

(WRGL use only):

Investigation(s):

W

DNA loc:

In before?

Initials

Referral reason:

Date of receipt:

**WESSEX REGIONAL GENETICS LABORATORY**

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**SPIRE REFERRALS FOR GENETIC ANALYSIS**

**PATIENT DETAILS**

<b>SURNAME</b>	<b>DATE OF BIRTH</b> _ _ : _ _ : _ _ _ _	<b>SEX</b>	Referring consultant
<b>FORENAME</b>	<b>NHS NUMBER</b>		Referring Spire hospital
Post code	Hospital number		All reports to be sent to Dr Wang, Frimley Park Hospital

Date of collection \_ \_ : \_ \_ : \_ \_

Priority: Routine  Urgent  *State reason for urgency below*

Collected by

**REASON FOR REFERRAL** Provide full clinical details including any relevant family history

**ANALYSIS REQUESTED**

**For infertility patients:**

- CKA2 Karyotyping (lithium heparin tube)
- YMIC Y Microdeletions (EDTA tube)

**For miscarriage patients:**

- MCYT Karyotyping (lithium heparin tube)

**Other - give reason:**

- Karyotyping (lithium heparin tube) \_\_\_\_\_
- Array-CGH (EDTA tube) *Please provide a detailed phenotype in the Reason for Referral box above: include details of any cognitive delay, behavioural or neurodevelopmental problems, growth abnormalities, dysmorphism, congenital malformations, or endocrine/metabolic conditions.*
- Other molecular genetic tests (EDTA tube) *Specify which gene(s) to be tested. If more than one test is required, please list preferred order.*

**Details of any previous genetic investigations**

*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 panel 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. **Please note:** even for private referrals, the implications of a positive or negative result and whether or not having the test would make any difference to the clinical treatment for the patient **must** be clearly explained by a specialist doctor or genetics counsellor. **A Clinical Geneticist must agree to any referrals for carrier testing or for tests that would predict any late-onset genetic disorders.**

### **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 **2 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 **2 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; prior to sending, 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 a 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](http://www.wrql.org.uk)**