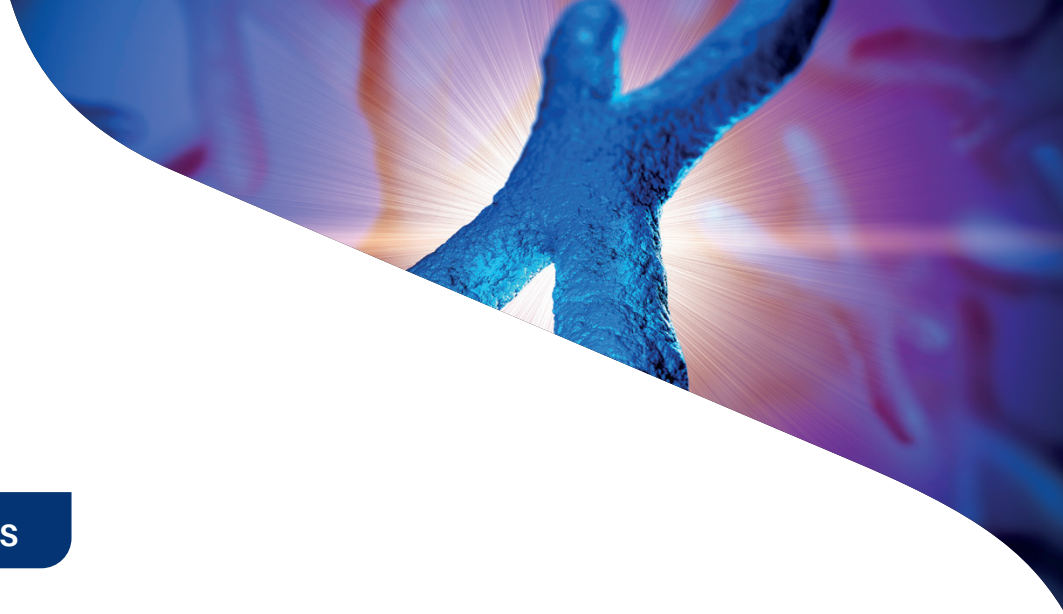

TDL GENETICS

Information for Patients

Products of conception for genetic investigations

Chromosome analysis by cytogenetic karyotype

Historically, karyotyping has been the primary method for determining chromosome status of miscarriage tissue or products of conception (POC) samples. Karyotyping analyses all 23 pairs of chromosomes to detect aneuploidy (missing or extra chromosomes), ploidy (extra sets of chromosomes), large deletions or duplications of chromosome material, chromosome rearrangements, and chromosome mosaicism (varying chromosome complements in different cells).

In around 50% of first trimester pregnancy losses a chromosome aberration is detected. In many cases the aberration has arisen as a sporadic event. In a very small percentage of cases the aberration may have been inherited from a chromosome rearrangement carried by a parent, in such instances karyotyping studies of both parents will be suggested.

Limitations of traditional cytogenetic karyotyping

Cytogenetic analysis or karyotyping first requires cell culture to be performed. POC cells are grown or cultured in the laboratory and stopped at the point in their division when the chromosomes become visible under a light microscope. This process typically takes 4-5 weeks to get a result. Cells that are degenerating, dying, or already dead are not suitable for this type of chromosome analysis since they often do not grow in the laboratory, therefore, some pregnancy tissue can sometimes be unsuitable for traditional karyotype analysis.

Additionally, with traditional karyotyping there is a risk of no results due to bacterial or fungal contamination of the sample and a chance of maternal cell contamination (MCC). If a normal female result is returned (reported as 46,XX), this may be the correct result for the fetus or it may be maternal cell contamination (MCC), where the maternal cells present in the pregnancy tissue were tested, not the fetal cells. At TDL Genetics the laboratory team takes great care to reduce the occurrence of maternal cell contamination (MCC) by sorting, dissecting and cleaning each POC sample when it arrives in the lab. Though the risk of maternal cell contamination can never be completely eliminated, sorting techniques to identify placental and fetal tissue combat much of the potential for contamination. It is important to keep in mind that even with microscopic tissue sorting there is still a chance that results will be contaminated.

Where no fetal or placental material has been recognised during dissection, the tissue is likely maternal in origin and not representative of the conceptus (an uninformative result). A report relaying this information is sent to the referring clinician and a small amount of tissue prepared and stored for 4 weeks. Testing of this tissue by molecular methods can be requested within this time; however, there is a high probability that any result will represent the maternal profile.

Chromosome analysis by SNP Array CGH

SNP Array testing can detect chromosome losses and gains across all 23 pairs of chromosomes. It can detect extra or missing whole chromosomes, but also smaller changes than is possible by conventional karyotyping. SNP array can also detect triploidy and some types of mosaicism.

As this assay does not require the cells to be cultured, those samples that may fail due to fungal or bacterial contamination can still be tested for chromosomal imbalances. Please note that this assay does not detect balanced rearrangements and most tetraploids.

Where chromosome analysis of a POC sample has been requested but fails to provide a result, we will reflex to SNP array CGH in its place. This enables us to provide a result for the significant majority of POC cases. Unfortunately, there will always be a small proportion of cases in which the above testing fails to yield a result (this is usually due to poor quality samples).

Sample Collection

Where samples are collected in a theatre environment, a small sample of placental or fetal material should be transported to the lab in a sterile container with suitable transport medium (transport medium is available on request from the lab), where transport medium is not available, a dry sterile container with or without normal saline can be used.

Where samples are collected at home, sterilise a leak proof container using either boiling water or steam. Once sterile the sample should be collected and sealed in the container, this should be stored in the fridge (NOT the freezer) until sending is practicable.

In order to collect the tissue, it may feel more comfortable to be sitting on the toilet. A sieve can be sterilised by running boiling water through it and placing in the toilet bowl. The retained tissue can be transferred from the sieve into the sterilised container.

Sample Dispatch to the Laboratory

Label the sample container with your name, DOB and a reference number (if available). When sending make sure that as well as being in a leak proof container, that the container is wrapped in absorbent material (household kitchen towel – enough to absorb any liquid should the sample leak) and that this is in turn sealed in another leak proof container preferably rigid to protect the sample in transit. The sample should be sent to the lab as soon as possible with a clinical referral form from your doctor. It should be clearly stated on the form the testing required (chromosome analysis in POC) and any relevant clinical details that apply, including if there is suspicion of a molar or partial molar pregnancy. It is important that the details of the doctor are clear for reporting as results cannot be sent directly to patients.

Sample Disposal

Once a small amount of tissue has been removed for testing, the remaining sample is disposed of.

If you would like the remaining tissue returned for burial or cremation, please ensure this is communicated on the referral form or that the laboratory is informed prior to receiving the sample.

Arrangements can be made to collect the sample from Patient Reception (details below). All samples with no special request will remain in storage for 4 weeks and then disposed of by sensitive incineration, the lab will keep a record of the collection and location of the incineration.

For further advice please contact TDL Genetics.

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Postal address for samples

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WC1H 9AX

Samples delivered in person

Patient Reception
76 Wimpole Street
London
W1G 9RT

Tel: 020 7307 7383

Opening hours: Monday to Friday
7.00am – 7.00pm, Saturday 7.00am – 1.00pm

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