

Genomic Medicine Service

National Genomic Test Directory Clinical Indication R89 Non-urgent Exome Sequencing Test Request

Please **electronically** complete this form and email to the laboratory rdurh.exeterexome@nhs.net **BEFORE** sending any samples. Ensure that email addresses are provided for the responsible clinician and/or 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.

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

Differential diagnosis: 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	NHS number	Gender	Deceased	Status
		dd/mm/yyyy		<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
		dd/mm/yyyy		<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: