

GENETIC TESTING: SPECIMEN REQUIREMENTS, COLLECTION AND DISPATCH



Registration No. 2000/026390/08

General Instructions:

- **Kits are available on request.** These include the relevant collection tubes, request form and envelope for despatch of the sample to the laboratory.
- Use **sterile technique**, close containers tightly.
- **Label all containers** with the patients name **and** another unique identifier e.g. hospital number or Date of birth/ID number. **NOTE** unlabelled tubes are considered non-conforming and cannot be processed.
- Living cells are needed for karyotyping. **DO NOT FREEZE, CENTRIFUGE OR ADD FIXATIVE TO ANY SAMPLE.**
- **Complete the request form in full**, including as much clinical information as possible as this may impact on the test report. **NOTE;** due to the invasive nature of the amniocentesis, please ensure that the patient completes the informed consent section of the amniotic fluid request form.
- Specimens must be sent to the nearest blood bank for dispatch to the laboratory and should be received in the Cytogenetics Laboratory **within 24 hour of collection.** If there is a delay in despatch keep sample at 2-6 C.

Note: Limitations of tests:

Karyotyping: detects numerical and structural abnormalities of ALL chromosomes. Result issued in 15-20 working days

QF-PCR: detects aneuploidies (numerical abnormalities) of chromosomes 13, 18, 21, X and Y only and NO structural abnormalities. Result issued in 48-72 hours.

Sample Type	Tubes	Collection	Storage and Dispatch	TAT	Technical reasons for no result on specimen.
Amniotic Fluid , for karyotyping, AFP and QF-PCR (an aliquot will be removed in the lab)	2 x 15ml sterile tubes, no anticoagulant	10- 15 ml fluid in each tube. Discard the first 1-2ml before placing in the tubes	Send sample within 24 hours of collection at room temp. If delayed, keep samples at 2-6°C	Karyotyping: 15 working days. QF-PCR : 48 – 72 hours	Maternal cell contamination (MCC) confounds the result. Bloody samples may impede growth of amniocytes in culture resulting in no results.
Peripheral and Cord blood , for karyotyping	Adult: 1 x 5ml Green top vacutainer tubes (Lithium heparin) Paediatric: 1 x Paediatric tube	Babies under 1 month : full paediatric tube Children > 2 years of age to 10 years: 1 – 4 ml Adults: 4 ml NB: invert the tube to mix well	Send sample within 24 hours of collection at room temperature. If there is a delay in despatch keep sample at 2-6°C	Karyotyping: 20 working days.	No growth and low mitotic index resulting in no result may occur when samples are clotted/ haemolysed or insufficient or there is a delay in receipt in the laboratory. Kleihauer test required on cord blood to determine whether foetal or maternal in origin.
Peripheral and Cord blood for QF-PCR	In addition to the above; 1 x 5ml purple top (EDTA) OR 1 x 5ml green top (Heparin) tube	Infants and young children (<2 years of age; 2 ml Children > 2 years and adults: 2-4 ml.	Send sample within 24 hours of collection at room temp. If despatch delayed keep sample at 2-6°C	QF-PCR : 48 – 72 hours	MCC and bloody samples as above for amniotic fluid. Cord blood – as above.
Products of conception and skin	Place in sterile container and add enough sterile saline to cover the tissue. Alternatively place in sterile container with culture media – request these from the lab.	Aseptically collect tissue according to your protocol. For Fetal demise cases DO NOT send the entire foetus.	Send sample within 24 hours of collection at room temperature. Refrigerate after collection until time of dispatch to the lab.	Karyotyping: 15 - 20 working days.	Tissue received in formalin will not grow in culture. Bacterial/fungal contamination of cultures due to collection not being done aseptically will result in no growth. If fetal demise occurred more than 48-72 hours prior to retrieval of tissue and receipt in the lab it is unlikely to grow.

Note: Laboratory Hours are Monday to Friday, 08h00 to 16h00

Contact Details:

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Revision Summary

VERSION NUMBER	REVISION DETAILS
0	New document.