Sláinte Leanaí Éireann ag Cromghlinn

Children's Health Ireland at Crumlin

Genomic Testing Request Form

Department of Clinical Genetics (DCG)

Cytogenetics: (01) 409 6737, Molecular Genetics: (01) 409 6733 Email: genetics.lab@childrenshealthireland.ie Laboratory Opening Hours: 09:30 – 17:00, Monday to Friday Lab Use Only

Date Received:

Lab No:

MF-LAB-DCGRequest Revision 3 Active Date:19/02/2025

Patient Details – Use sticker if available, otherwise add any missing information **Ensure all information matches on the paperwork and sample container**			Referring Clinician/Healthcare Professional Details		
Surnan				Consultant/GP: (Name in full)	
Forename:				Telephone No.:	
DOB:				Email:	
Hospital No:		Patient Address:		Hospital Name: (in full)	
Path N	o.:			Dept. / Ward:	
Ped. N	p.:	Eircode:		Cc. Report To:	
Biological Sex:		Ethnicity:			
Sample Type(s):					Sample Date:
Taken By:		Foetal gestation:			Sample Time:
	atient has had a bone marrow/ste ransfusion, please contact the lab				
Sample Type Guidance		EDTA Blood: For the majority of DNA-based Testing			
Sample		Lithium Heparin (Li-Hep	Lithium Heparin (Li-Hep) Blood: For Karyotyping and FISH		
Test(s) Required:					
Test [Details	Clinical Details and			
Test [Details Microarray	Please give clinical detail	ls & details of previous	genetic investigations	in the family, if known. tails of affected family members and familial variants.
	Microarray Diagnostic Screen/Test	Please give clinical detail	ls & details of previous	genetic investigations	
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	Microarray Diagnostic Screen/Test Predictive/Pre-symptomatic	Please give clinical detail	ls & details of previous	genetic investigations	
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	Microarray Diagnostic Screen/Test Predictive/Pre-symptomatic Test Carrier Test Family Segregation Studies	Please give clinical detail	ls & details of previous	genetic investigations	
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Patient Details

The following patient details are mandatory, other details should be completed as fully as possible:

- Surname & Forename
- **DOB** Date of Birth
- Patient's Biological Sex

Please ensure a minimum of 2 unequivocal identifiers are present and matching on tubes and request form.

- Patient full name
- Date of birth and/or
- MRN

NOTE: For referral of a baby, where the baby's first name is not present on both the request form and sample tube (e.g. indicated as Baby, Infant, Baby of Mary, Mary's baby, Twin one or no first name provided), the surname, date of birth and MRN must be provided on both the request form and sample tube.

Samples that do not conform to the acceptance criteria will be rejected and will not be processed by the laboratory. A record of the specimen will be made in the relevant Laboratory Information System and the reason for its rejection noted. A report/letter will be sent to the requesting clinician detailing the reason for rejection.

Test Required and Clinical Details

Full details of 'Test(s) Required' and 'Clinical Details and Relevant Family History' must be supplied to ensure the correct analysis is performed. It is the responsibility of the Referring Clinician to provide as much detail as possible to ensure the correct test(s) are completed on the patient. If required, additional reports and letters can be attached to this request form.

Refer to the CHI website https://www.childrenshealthireland.ie/list-ofservices/clinical-genetics/ for full details of all clinical genetics tests.

Referring Clinician/Healthcare Professional

The following referring clinician/healthcare professional details are mandatory:

- Consultant/GP name: Initials are not acceptable as the laboratory cannot identify the clinician/healthcare professional. A minimum of first initials and surname must be provided. Names must be legible.
- Hospital name: Should be clearly identifiable; initials are not acceptable, as the laboratory cannot identify the hospital.

Other details should be completed as fully as possible:

- Department/Ward: Must be clearly identifiable.
- Telephone Number: This will aid queries relating to test request.
- Email: This will aid gueries relating to test request.
- **Cc. Report To:** Use this space if the referring consultant requires other healthcare professionals to receive a report copy.

