

## Routine Intellectual Disability OR Congenital Malformation and/or Dysmorphism Sequencing Request Form

**SAMPLE REQUIREMENTS**

**PROBAND-** >1ml, whole blood in EDTA, **BOTH PARENTS** (if trio required) – 3-4ml each, whole blood in EDTA

**Please send samples with filled in referral form to: AWMGS, University Hospital Wales, Cardiff, CF14 4XW**

Tel: +44(0)2921844023 | [Lab.genetics.cav@wales.nhs.uk](mailto:Lab.genetics.cav@wales.nhs.uk) | <https://medicalgenomicswales.co.uk>

**TEST REQUIRED (please only tick one where possible) – ONLY available through referral from Clinical Genetics**

**Congenital malformation and/or dysmorphism syndromes (R27)**  
including copy number analysis by array where not done previously  
(except Rasopathies panel)

- |  |                          |
|--|--------------------------|
| 1. TRIO WGS                                      | <input type="checkbox"/> |
| 2. TRIO WES for fetal testing                    | <input type="checkbox"/> |
| 3. Clefting SINGLETON WES PANEL                  | <input type="checkbox"/> |
| 4. Rasopathies SINGLETON WES PANEL (no array)    | <input type="checkbox"/> |
| 5. Skeletal dysplasia (R104) SINGLETON WES PANEL | <input type="checkbox"/> |
| 6. Severe microcephaly (R88) SINGLETON WES PANEL | <input type="checkbox"/> |

**Intellectual Disability (R29)** including copy number analysis by array where not done previously

- |                  |                          |
|------------------|--------------------------|
| 1. TRIO WGS      | <input type="checkbox"/> |
| 2. SINGLETON WES | <input type="checkbox"/> |

**Urgent - ongoing pregnancy in mother**

If a trio is unavailable, please contact the laboratory to discuss further testing options.

Requesting Consultant	Requested by (if not Consultant)
Name:	Name:
Email:	Email:
Referring unit:	Copies to:
Phone:	

Attach patient addressograph below if needed

<b>Proband's first name:</b>	Sex:	<b>Previous genetic testing:</b> <b>Array:</b> YES <input type="checkbox"/> NO <input type="checkbox"/> <b>Fragile X:</b> YES <input type="checkbox"/> NO <input type="checkbox"/> <b>Other:</b> YES <input type="checkbox"/> NO <input type="checkbox"/> Please provide details or reports:
<b>Proband's last name:</b>	Genetics family no:	
Date of birth:	Hosp no:	
NHS no.:	Ethnicity (required):	
Address:	Sample date and time:	
Postcode:	Sample type:	

Consent for genetic testing and DNA storage is assumed when the request for testing is received  
It is the responsibility of the referring clinician to ensure that appropriate consent has been obtained

<b>Proband name:</b>		<b>Proband DOB:</b>	
----------------------	--	---------------------	--

Family members to be tested (for TRIO testing only, not required for singleton testing)							
First name:	Last name:	DOB:	NHS no:	Sex:	Ethnicity:	Relationship to proband:	Please tick if affected:
							<input type="checkbox"/>
							<input type="checkbox"/>

**Please note:** Choosing that a parent is affected will initiate additional analysis of inherited variants when requesting Intellectual Disability trio WGS. See WGS test information sheet on our website for details.

<p><b>HPO Terms</b> – please use description and number as available on the website (<a href="https://hpo.jax.org/app/">https://hpo.jax.org/app/</a>)</p> <p>To ensure correct interpretation of the results, please provide as much phenotypic and family history information as possible.</p>	<p><b>Family history / Pedigree</b></p> <p>Please include information about health problems in relatives and relationships to other people, including disease status and age of onset. Include details about miscarriages and stillbirths.</p> <p>Consanguinity (if YES, please provide details) <input type="checkbox"/></p>
---	---

<b>Additional information to include specific genes or differential diagnosis that are considered likely</b>

Consent for genetic testing and DNA storage is assumed when the request for testing is received  
It is the responsibility of the referring clinician to ensure that appropriate consent has been obtained