

# Genetic Medicine Referrals

## Sample Types

### Cellbank Samples

#### Blood samples

Use AcidCitrateDextrose-A or sodium heparin tube only. We require 10-20ml for adults, 2-10ml for children.

#### Other samples

By prior arrangement only. Please contact the cellbank for more details. (Contact details available on the website).

### Chromosome (Cytogenetic) Analysis

#### Prenatal samples

Do not freeze or expose to heat. Must arrive in lab within 24 hours.

**Amniotic fluid** - Send 10-20ml in a sterile, leak-proof, plain (no anticoagulant) plastic container. Samples which are blood-stained or discoloured cannot be tested using rapid aneuploidy screening techniques due to the risk of false negative results caused by maternal cell contamination.

**Chorionic villi** - The minimum size requirements for the specimen are dependent on the reason for referral, usually 10-30mg in a sterile transport medium; contact the cytogenetics lab for more details. CVS samples should be received on the same day as taken and preferably no later than 4.00pm. For samples referred for a parental balanced rearrangement (ie, inversion, translocation) a maternal blood sample (5ml in EDTA) is required.

**Fetal blood** - Send 1ml in a 2ml paediatric lithium heparin tube and mix well to avoid clotting. Please clearly mark the sample as "fetal blood".

#### Postnatal samples

Do not freeze or expose to heat. Can be stored overnight at 4°C if required.

**Venous blood** - Use lithium heparin tube only. We require at least 5ml for adults and children, at least 1ml for neonates. Send within 48 hrs of venepuncture and mix well to avoid clotting.

Chromosome breakage tests are initiated on a Monday, Tuesday or Friday morning ONLY. Please take this into consideration when sending blood samples to the laboratory for testing. Testing for Fanconi anaemia or ataxia-telangiectasia requires a minimum of 2ml blood in lithium heparin (1ml for neonates). Testing for Nijmegen breakage syndrome requires a minimum of 3ml blood in lithium heparin.

**Solid tissue** - Do not expose to formalin. Send in dry, sterile, plastic container, or sterile saline if stored overnight. The container should be wrapped with absorbent material and placed in a sealable polythene sample bag. Sending multiple sample types improves success rates. The most appropriate tissue types to send us are:

- A full depth skin sample (0.5cm<sup>3</sup>).
- A sample of cord (2-3cm in length; please do not include a cord clip) along with a sample of placental membrane from around the cord insertion site.
- A cord or cardiac blood sample (at least 0.5ml collected into a 1-2ml lithium heparin paediatric tube).

## Gene (DNA) Analysis

### **Blood samples**

Use EDTA tube only. We require at least 5ml for adults and children, at least 1ml for neonates. Store at 4°C and send by first-class post.

### **Mouthwash samples**

Use Oragene container only; these are available from the molecular genetics lab.

### **Other samples**

By prior arrangement only. Please contact the molecular genetics lab for more details. (Contact details available on the website).

## Enzyme (Biochemical) Analysis

Due to the wide range of tests carried out for biochemical genetic indications, it is not possible to list the full sample requirements. Please see the relevant service profile for detailed sample requirement. (Service profiles are available on the website). However, for guidance, the following sample types are used:

- Blood - use EDTA or lithium heparin tube, depending on test.
- Plasma
- Urine
- Liver
- Amniotic fluid
- Chorionic villi
- Cerebrospinal fluid
- Cultured skin fibroblasts
- Cultured amniotic fluid cells
- Cultured chorionic villi