



CRIM status can be determined in 2 ways:

1. Prediction based on GAA mutations by means of this database:
www.pompevariantdatabase.nl
2. Determine the presence or absence of endogenous GAA through Western Blot (see instructions on next page)



CRIM testing request



Please note: It is essential to record the sample collection date/time for accurate result interpretation and contact information for the attending physician to communicate the laboratory report.



Patient Details:

Family name:

First name:

Date of birth (DD/MM/YY):

Sex (please circle): Male - Female

Relevant clinical details:

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.....
.....

Relevant family history:

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.....
.....

Sample date (DD/MM/YY): Sample time:



Hospital Details:

Requesting physician:

Hospital name:

Address line 1:

Address line 2:

Address line 3:

City/Town:

Postal/ZIP Code: Country:

Contact phone number:

Fax number (for results):

E-mail:



Sample collection procedure for blood CRIM

Blood collection tube requirement:

BD Vacutainer® CPT™ (Cell Preparation Tube) with Sodium Citrate, 4 ml (Becton Dickinson), stored at room temperature (18-25°C) and protected from light. If your hospital does not carry CPT tubes, please contact us and we can provide the tubes. Please note that the cost of a courier for shipping is 35 GBP. Contact Katie Harvey to arrange a shipment and/or for additional information.

Important notes:



1. Samples must be collected **before** enzyme replacement therapy initiation. If samples are collected after treatment has commenced, we are **not** able to reliably determine CRIM status.
2. CPT tubes must be centrifuged before shipping to prevent sample degradation. If degradation is observed, we are not able to reliably determine CRIM status.
3. The CPT tubes must be labeled with the patient's details (name and date of birth at a minimum).
4. You must contact the laboratory prior to collecting and sending a sample to ensure the sample can be processed immediately upon arrival.



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Collection procedures:

1. Blood collection:

Collect blood into the tube using the standard technique for BD Vacutainer® Evacuated Blood Collection Tubes. **Invert the tube 8 to 10 times to mix the anticoagulant additive with blood after draw.** Do not shake.

2. MNC (Mononuclear cell) isolation by centrifugation:

a. After collection, store tube upright at room temperature until centrifugation. Blood samples should be centrifuged within four hours of collection.

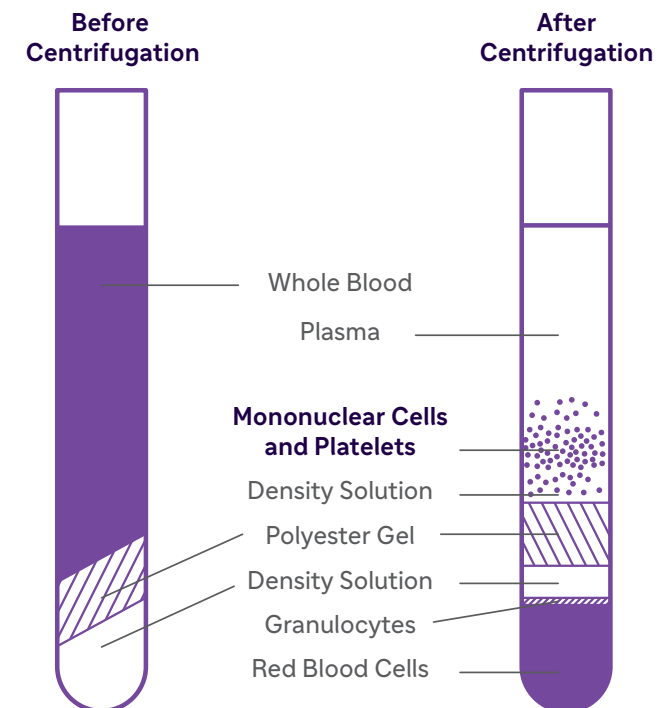


b. Re-mix the blood sample immediately prior to centrifugation by gently inverting the tube 8 to 10 times. Do not shake.

c. Centrifuge the tube/blood sample at room temperature (18-25°C) in a horizontal rotor (swing-out head) for 30 minutes at 1800 x g.

d. After centrifugation, mononuclear cells and platelets will be in a whitish layer just under the plasma layer (Figure below).

e. Resuspend the cells into the plasma by inverting the unopened BD Vacutainer® CPT™ Tube gently 5 to 10 times. Store the tubes at 2-8°C for shipping.





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3. Shipping:

- a. The tubes **must be kept cold** (at 2-8°C) after processing and shipped with cold pads. **Samples cannot be frozen at any point.**
- b. **CRIM request form** should be completed. It is essential to record the sample collection draw date/time for accurate result interpretation and contact information for the requesting physician to communicate the laboratory report.
- c. The blood sample(s) must reach the laboratory **within 48 hours** of draw. Please enclose the CRIM request form with the samples and **notify the lab before shipping.**
- d. **Address to:**

Great Ormond Street Hospital for Children NHS Foundation Trust

Dpt: Enzyme Unit - Chemical Pathology - Camelia Botnar Laboratories

Address: London WC1N3JH United Kingdom

Contacts: Katie Harvey, Katie.Harvey@gosh.nhs.uk, Ext 7843

Derek Burke, Derek.Burke@gosh.nhs.uk, Ext 5290

Emmaline Cullen, Emmaline.Cullen@gosh.nhs.uk

Tel: +44 0207 405 9200 Ext 1785 or 6751

Fax: +44 0207 829 8624