AWMGS All Wales Medical Genomics Service

GIG CYMRU NHS WALES

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 | 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 ☐
2. TRIO WES for fetal testing ☐
3. Clefting SINGLETON WES PANEL ☐
4. Rasopathies SINGLETON WES PANEL (no array) ☐
5. Skeletal dysplasia (R104) SINGLETON WES PANEL ☐
6. Severe microcephaly (R88) SINGLETON WES PANEL ☐

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

1. TRIO WGS ☐
2. SINGLETON WES ☐

☐ **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

Proband's first name:	Sex:	**Previous genetic testing:** **Array:** YES ☐ NO ☐ **Fragile X:** YES ☐ NO ☐ **Other:** YES ☐ NO ☐ Please provide details or reports:
Proband's last name:	Genetics family no:	
Date of birth:	Hosp no:	
NHS no.:	Ethnicity (required):	
Address: Postcode:	Sample date and time: 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

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Proband name:		Proband DOB:	

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:
							☐
							☐

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.

HPO Terms – please use description and number as available on the website (https://hpo.jax.org/app/)

To ensure correct interpretation of the results, please provide as much phenotypic and family history information as possible.

Family history / Pedigree

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.

Consanguinity (*if YES, please provide details*) ☐

Additional information to include specific genes or differential diagnosis that are considered likely

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