Oncology - Sample Requirements

Sample Types

- **Bone Marrow (BM)** is the tissue of choice to investigate patients suspected of having leukaemia or related haematological neoplasms. BM aspirates are routinely received although a BM trephine crush is an option if the marrow is fibrotic or otherwise difficult to aspirate.

    - **Conventional cytogenetics/FISH**: **1-2mls BM** in transport medium or in lithium heparin (please try to avoid excess haemodilution)
        - For paediatric acute leukaemias also add three drops of BM to KCH solution, when available; this will then be fixed at the referring laboratory
        - For **FISH** analysis, we can also accept unstained BM smears or trephine rolls; these may be useful for many months after being made. Please note that FISH results on trephine biopsies are difficult to obtain; however, we will attempt FISH investigations if required.

    - **Molecular investigations**: **2-3mls BM** is the preferred sample for *KIT* investigations for systemic mastocytosis, together with *FLT3 , NPM1* and myeloid NGS panel investigations, otherwise peripheral blood in EDTA (see below)

- **Peripheral blood (PB)**: this is perfectly adequate for FISH studies in lymphoproliferative disorders if there is PB lymphocytosis and for most molecular studies. PB can be used for conventional cytogenetics provided that immature cells capable of division are present (i.e. frequently in acute leukaemias and diagnostic CML, but rarely in MDS or MPD). If the BM is a dry tap or you are doubtful about its quality, consider sending a blood sample as well.

    - **Conventional cytogenetics/FISH**: at least **5mls** in lithium heparin; if fresh material is not available, FISH can be performed on unfixed, unstained blood smears (at least 4 slides);

    - **Molecular studies:** *JAK2, CALR* and *MPL* (MPN gene panel), *TP53*, *KIT* (for AML) at least **5mls** in EDTA (**10~20mls** for *FIP1L1-PDGFRA* testing)

- **Unfixed air-dried tissue imprints or 2-4µm thick paraffin embedded tissue sections** (mounted on positively charged slides) are used for interphase FISH analysis for NHL and solid tumours. Both sample preparations are acceptable for rearrangement testing, while imprints are the only suitable tissue for numerical abnormalities. Samples should only be forwarded to the laboratory once the pathologist has determined the need for a specific genetic result. Imprints should be stored at room temperature prior to sending and they are likely to be successful even when several weeks old. For those samples, where there is limited infiltration of the tissue, the involved areas must be clearly marked on an additional H&E slide. In the context of B cell NHL, fresh biopsy material for cell culture will only be accepted in specific cases after discussion with laboratory personnel. **Please note that slides should have two patients' identifiers, including the patient's name and the histology ID number**.

- **Fresh biopsy samples** for paediatric solid tumours; these should be placed in sterile culture medium and sent to the laboratory as quickly as possible. Where material is extremely limited and a FISH marker could be used, touch preparations of biopsy specimens may be adequate. These should be placed on slides in a suitable way for the FISH to be carried out under 22x22mm coverslips. For tumours with classic

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chromosomal rearrangements that help in diagnosis or prognosis, we suggest that imprints are always made in the referring laboratory and sent with the fresh tissue.

- **Other fresh tissues** like lymph nodes, spleen, ascitic fluid, CSF, buccal swabs and non-paediatric solid tumours can also be analysed following discussion with the laboratory.

Specimen Containers

For samples which require **cytogenetic analysis**, the laboratory will provide containers to regular referrers for sample collection (transport medium and KCH with full instructions are sent out by the WRGL on the FIRST Tuesday of every month). These bottles contain heparinised tissue culture medium with antibiotics, to facilitate the transport of the small amount of BM and avoid desiccation. More bottles can be sent upon request, at any time, by hospital transport or by post. However, please note that i**n emergency, a tube containing lithium heparin (green top) can be used** for both BM and PB.

Fresh solid tissues should be placed in one of our transport containers or other a sterile liquid such as culture medium, Hank's balanced salt solution or saline. The department is pleased to advise on the use of alternative specimen containers.

Identification of the Tube

Label the tube with surname and first name, date of birth as well as date of withdrawal.

Completing the Order Form

Please send a COMPLETED referral form with all samples. The reason for referral is essential to determine which culture types need to be set up, which tests to perform, numbers of cells to analyse and sample prioritisation. All relevant clinical and haematological information and likely diagnosis should be included. If the patient is a participant of a research trial, it is important to give details as certain trials can have specific requirements. For samples from patients who underwent BM transplant, the sex of the donor should be stated.

Transport and Mailing

Sample and referral card should be sent **together** in a secure leakproof package (hard cardboard box not a padded envelope) according to UN P650 packaging instructions, to arrive as soon as possible after collection. Please note that the laboratory no longer provides an emergency weekend rota; however, for **urgent** referrals (?APML, ?Burkitt, paediatric acute leukaemia), samples can still be sent during the weekend. For these cases, arrange special transport and phone the lab on Friday afternoon or the Hospital switch board during the weekend (the hospital switchboard will contact WRGL staff). **It is advisable to telephone about any samples that could arrive at the laboratory late in the day or out of hours.**

All samples should be sent directly to the Wessex Regional Genetics Laboratory by guaranteed next day delivery, courier service, hospital transport, etc. While samples from the majority of chronic myeloid conditions will survive comfortably for 48 hours, ALL and Burkitt lymphoma samples die very rapidly, so they should reach the laboratory within a few hours of being taken, and myeloma samples cannot be processed effectively if they are more than 24 hours old. It is advisable that all myeloma samples sent on Friday arrive before 2pm to allow time for cell separation; all myeloma samples arriving on Friday after this time will be refrigerated over the weekend and processed on Monday morning, in such cases the success of the investigation is likely to be compromised. Samples for RNA analysis should ideally reach the laboratory within 24 hours of being taken as degradation of RNA may compromise the assay.

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