

# Genomic Medicine Service

## National Genomic Test Directory Clinical Indication R312 Parental Sequencing for Autosomal Recessive Disorders Test Request

Please **electronically** complete this form and email to the laboratory [rdh.exeterexome@nhs.net](mailto:rdh.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.

Please indicate the type of referral: ☐ Non-urgent ☐ Urgent

If **urgent:** Current pregnancy (EDD: )

### Parental DNA samples provided

Surname	Forename	Date of birth <small>dd/mm/yyyy</small>	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

### Proband/ pregnancy details (sample unavailable) – complete as applicable

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

☐ Multiple affected children/ pregnancies – please provide additional details below

### 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

### Clinician details

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