Sample Details

- There are no special requirements or preparation of the patient prior to taking blood samples for genetic tests. Fasting is not required.
- Once samples have been collected, do not freeze or expose the samples to excessive heat (>40°C).
- Please ensure samples are received by the laboratory in a timely manner.
- Ensure all sample collection containers are within their expiry date, using out of date tubes may compromise the patient result.
- In the interest of patient safety and ensuring a genetic result is not compromised, please do not send blood samples to the DCG laboratory that have been previously processed/analysed on another instrument. Such samples will not be processed by the DCG laboratory.

High Infection Risk: The laboratory must be informed of any infection risk associated with submitted samples. The sender has the responsibility for minimising the risk to laboratory staff by giving sufficient information to enable the laboratory to take appropriate safety precautions when testing a specimen.

Factors known to affect the performance of the examination/interpretation of the results: If the patient has had a bone marrow or stem cell transplant/blood transfusion, please contact the laboratory to discuss testing options prior to sending a sample.

Postnatal Samples for DNA Testing (all genomic testing except Karyotyping and FISH)

- Mix well to avoid clotting, Store sample at 4°C if required.
- Venous Blood: Use EDTA tube only.
- Minimum 1ml for adults, children, & neonates.
- Other Sample Types: By prior arrangement only.

Postnatal Samples for Karyotyping and FISH

- Store overnight at 4°C if required.
- The sample must arrive in laboratory within 48 hours of being taken.
- Venous Blood: Use Lithium Heparin (Li-Hep) tube only.
- Minimum 1 ml adults, children, & neonates. (NOTE: 3-5ml is preferred if possible)

Tissue Type: For solid tissue samples the tissue type and anatomical site (if relevant) should be specified. Please contact the laboratory for further information.

Sample Packaging: The sample container should be sealed in a biohazard bag in case of a leakage. To prevent contamination of referral form and paperwork this should not be sealed with the sample. All packaging should adhere to the HSE National Guidelines for the Preparation for Transport of Patient Specimens and other Biological Materials. (See

https://assets.hse.ie/media/documents/ncr/Guidelines_for_the_Preparation_for_Tr ansport_of_Patient_Specimens_and_othe_uUk13xH.pdf)

Consent

- The Referring Clinician is responsible for ensuring valid consent is obtained from the patient for testing. Consent should be obtained, and documented, as per the
 HSE National Consent Policy. Also, refer to the HSE Genetic and Genomic Testing Patient Information Leaflet (https://www.hse.ie/eng/about/who/nationalgenetics-and-genomics/hse-genetic-genomic-testing.pdf).
- If the patient has consented to DNA/sample storage (see section below), please tick the box on the request form.
- The patient must be advised that the sample may be used for quality assurance and training purposes (see section below) as per CHI policy on secondary use of patient data. Please advise the laboratory of any restrictions.

Storage of Remaining DNA or Cells

- Patients/guardians can consent to the long-term storage of the DNA sample (i.e. for DNA to be stored in the Department of Clinical Genetics according to HSE retention policy or until the patient removes consent for storage). The benefit of storing DNA is that it can be used to aid family studies if required and/or for use in further genetic testing (if consent provided), meaning that a repeat blood sample is not required from the patient.
- There is no obligation to provide consent for DNA storage; testing can proceed in the absence of consent to store.
- However, without consent to store, the DNA sample will be discarded following testing.

Quality and Training Purposes

- Quality assurance and audits are an integral part of the quality management system of any diagnostic laboratory, and are frequently used in the introduction of new technology and/or improving testing methods.
- The use of patient samples greatly improves the data generated from quality assurance testing and audits.
- NOTE: The patient sample will never be compromised if/when it is used for quality assurance testing.
- There is no obligation to provide consent for the use of patient sample for quality assurance and audit purposes.