ guidelines). A high proportion of these will be aneuploid (either 45, X or single or double trisomies) and although this provides an explanation for the miscarriage it is unlikely to be of significance for future pregnancies.

If a fetus falls outside these referral categories, please contact Lee Grimsley or Lucy Platts, Cytogenetic Team Leaders, to discuss the test required before sending a sample.

Karyotyping parents

- Where molecular cytogenetic techniques indicate that parental karyotyping follow-up is warranted (see above).
- After pregnancy loss of a fetus with multiple congenital abnormalities or severe IUGR, where tissue is unavailable from the fetus or placenta for molecular cytogenetic testing both parents should be karyotyped instead to exclude a balanced rearrangement.

Please note that parental karyotyping after three or more unexplained miscarriages is no longer funded by the London Specialist Genetics Commissioners. Please see ‘Indications for cytogenetic testing after pregnancy loss’ for further information on appropriate tests.

If the parents fall outside these referral categories, please contact Lee Grimsley or Lucy Platts, Cytogenetic Team Leaders, to discuss the test required before sending a sample.

Sample types and Requirements

Pregnancy loss

- Amniotic fluid from planned terminations: 10-20 mls
- Products of conception: (including chorionic villi or fetal tissues)
- Placenta: 1cm³ piece including chorionic villi or membrane (preferably close to the cord insertion site).
- Cord : 1cm length taken from placental insertion site and including 1cm³ piece of placenta
Post Mortem

- **Internal tissues**: 1cm³ muscle, cartilage, lung or liver
- **Skin**: 1cm² full thickness including the dermis

Requirements

All samples **must** be put into a clean, sterile, leak proof container and transported to the laboratory on the day that they are taken.

If samples are likely to be delayed, then they **must** be placed into a sterile saline solution, refrigerated and transported early the next working day.

Please note that any **delay** in receipt can affect the success rate of the samples and can also compromise the quality of preparations obtained.

Please ensure that if a fetus and/or placenta or fetal and/or placental tissue is required to be returned for burial that this is clearly stated on the referral form provided (downloadable from this website and that the appropriate paperwork is completed.

**Parental samples (when requested)**

5ml of blood in a plastic lithium heparin tube and 5ml in a plastic EDTA tube from each parent.