

(WRGL use only):

Investigation(s):

W

DNA loc:

In before?

Initials

Referral reason:

Date of receipt:

WESSEX REGIONAL GENETICS LABORATORY

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BLOOD SAMPLES FOR CHROMOSOME AND DNA ANALYSIS

PATIENT DETAILS

Addressograph label

SURNAME	DATE OF BIRTH _ _ : _ _ : _ _ _ _	SEX	Referring consultant
FORENAME	NHS NUMBER		Hospital / Department
Postcode	Hospital number / Genetics number		Clinician's contact number
NHS / Private (address for invoicing):	Additional copies to		Clinician's NHS.net email @nhs.net

Date of collection _ _ : _ _ : _ _ Collected by _____ State reason for urgency below
Priority: Routine Urgent

REASON FOR REFERRAL Provide full clinical details including any relevant family history.
For NHS referrals in England, please enter the National Genomics Test Directory clinical indication below . Please note that any tests not included in the National Genomics Test Directory will not be centrally funded and will incur a charge.

National Genomics Test Directory indication: **R**_____

Molecular genetic test (EDTA) Specify which gene(s) to be tested. If more than one test is required, please list preferred order.

Diagnostic (specify): _____ Full screen / Targeted mutation screen
Follow-up (specify): _____ Carrier test / Predictive test / Other (provide details)

Array-CGH (EDTA) Please include a detailed phenotype in the Reason for Referral box.

Specify/state (where applicable):
Degree of cognitive delay; the developmental disorder; neurodevelopmental/behavioural problems; the neurological disorder; growth abnormalities; congenital malformations/dysmorphisms; endocrine/metabolic conditions.

Karyotyping (lithium heparin) **Additional related tests (EDTA)**

Infertility CF Y microdeletion
Premature ovarian failure NB: For FMR1 premutation testing an EDTA blood is also required
Other (provide details)

Details of any previous genetic investigations

Are Rare and Inherited Disease eligibility criteria fulfilled? **Y / N**

In submitting this sample the clinician confirms that consent has been obtained for testing and storage. Anonymised stored samples may be used for quality control procedures including validation of new genetic tests.



SAMPLE REQUIREMENTS: Chromosome and DNA analysis (Includes Array-CGH, FISH, MLPA, gene panels and single-gene testing)

ACCEPTANCE CRITERIA

The National Genomics Test Directory specifies which tests are funded by NHS England, together with their eligibility criteria (<https://www.england.nhs.uk/publication/national-genomic-test-directories/>). Clinical Genetics services are available if required for advice or discussion of possible specific genetic diagnoses. For rare or unusual cases, please contact Wessex Clinical Genetics Service, Level G, Princess Anne Hospital, Coxford Road, Southampton, Hants. SO16 5YA, tel: 02381 206170.

SAMPLE COLLECTION

Blood for array-CGH analysis:

Please collect **5 ml** of blood taken into an **EDTA** tube. **Mix well** by inverting the tube after collection. For infants, a minimum of **1 ml** is required.

Blood for DNA analysis:

Please collect **5 ml** of blood in an **EDTA** tube. **Mix well** by inverting the tube after collection. For infants, a minimum of **1 ml** is required.

Blood for karyotype and/or FISH analysis:

Please collect **5 ml** of blood into a **lithium heparin** tube. **Mix well** by inverting tube after collection. For infants, a minimum of **1 ml** is required.

OTHER TISSUES

Other tissue types may be processed under special circumstances; please contact the laboratory to discuss requirements.

Tumour tissue for DNA analysis may be sent as formalin-fixed wax block sections.

Fresh tissue samples should be collected in **sterile empty containers**. **DO NOT fix these tissues**.

Details on both the referral form and the sample tube should be **complete and legible**. We reserve the right to refuse to process samples with incomplete, illegible or ambiguous patient information.

Any samples in the wrong tube or medium, or which are subject to significant delay in transit, are liable to be rejected. Blood samples from patients who have had a recent white cell blood transfusion may not be suitable for testing.

SAMPLE DESPATCH AND TRANSPORT

Sample and referral form should be sent **together** in a secure leakproof package according to UN P650 packaging instructions, to arrive as soon as possible after collection (e.g. by first class post, courier service or hospital transport) and **within 48 hours for optimum results**. Outside packaging should be clearly labelled '**PATHOLOGICAL SAMPLE FOR DELIVERY TO GENETICS**'.

WRGL opening hours are 9 am - 5.30 pm Mon - Fri; please inform the laboratory of any samples likely to arrive over a weekend or bank holiday, or of anything sent by courier which might arrive outside normal working hours. If there is an unavoidable delay between the sample collection and despatch, blood or tissue may be stored in a refrigerator at 4 °C.

For current information and to download copies of our referral forms and service guides, please refer to our website: www.wrql.org.uk