All Wales Medical Genomics Service-Whole Exome Sequencing Request Form

Patient details are required. Please ensure patient name is included on pedigree (below)

Surname	Forename	Date of Birth	NHS Number	Sex
****Please note any remaining sample is to be returned to AWMGS on a monthly basis*****				

Requesting Clinician (Full Name)		**Hospital**	
Email address (For Report*)		**Ward/Clinic**	
Signature			
*Reports will be received by AWMGS from the testing laboratory. AWMGS Administration will arrange for a printed copy of the report to be posted and an electronic copy of the report to be emailed to the referring clinical team(s) via the hospital and email address(es) given above.			
TEST REQUESTED	SINGLETON GENE PANEL ✓ **Details of gene panel required:** (Please include 'R' number as stated in the National Genomic Test Directory): https://www.england.nhs.uk/publication/national-genomic-test-directories/		

I confirm that informed consent has been obtained for all family members being tested and the possibility of incidental findings has been discussed (please see appendix 1) - please select one of the following:

Patient **CONSENTS** to receive incidental findings	☐
Patient **DOES NOT CONSENT** to receive incidental findings	☐

Referral forms without one of the two incidental findings consent options ticked will not be processed and a request to provide this information will be issued by the AWMGS laboratory before testing can be initiated

Description of Clinical Features	**Family History/Pedigree**
Please list the patient's clinical features using HPO terms where possible, see: https://hpo.jax.org/. Please see Appendix 1 for further information.	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. If it is indicated below that parents are affected, please describe the phenotype relative to the child.
Please enter all clinical feature terms and HPO codes e.g. Unsteady gait HP:0002317	

Referral forms without clinical details and appropriate HPO codes will not be processed and a request to provide this information will be issued by the AWMGS laboratory before testing can be initiated

Previous Genetic Testing – Please include previous tests ordered and results. Where possible, reports should also be provided.

Consanguinity? If yes, please provide details.

Copies of report sent to (quoting AWMGS lab number):
Requesting Clinician (1):
Requesting Clinician (2):

Requesting Laboratory:
All Wales Medical Genomics Laboratory Service
Institute of Medical Genetics
University Hospital of Wales
Heath Park
Cardiff
CF14 4XW

AWMGS Admin team: **Pt-lhb.admin-genetics@nhs.net**

******PLEASE NOTE: Our IT policies prohibit the use of any email other than @nhs.net accounts as a means of accepting patient reports. Please send by post, FAX or to an @nhs.net email address******

Invoice ONLY (quoting AWMGS lab number):

CAV Accounts Payable:
Finance Department
University Hospital of Wales
Cardiff
CF14 4XW

PLEASE ONLY EMAIL INVOICES TO:
NWSSP.APCAV@wales .nhs.uk
AND
accounts.payable2@wales.nhs.uk

******DO NOT SEND REPORT TO FINANCE DEPARTMENT*******

Appendix 1:

Incidental findings

Reporting of incidental findings is restricted to findings within the diagnostic genes on the gene panel, only. It is possible the sex chromosome imbalances may also be detected as a part of the sequencing quality control process; such findings would be included on the patient's report, if consent is given to receive incidental findings.
Should you have any questions or wish to clarify understanding around this area, please contact the All Wales Clinical Genetics service https://medicalgenomicswales.co.uk/index.php/en/

Human Phenotype Ontology (HPO)

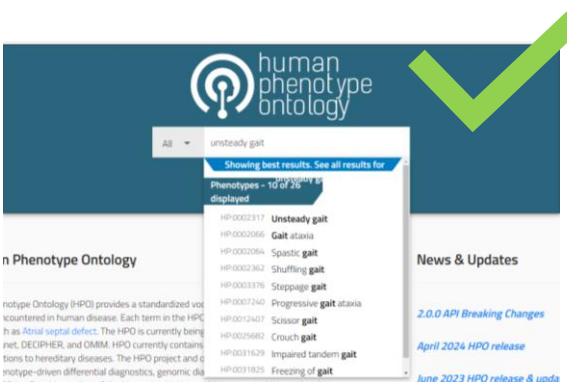
For information and to search for the specific HPO terms relating to your patient phenotype, please follow this link/refer to: https://hpo.jax.org/

Providing details of the phenotype displayed by the patient is very important in enabling the testing laboratory to properly interpret and classify genomic test findings. Inclusion of the standardized HPO codes on the referral form, where possible, will avoid any ambiguity in interpretation of the features described. Referrals received without details of the patient **phenotype and the associated HPO code** will not be processed and a request to the referring clinician to provide this information will be issued by the AWMGS laboratory before testing can be initiated.

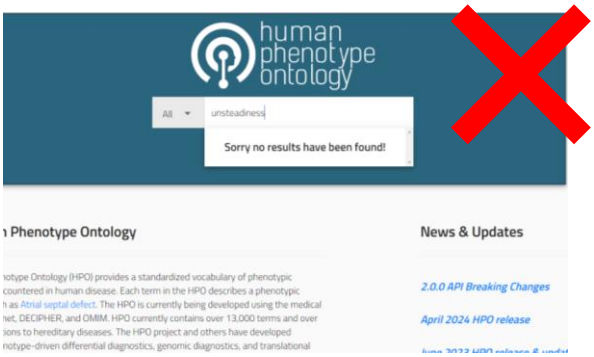
Completing the 'Description of Clinical Features' field with HPO terms and codes:

1) Search for term in HPO search field:

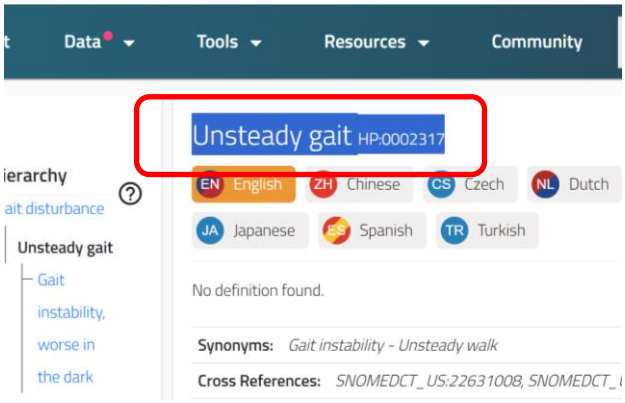
'*Unsteady gait*' – several returns to select from



'*Unsteadiness*' – not recognised – no code Search for a more specific term



Select appropriate term. In the new window, select and copy the term and HP: code text to clipboard



Paste term and HP: code into the Description of Clinical Features field of the WES request form

Description of Clinical Features	Family History/Pedigree
Please list the patient's clinical features using HPO terms where possible, see: https://hpo.jax.org/. Please see Appendix 1 for further information.	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. If it is indicated below that parents are affected, please describe the phenotype relative to the child.
Unsteady gait HP:0002317	

Repeat for all clinical features to be shared with the laboratory.

To ensure that the testing laboratory has the specific phenotypic information required to fully interpret genomic test results, it is important that the referring clinician, with knowledge of the specific phenotype in the patient, enters the most appropriate HPO code(s) and term(s) on the referral form, with reference to the HPO website.