

Sample requirements

Samples must be labelled with:

- the patient's full name
- the patient's date of birth
- NHS or genetics number
- date and time sample was collected

The m.3243A>G mutation can be detected in virtually all tissues; however heteroplasmy differs between sample types. Blood leukocytes generally contain the lowest level of the m.3243A>G mutation. Therefore it is possible to obtain a negative result for m.3243A>G in blood, despite the presence of the mutation in other tissues. Of the readily accessible tissues available for testing, urine epithelial cells contain the highest level of the m.3243A>G mutation and **urine is therefore the preferred sample to test** ([de Laat *et al* 2012 J Inherit Metab Dis 35: 1059-1069](#))

Urine: Collect the first 20ml of an early morning urine sample into a sterile universal container. Send to the laboratory with a completed request form by first class post on the day the sample is taken

Blood: Please send at least two 4ml EDTA blood samples (1ml minimum for neonates, 5-10ml for children and 10-20ml for adults). **Samples should be transported at ambient temperature.** Blood samples in glass bottles are not accepted by the laboratory. Blood samples should be received by our laboratory **within 5 days of venesection.** Do not freeze blood samples – if storage is required prior to dispatch, blood samples can be stored at 4°C (39.2°F).

DNA: Please send a minimum of 5µg of DNA. **DNA samples can be sent at room temperature.**

Other: In special circumstances, a **saliva sample** is acceptable. Please contact the laboratory for sample collection kits and/or instructions for collection ([Oragene](#)). Please contact the laboratory prior to sending any other samples (e.g. paraffin-embedded tissue sections).

High Risk samples:

It should be noted that blood samples from patients who are likely to be Hepatitis B antigen or HIV positive, who have infectious hepatitis or who are jaundiced without obvious cause are potentially dangerous to all who handle them. Blood from febrile, undiagnosed patients, especially from abroad, may also be dangerous. Great care should be observed when submitting these samples for laboratory investigations, with strict adherence to the recognised methods of handling, particularly:

1. Forms and sample bottles must be clearly marked with a warning sticker
2. The samples must be sealed within two plastic bags.
3. The accompanying form must not come into contact with the sample.

Request form

Completing Request Forms and sample acceptance criteria

Samples must be accompanied by a correctly completed request form. All request forms must indicate either a specific disorder/gene(s) to be investigated or, a request to extract and store DNA

The request form must be completed with the following patient identifiers for the sample to be accepted:

- Surname
- Forename (in full)
- Date of birth
- NHS number, Hospital number
- Genetics number (if applicable)

Samples will be rejected if there are fewer than 3 unique points of identification on the request form. The sample must be the correct sample type to be accepted and processed. The sample must not have been used on any Chemistry of Haematology department analysers.

Clinical context is often crucial for determining the pathogenicity of genetic variants. Therefore, please provide as much clinical information as possible about the case on the request form. This will decrease the likelihood the laboratory will need to contact a clinician for additional information to produce the clinical report.

Consent

In submitting a sample with a request form, the clinician confirms that informed consent has been obtained for (a) testing and storage (indefinitely) (b) the use of this sample and the information generated from it to be shared with members of the donor's family and their health professionals (if appropriate). The patient should be advised that the samples may be used anonymously for quality assurance and training purposes.

Postage

Please send samples by first class post or courier. Packaging should comply with UN3373 regulations for packaging and transportation of samples (See Table A4 in ['Biological agents: managing the risks in laboratories and healthcare premises'](#)):

1. The sample should be wrapped in enough tissue to absorb the entire contents of the tube in the event of a breakage.
2. Seal the tissue with tape and place it into a specimen bag and seal.
3. Samples should then be placed in a sample box or padded envelope along with a copy of the referral information and the package marked 'Pathological Specimen – Fragile With Care'.



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