

R14 Test Request to be treated as URGENT

Genomic Medicine Service

National Genomic Test Directory Clinical Indication R14 Rapid Genome Sequencing Test Request

Please **electronically** complete this form and email to the laboratory (rduh.exeterexome@nhs.net) **BEFORE** sending any samples. Ensure that email addresses are provided for the responsible clinician and clinical geneticist.

CONSENT: Receipt of samples for testing assumes that **informed consent** has been obtained for all family members being tested and the possibility of incidental findings has been discussed.

Please send at least 1µg of DNA per individual to: Exeter Genomics Laboratory, RILD Level 3, Royal Devon University Healthcare NHS Foundation Trust, Barrack Road, Exeter, EX2 5DW.

Please indicate the type of referral: ☐ NICU ☐ PICU ☐ Other:
☐ Current pregnancy (EDD:)

Patient first name:		Life status: <input type="checkbox"/> Alive <input type="checkbox"/> Deceased	Ethnicity:
Patient last name:		Family test: <input type="checkbox"/> Trio <input type="checkbox"/> Singleton <input type="checkbox"/> Duo <input type="checkbox"/> Parental	Consanguinity: <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown
Date of birth: dd/mm/yyyy	Hospital number:	Trio – affected child and both parents (gene-agnostic) Duo – affected child and one parent (gene panel) Singleton – affected child only (gene panel) Parental – both parents but sample unavailable from affected child (autosomal recessive gene-agnostic)	
Gender (if phenotypic sex is different please state): <input type="checkbox"/> Male <input type="checkbox"/> Female <input type="checkbox"/> Other:			
NHS number (or postcode if not known)			
Postcode:			

Clinical information and any relevant family history

Reason for testing – clinical summary (this text, or an abbreviated version of it, will be included in the report): Please also include relevant information for relatives and relationship to other tested individuals, including disease status and age of onset

Specific rare or inherited diseases that are suspected or have been confirmed: Please list

HPO terms (<https://hpo.jax.org/app/>) to describe phenotypes in this individual (OPTIONAL): Please list

Family DNA samples provided (parents for trio tests)

Surname	Forename	Date of birth dd/mm/yyyy	NHS number	Gender	Deceased	Status
				<input type="checkbox"/> Male <input type="checkbox"/> Female <input type="checkbox"/> Other:	<input type="checkbox"/> No <input type="checkbox"/> Yes	<input type="checkbox"/> Affected <input type="checkbox"/> Unaffected
				<input type="checkbox"/> Male <input type="checkbox"/> Female <input type="checkbox"/> Other:	<input type="checkbox"/> No <input type="checkbox"/> Yes	<input type="checkbox"/> Affected <input type="checkbox"/> Unaffected

Clinician details

Responsible clinician / consultant paediatrician: Name Department Hospital	Email address for report: (nhs.net)
	Telephone number:
Clinical geneticist: Name Department Hospital	Email address for report: (nhs.net)
	Telephone number: