

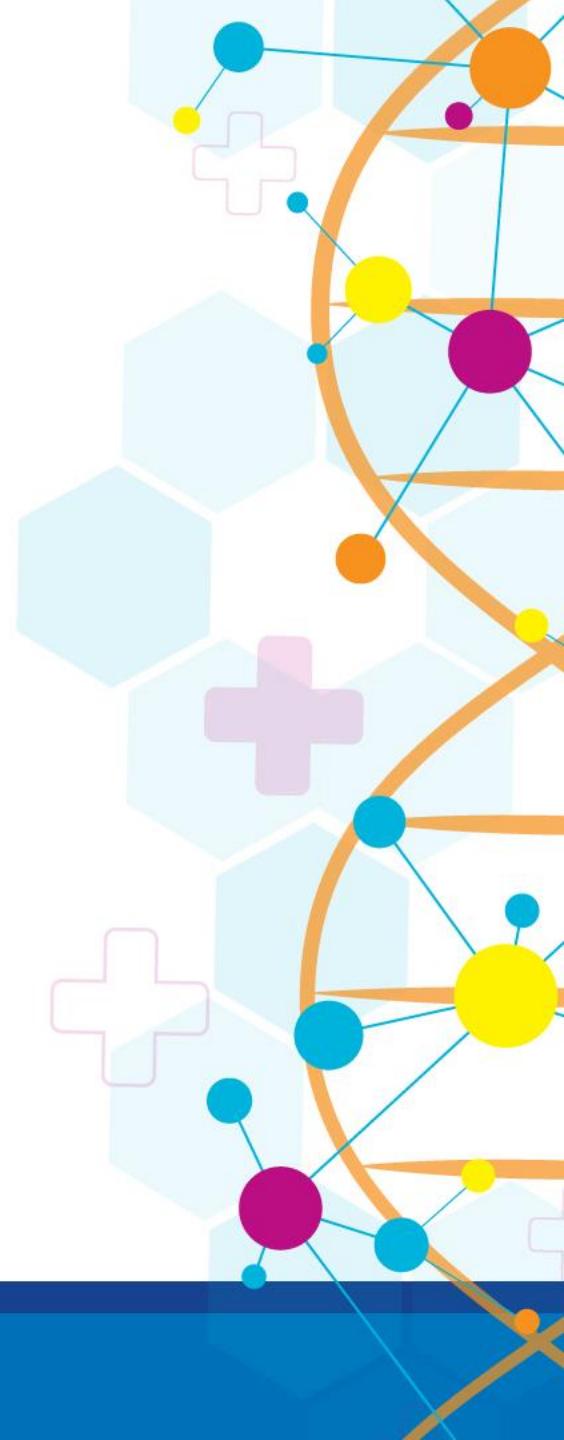
Right Test, Right Time: Essential Updates in Genomic Testing

Please complete pre-
survey:



Objectives

- The main changes
- How to navigate the new forms
- When to test, when not to test
- Why are the changes happening now
- Where can I get further information/guidance
- Q+A



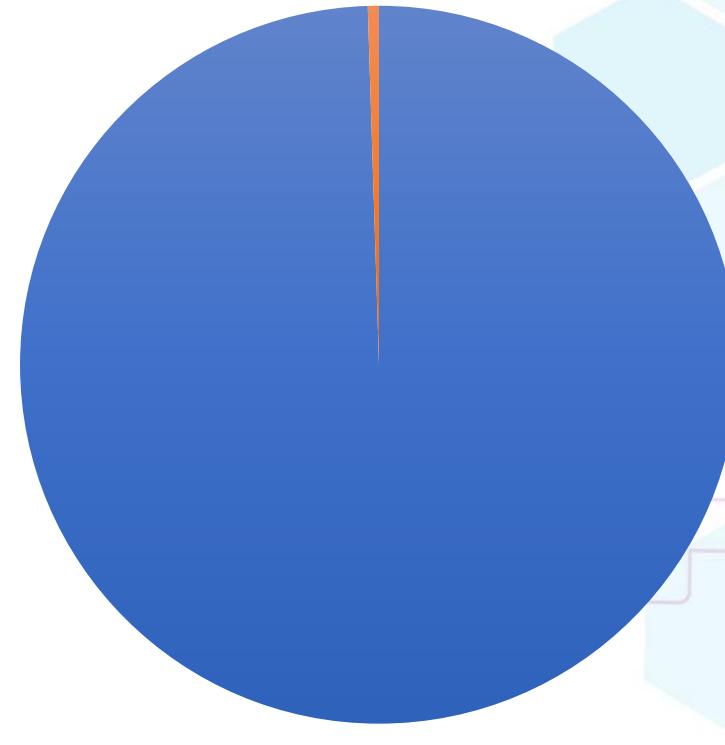
The main changes



1. Whole Genome Sequencing should be the first line test

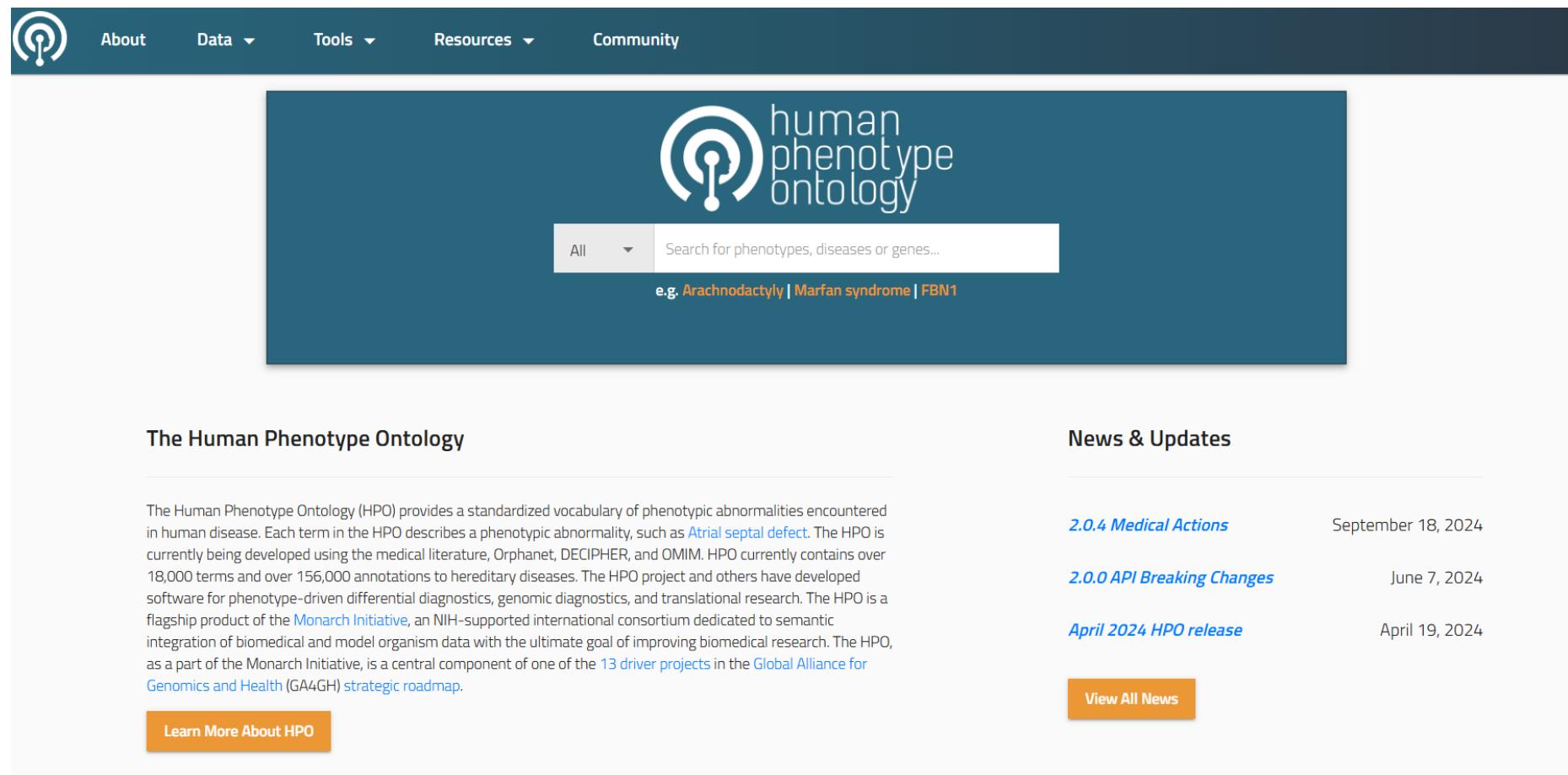
- WGS is **most comprehensive** test and able to detect almost every type of genomic variant
- Has a **higher sensitivity** for majority of variant types
- Alternatives requests, such as microarray are expected to be **exceptionally rare** and only available when there is demonstrable clinical utility

Primary test for a rare disease clinical indication



■ WGS ■ Exceptional cases*

2. HPO



The Human Phenotype Ontology (HPO) provides a standardized vocabulary of phenotypic abnormalities encountered in human disease. Each term in the HPO describes a phenotypic abnormality, such as [Atrial septal defect](#). The HPO is currently being developed using the medical literature, Orphanet, DECIPHER, and OMIM. HPO currently contains over 18,000 terms and over 156,000 annotations to hereditary diseases. The HPO project and others have developed software for phenotype-driven differential diagnostics, genomic diagnostics, and translational research. The HPO is a flagship product of the [Monarch Initiative](#), an NIH-supported international consortium dedicated to semantic integration of biomedical and model organism data with the ultimate goal of improving biomedical research. The HPO, as a part of the Monarch Initiative, is a central component of one of the [13 driver projects](#) in the [Global Alliance for Genomics and Health \(GA4GH\) strategic roadmap](#).

[Learn More About HPO](#)

News & Updates

[2.0.4 Medical Actions](#) September 18, 2024

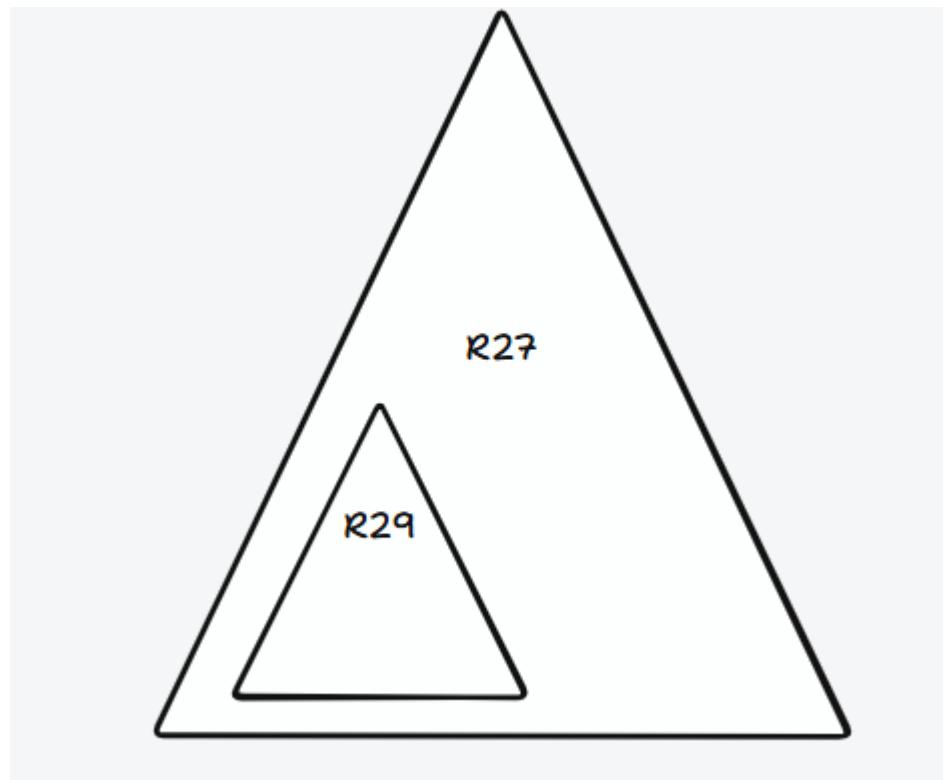
[2.0.0 API Breaking Changes](#) June 7, 2024

[April 2024 HPO release](#) April 19, 2024

[View All News](#)

[HPO website](#)

3. No R29 – amalgamation



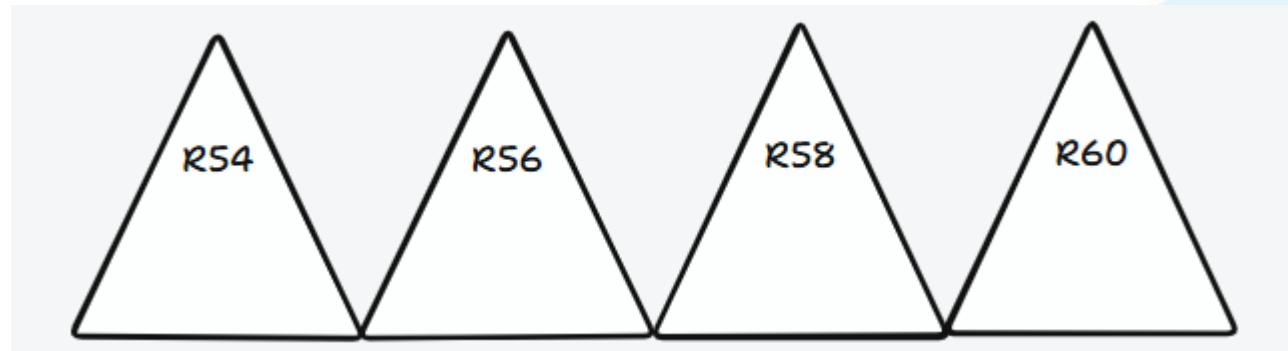
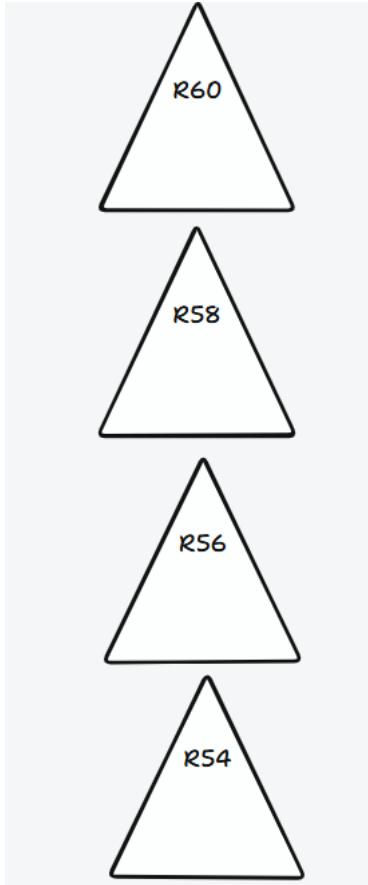
- R27 (Paediatric disorders)
- This panel contains these 13 panels:
- DDG2P
- **Intellectual disability (R29)**
- Early onset or syndromic epilepsy
- Likely inborn error of metabolism
- Skeletal dysplasia
- Monogenic hearing loss
- Paediatric disorders - additional genes
- Clefting
- Neurological ciliopathies
- Renal ciliopathies
- Ophthalmological ciliopathies
- Limb disorders
- Skeletal ciliopathies



4. Adult Neurology

Request any below...

..get all (Neurology super panel)



- ✓ Increases diagnostic yield
- ✓ Reduces likelihood of time-consuming re-analysis.

Worries

- Incidental findings
- Turn around times
- “I already know what it is” – Is there a clinical utility then?
- Good clinical reason to not? Can still request to not amalgamate



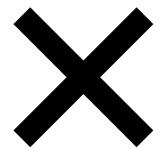
5. Drive to reduce Re-analysis



Laboratories receive large volumes of **R387 WGS re-analysis** requests because original panel selections were not comprehensive enough

Request all relevant panels as part of the primary investigation

10% of lab work is re-analysis and a significant amount of that is from adult neurology



- R60



- Neurology Super Panel
(R54, R56, R58, R60)



How to navigate the new forms



RARE DISEASE AND REPRODUCTIVE GENOMICS TEST REQUEST

PATIENT DETAILS		CLINICIAN DETAILS			
Forename:		Consultant/Clinician (full name & specialty):			
Surname:					
Date of birth:	Sex:	Report copies to:			
Address:					
Hospital number:		Tel number/Bleep:			
NHS No:		Email address:			
Ethnicity:					
In / Out patient	NHS / Private patient	Urgent / Routine	Hospital (please specify hospital site within Trust):		
Date of next appointment/MDT:					
CLINICAL DETAILS Please provide detailed information. Incomplete referral forms will not be processed.					
GENOMIC TESTING REQUIRED Please include NHSE test directory (TD) indication and code and details of how the patient meets the National Genomic Test directory eligibility criteria: https://www.england.nhs.uk/publication/national-genomic-test-directories/					
Test required and TD code(s):					
CLINICAL UTILITY (Please provide additional information with other relevant clinical information above)					
<input type="checkbox"/> Patient management (determining therapeutic decisions and/or clinical investigations and/or surveillance programme) <input type="checkbox"/> Patient, parents or adult relative reproductive decision making <input type="checkbox"/> Unaffected relatives are seeking predictive testing					
Status: (please circle)		Prenatal	Presymptomatic	Diagnostic	Carrier testing
Details of prenatal screening: (if applicable)		Current gestation:	Type of screening:	Screening risk:	
DETAILS OF AFFECTED FAMILY MEMBERS IF RELEVANT (please state relationship to the patient)					
Forename and Surname:		DOB:	Lab/NHS No:		
Details of previous genomic testing:					
SPECIMEN DETAILS <small>If a specimen is known to present an infection risk, please label it with a red 'DANGER OF INFECTION' label.</small>					
Sample type:		Venous blood			
(please circle)		Tissue (please circle)			
Date collected:					

FOR INTERNAL GLH LAB USE ONLY		
Date of receipt:	Number & volume & type of sample(s) received:	Place lab reference sticker here:

SAMPLE REQUIREMENTS																							
Venous blood For molecular genetic testing (e.g. NGS, SNP array, QF-PCR) please send DNA or 3-5ml VB in EDTA. For conventional cytogenetics (e.g. karyotype, FISH) please send 3-5 ml VB in lithium heparin. For Fanconi Anaemia or Bloom Syndrome, please send 5ml PB in LH, to arrive within 48 hours (samples received in EDTA will not be processed).																							
Prenatal CVS: 10-30mg in transport medium. Amniotic fluid: 10-20ml in universal container. Fetal blood: Lithium heparin and EDTA (min 0.5ml). Maternal/paternal blood: 3-5ml VB in EDTA.																							
Non-invasive prenatal NIPT (trisomy 13, 18, 21): 10ml maternal blood in Streck BCT tube. NIPD (fetal sexing/single gene disorder): 10-20ml maternal blood in Streck BCT tube. Invert Streck tubes x10 and store at room temperature. NIPD familial control samples: DNA or 3-5ml VB in EDTA.																							
Tissue Fresh in a sterile container and NOT fixed in formalin. POC/Placental biopsy (containing chorionic villi): 15mm ² in tissue culture medium or sterile saline. Fetal or postnatal tissue biopsy (e.g. skin, muscle, cord): 5mm ² in tissue culture medium or sterile saline. Cardiac/cord blood: 1-2ml in EDTA.																							
SAMPLES SHOULD BE SENT TO THE LABORATORY WITHIN 24 HOURS OR RISK BEING COMPROMISED Please send via hospital transport, courier or 1st class post.																							
GENOMICS LAB CONTACT DETAILS																							
Tel: 0121 335 8036		SHIPPING ADDRESS		LABORATORY OPENING TIMES																			
Email: bwc.genetics.lab@nhs.net		Birmingham Women's Hospital Genetics Laboratory – Ground Floor Mindelsohn Way Edgbaston Birmingham B15 2TG		Monday to Friday: 07:00 - 18:00 Saturday: 09:00 - 14:00																			
TRIAGE DETAILS (FOR INTERNAL GLH LAB USE ONLY)																							
<table border="1"> <tr> <td>Priority</td> <td>Prenatal</td> <td>Urgent</td> <td>Presymptomatic</td> <td>Priority</td> <td>Routine</td> </tr> <tr> <td>B number indication</td> <td></td> <td></td> <td></td> <td>Culture</td> <td>Culture</td> </tr> <tr> <td>Additional information</td> <td colspan="4"></td> <td>No culture</td> </tr> </table>		Priority	Prenatal	Urgent	Presymptomatic	Priority	Routine	B number indication				Culture	Culture	Additional information					No culture				
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D Indication 1		Reason																					
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		Reason																					
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West Midlands

PATIENT DETAILS <small>(Printed label if available)</small>		REFERRER DETAILS	
Family name:		Referring Clinician:	Job Title:
First name(s):		Consultant: <small>(if different from above, mandatory)</small>	
Date of birth:	Sex: <input type="radio"/> M <input type="radio"/> F <input type="radio"/> U <small>(Please state if karyotypic and/or phenotypic sex differs from given sex)</small>	Contact Name: <small>(if different from above)</small>	
NHS number:		Additional copies to:	
Hospital number:		Email for report: <small>(PTO for more information)</small>	Tel No:
Address:	Ethnic Origin:	Hospital address:	
Postcode:	NHS Private <small>Please supply the name and address for</small>		
CLINICAL DETAILS AND FAMILY HISTORY <small>PLEASE PROVIDE SPECIFIC INFORMATION DETAILING HOW THE PATIENT MEETS THE NATIONAL GENOMIC TEST DIRECTORY ELIGIBILITY CRITERIA FOR THE TEST BEING REQUESTED (see www.England.nhs.uk/publications/internal-genomic-test-directory for further information)</small> <small>For pedigrees, please mark <input checked="" type="checkbox"/> against person sampled with this request card. Where appropriate identify other family members that may be known to the lab with their full name and date of birth.</small>			
Is the patient or their partner pregnant?		If YES: gestation at sampling by scan?	
For infertility referrals please give partner's name and DOB:			
If this case has been discussed with the Clinical Genetics department, please give name of contact in Genetics:			
TEST(S) REQUESTED – please read consent information overleaf			
NHSE Clinical Indication code/Test ID (R/M Code):			
CLINICAL UTILITY (Please indicate how testing will impact patient care – tick below) <small>Patient management (determining therapeutic decisions and/or clinical investigations and/or surveillance programme). Patient, parents, or adult relative reproductive decision making. Unaffected relatives are seeking predictive testing.</small>			
HIGH RISK SAMPLES: If a specimen is known to present an infection hazard it must be clearly labelled 'DANGER OF INFECTION' and the infection hazard stated.			
Sample requirements – further details available on our website: www.ouh.nhs.uk/geneticslab			
<small>(Clinician – Please tick which tube is required)</small> <small>For Chromosome analysis, Fluorescence In Situ Hybridization (FISH): Blood in LITHIUM HEPARIN (1.5ml)</small> <small>For gene sequencing, specific mutation tests, dosage, SNP array: Blood in EDTA (1.5ml)</small> <small>Fetal tissue requirements: Placenta/ spleen/ POC/ skin/ kidney to be sent in tissue transport media (available on request to the laboratory). Please DO NOT fix in formalin.</small>			
Has this patient had a recent blood transfusion or ever had a bone marrow transplant?		If yes, give details below	
Other (Please state)		Date sample taken:	
TWO patient identifiers are required on all sample tubes.			
For Lab use only: Date of receipt:		Sample cond./Vol.:	
Version: 6.0		Comments:	

In submitting this sample, the clinician confirms that consent has been obtained for testing and storage. Anonymised stored samples may be used for quality control procedures including validation of new genetic tests.

Further information:

In complying with the Human Tissue Act 2004 all surplus tissue samples are discarded once DNA/RNA has been extracted.
 Please be aware that anonymised genomic and clinical data may be shared within and beyond the NHS for diagnostic and research purposes.

Electronic Reporting via Email:

The Oxford Genetics Laboratories are now offering the option to receive reports by Email. If you would like to receive future reports via this method please provide your email address in the referrer details section (securely accredited DCB1596 domain preferred). To set this up, the laboratory will contact you with further information.

Laboratory contact details:

General Enquiries Tel: +44 (0)1865 226001

Duty scientist e-mail: dutyscientist.oxfordgenetics@ouh.nhs.uk

Opening hours: 9.00am – 5.00pm Monday – Friday (excluding bank holidays)

Sample dispatch:

Please send blood samples at room temperature via your local pathology sample transport pathway or by 1st class post or courier to:
(For other samples please enquire or consult website)

Oxford Genetics Laboratories
 Churchill Hospital
 Old Road
 Headington
 Oxford
 OX3 7LE
 UK

N.B. Samples for chromosome analysis should be sent to arrive at the laboratory within 24 hours.

For further information about sample requirements and tests available see:
www.ouh.nhs.uk/geneticslab

Information for patients:

Blood samples can be arranged via your GP or the phlebotomy clinic of your local hospital. This form must accompany the sample.
 Following receipt of the sample, laboratory staff are unable to provide information on samples and test results directly to patients or their relatives. Such enquiries should be directed to the referring clinical teams or the GP.

Oxford

(WGLS use only):

W					
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Investigation(s):

DNA loc:
In before?
Initials
Date of receipt:

Wessex Genomics Laboratory Service (Salisbury)
Salisbury District Hospital, Salisbury, Wiltshire SP2 8BJ
Tel.: +44(0)1722 429080
E-mail: sho-dr.WRGLdutyscientist@nhs.net
Web: www.wrgl.org.uk



Genomic Medicine Service Rare Disease test referral form

PATIENT DETAILS

Address/paragraph label			
SURNAME	DATE OF BIRTH	SEX	Referring consultant (full name)
FORENAME	NHS NUMBER		Hospital / Department
Patient's postcode	Hospital number / Genetics number		Clinician's specialty
NHS England / Other NHS / Private (Address for invoicing if not NHS England):		Additional copies to (name, address, specialty) Clinician's contact number / NHS.net email @nhs.net	

Date of collection ____ : ____ : ____ Collected by: Priority: Routine Urgent
State reason for urgency below
Sample type: EDTA for all referrals + Lithium Heparin if FISH and/or karyotype required

Test selection: please select a test from the National Genomic Test Directory + provide information on the clinical utility of the requested test

N.B.: Both the R number and the test code name (clinical indication) must be provided, and the referral must fulfil the associated eligibility criteria. Refer to <https://www.england.nhs.uk/publication/national-genomic-test-directories/> for details.

National Genomic Test Directory test code(s): R_____

Test code name(s):

Is there a clinical utility? If yes, tick box:

- Patient management (determining therapeutic decisions and/or clinical investigations and/or surveillance programme).
- Patient, parent, or adult relative reproductive decision making.
- Unaffected relatives are seeking predictive testing.

For molecular tests (single gene / gene panels) - tick one of the following boxes:

- Diagnostic full screen (enter R code above)
- Predictive (R242)
- Carrier (R244)
- Diagnostic targeted test (R240)
- Segregation (R375)
- DNA storage only (R346)

Referral reason: please provide full clinical details (including any relevant family history).

DNA will be stored and not tested unless these details are provided

Details of any previous genetic investigations:



University Hospital Southampton
NHS Foundation Trust

In submitting this sample the clinician confirms that consent has been obtained for testing and storage.
Anonymised stored samples may be used for quality control procedures including validation of new genetic tests.

NHS
Central & South
Genomic Laboratory Hub

SAMPLE REQUIREMENTS

ACCEPTANCE CRITERIA

The Genomic Medicine Service came into operation in England in 2020. The Wessex Regional Genetics Laboratory and the Molecular Pathology Department at University Hospital Southampton together form the Wessex Genomics Laboratory Service, within the Central and South Genomics Laboratory Hub (GLH).

The National Genomic Test Directory specifies which tests are funded by NHS England, together with their eligibility and referral criteria: <https://www.england.nhs.uk/publication/national-genomic-test-directories/>.

Please note that any test not included in the National Genomic Test Directory will not be centrally funded and will incur a charge. Please contact the laboratory for further information.

Clinical Genetics services are available if required for advice on rare or unusual cases. Please contact Wessex Clinical Genetics Service, Level G, Princess Anne Hospital, Southampton, SO16 5YA: tel. 02381 206170, <https://www.uhs.nhs.uk/departments/genetics>.

SAMPLE COLLECTION

For all referrals:

Please collect 5 ml of blood taken into an EDTA tube. Mix well by inverting the tube after collection. For infants, a minimum of 1 ml is required.

For referrals requiring karyotype and/or FISH analysis:

Please collect 5 ml of blood into a lithium heparin tube. Mix well by inverting tube after collection. For infants, a minimum of 1 ml is required.

OTHER TISSUES

Other tissue types may be processed under special circumstances; please contact the laboratory to discuss requirements.

Tumour tissue for DNA analysis may be sent as formalin-fixed wax block sections.

Fresh tissue samples should be collected in sterile empty containers. DO NOT fix these tissues.

Tissue samples requiring culturing should be collected into tissue transport medium.

Details on both the referral form and the sample tube should be **complete and legible**. We reserve the right to refuse to process samples with incomplete, illegible or ambiguous patient information.

Any samples in the wrong tube or medium, or which are subject to significant delay in transit, are liable to be rejected. Blood samples from patients who have had a recent white cell blood transfusion may not be suitable for testing.

SAMPLE DESPATCH AND TRANSPORT

Sample and referral form should be sent together in a classification and packaging instruction P650, to arrive by post, courier service or hospital transport and within 48 hours. The sample tube should be clearly labelled 'PATHOLOGICAL SAMPLE FOR GENETIC TEST'.

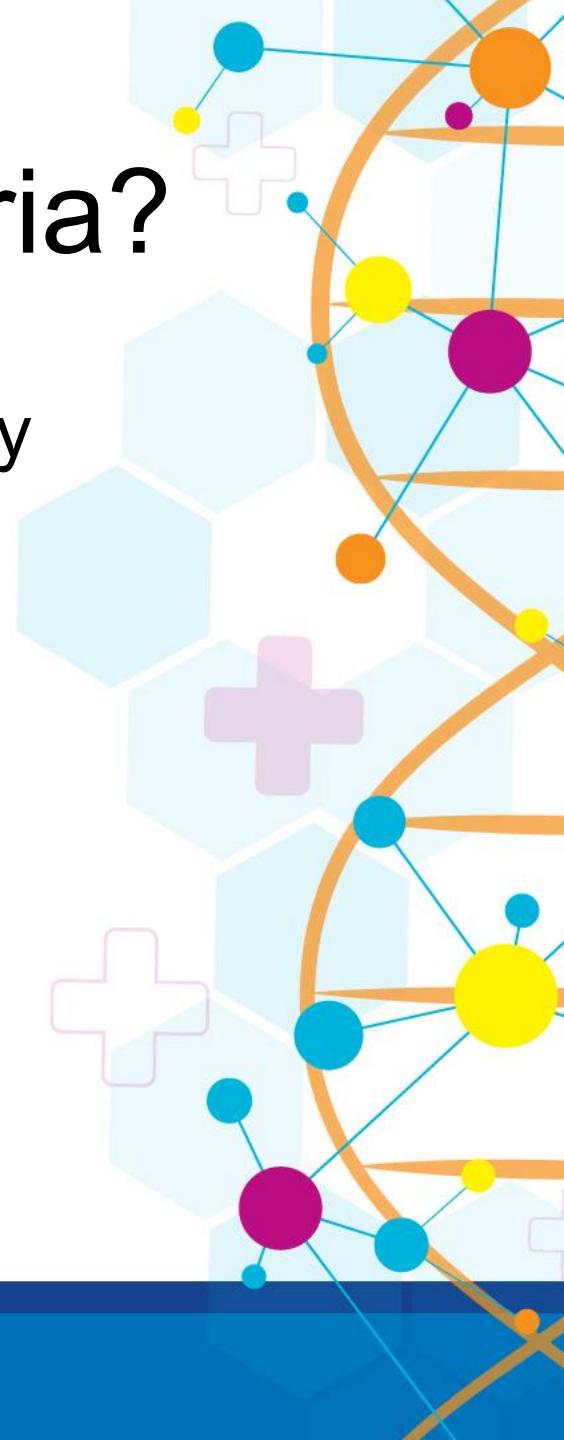
Opening hours are 8 am - 5.30 pm Mon - Fri; please inform the laboratory if there is a weekend or bank holiday, or if anything sent by courier. There is an unavoidable delay between the sample collection and despatch, blood or tissue may be stored in a refrigerator at 4 °C.

For current information and to download copies of our referral forms and service guides, please refer to our website: www.wrgl.org.uk

Wessex

How do they meet the testing criteria?

- Clinical details cross referenced with the test directory
- Family history/ pedigree



How do they meet the testing criteria?



R186 Hereditary haemorrhagic telangiectasia

Testing Criteria

Test where any THREE of the following criteria are met:

1. Epistaxis: spontaneous, recurrent nose bleeds
2. Telangiectases: multiple, at characteristic sites (lips, oral cavity, fingers, nose)
3. Visceral lesions such as gastrointestinal telangiectasia (with or without bleeding), pulmonary arteriovenous malformation (AVM), hepatic AVM, cerebral AVMs, spinal AVM
4. Family history: a first degree relative with HHT according to these criteria (as above) or an autosomal dominant family history of nosebleeds or first degree relative with cerebral AVM / cerebral haemorrhage / pulmonary or hepatic AVM.

Alternatively, test where any ONE of the following criteria are met:

- A) Personal history of at least one pulmonary AVM*
- B) Personal history of two or more AVMs at one or more characteristic sites (pulmonary*, cerebral, hepatic or spinal)
- C) Personal history of at least one AVM and severe epistaxis history
- D) Personal history of telangiectasia, and refractory or frequent transfusions)*

*Pulmonary AVM only if confirmed by cross sectional imaging (therapeutic angiography/surgery). Do not diagnose if only suspected ("bubble echo") or chest x-ray.

In the past:
HTT test please



CLINICAL DETAILS AND FAMILY HISTORY

PLEASE PROVIDE SPECIFIC INFORMATION DETAILING HOW THE PATIENT MEETS THE NATIONAL GENOMIC TEST DIRECTORY ELIGIBILITY CRITERIA FOR THE TEST BEING REQUESTED (see www.england.nhs.uk/publication/national-genomic-test-directories for further information)

For pedigrees, please mark against person sampled with this request card. Where appropriate identify other family members that may be known to the lab with their full name and date of birth.

**Cross reference with test directory and say WHY they are eligible
Nose bleeds present, Blood vessels in skin present, AVM present**

Is the patient or their partner pregnant?

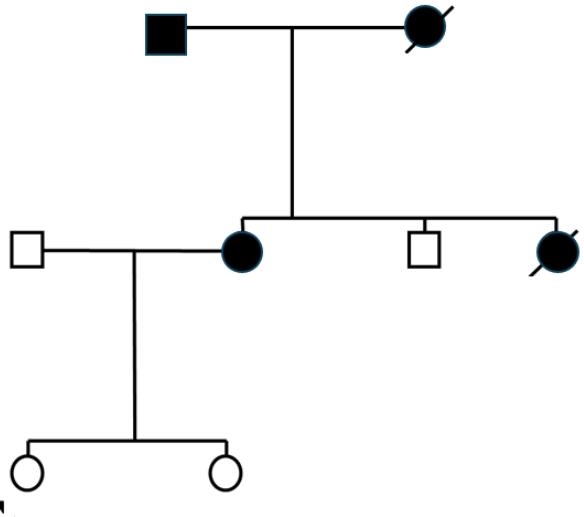
If YES: gestation at sampling by scan?

For infertility referrals please give partner's name and DOB:

If this case has been discussed with the Clinical Genetics department, please give name of contact in Genetics:

Family History/Pedigree

Family Member	Name	DOB
Mother		
Maternal Aunt		
Maternal Grandma		
Maternal Grandfather		



CLINICAL DETAILS AND FAMILY HISTORY

PLEASE PROVIDE SPECIFIC INFORMATION DETAILING HOW THE PATIENT MEETS THE NATIONAL GENOMIC TEST DIRECTORY ELIGIBILITY CRITERIA FOR THE TEST BEING REQUESTED (see www.england.nhs.uk/publication/national-genomic-test-directories for further information)

For pedigrees, please mark against person sampled with this request card. Where appropriate identify other family members that may be known to the lab with their full name and date of birth.

Is the patient or their partner pregnant?

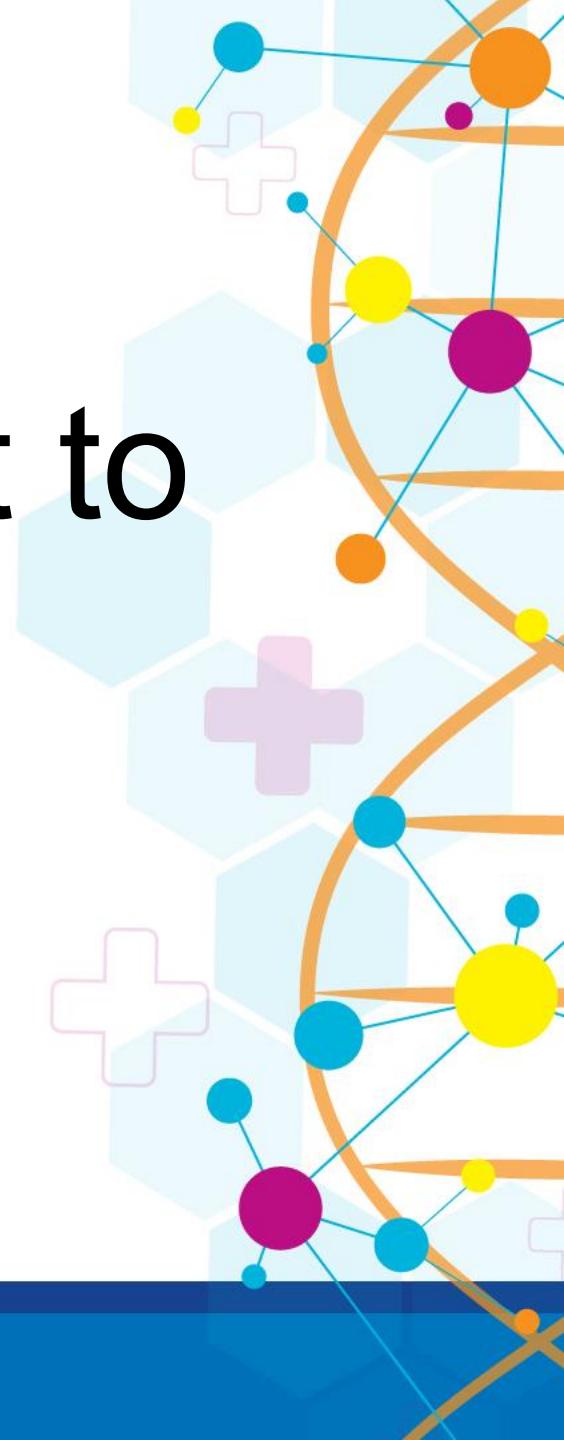
If YES: gestation at sampling by scan?

For infertility referrals please give partner's name and DOB:

If this case has been discussed with the Clinical Genetics department, please give name of contact in Genetics:



When to test, when not to test



Is there clinical utility?

CLINICAL UTILITY (Please indicate how testing will impact patient care – tick below)

Patient management (determining therapeutic decisions and/or clinical investigations and/or surveillance programme).

Patient, parents, or adult relative reproductive decision making.

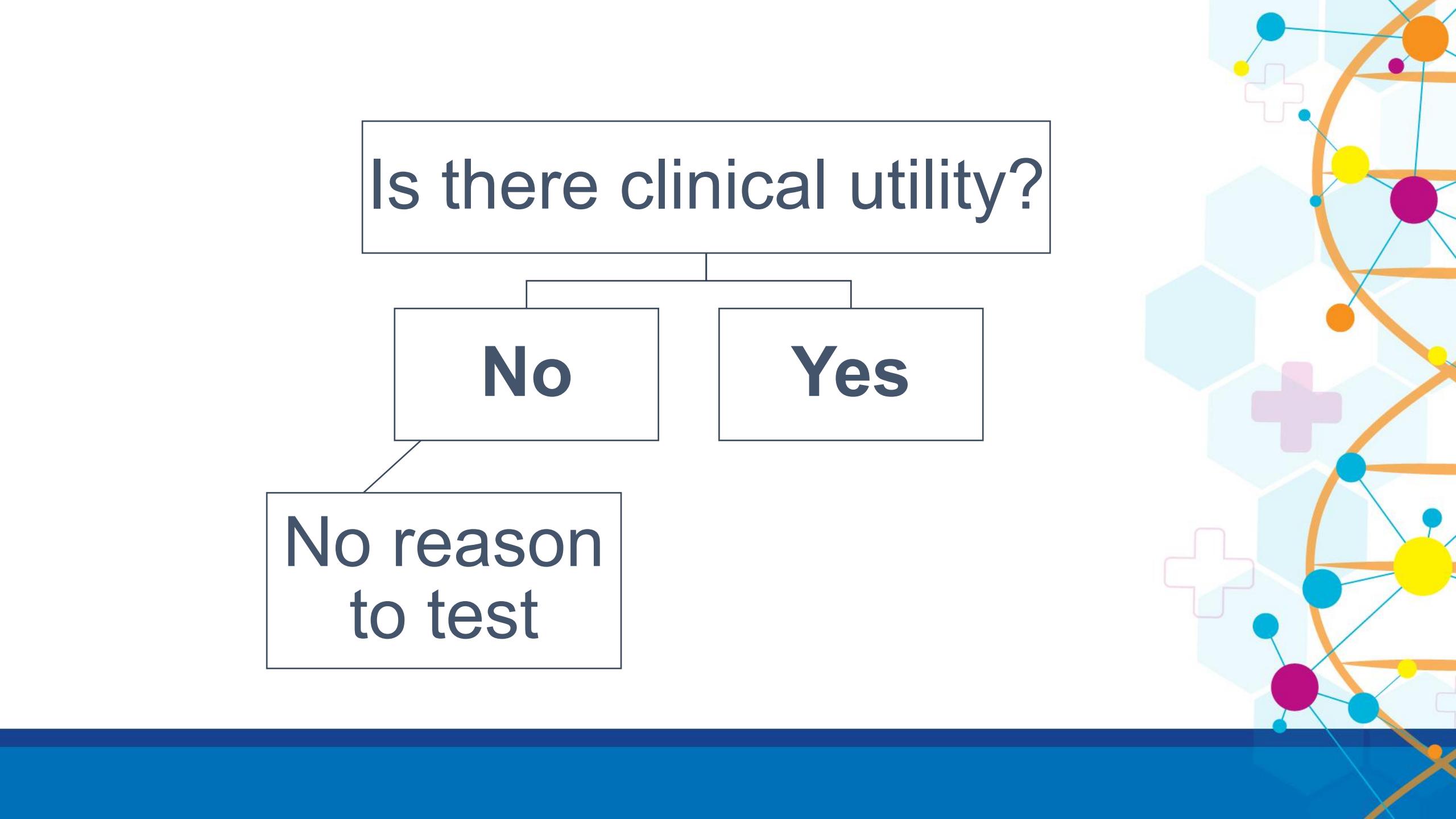
Unaffected relatives are seeking predictive testing.

CLINICAL UTILITY (Please provide additional information with other relevant clinical information above)

- Patient management (determining therapeutic decisions and/or clinical investigations and/or surveillance programme)
- Patient, parents or adult relative reproductive decision making
- Unaffected relatives are seeking predictive testing

Is there a clinical utility? If yes, tick box:

- Patient management (determining therapeutic decisions and/or clinical investigations and/or surveillance programme).
- Patient, parents, or adult relative reproductive decision making.
- Unaffected relatives are seeking predictive testing.



Is there clinical utility?

No

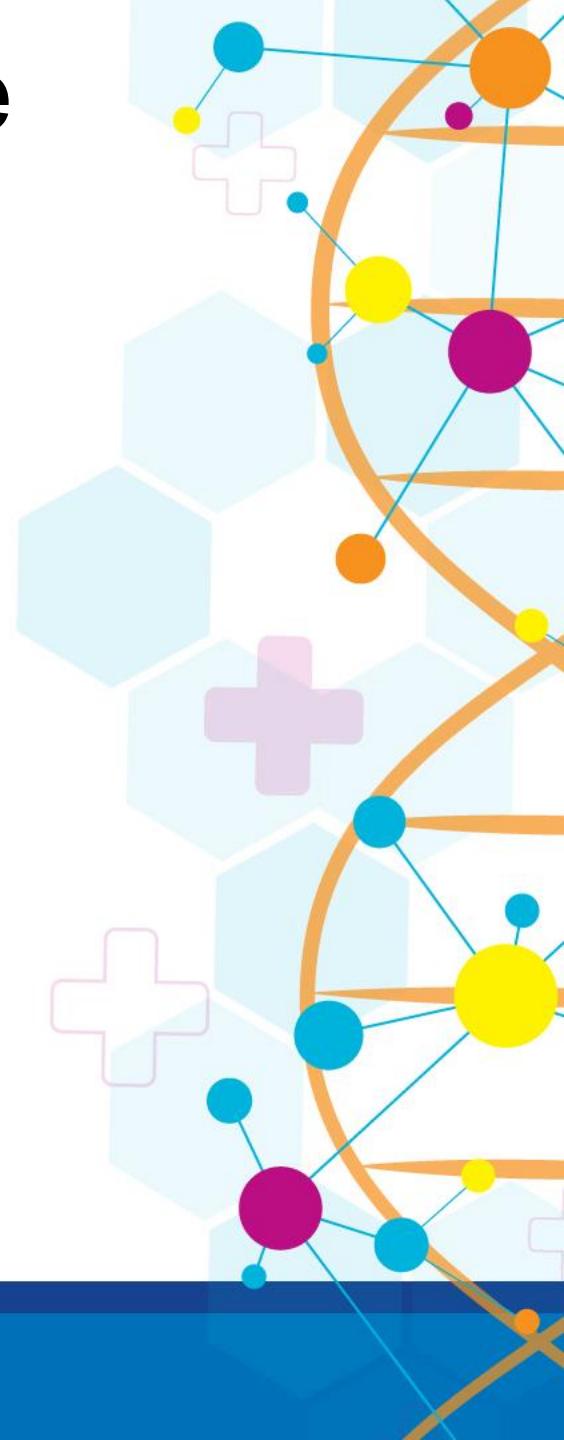
Yes

No reason
to test

Tests should **only** be requested where there is clear evidence that a result is highly likely to change **clinical management** of the patient or their family.

This includes:

- determining clinical investigations
- therapeutic/treatment decisions or strategies
- enrolling in nationally approved surveillance programme
- informing or supporting reproductive choices



Reasons not to test

- **Not eligible** – e.g. Mild Learning Difficulties or normal baby after normal testing during pregnancy or low risk sibling (tests will simply be rejected if they don't meet eligibility)
- **Low utility** - Clinically obvious diagnosis and/or no family implications and will not impact management



Reasons not to test

- Not testing people where there is no clinical reason to do so means patients that **will benefit** will get tested, diagnosed and treated in a clinically relevant time frame



Why are the changes happening now?

- These steps will **increase the diagnostic yield** of the test and helps to get a diagnosis **promptly**.
- **Reduce the need for costly and time-consuming re-tests**
- **Reduce total tests**
- **Adapt to meet the population needs and the financial budget**

Where can I get further information/guidance?



Resources

- <https://panelapp.genomicsengland.co.uk/> Panel app
- [NHS England » National genomic test directory NGTD](#)
- [Human Phenotype Ontology](#) –HPO website
- [NHS Guide to HPO](#) – short video
- [Essential updates in genomic testing « Central and South Genomics](#) –resources, training and updates

Please see end of presentation for more guidance on using all of the above

General enquiries

West Midlands Regional Genetics Laboratory

For general enquiries: BWC.genetics.lab@nhs.net

Telephone: 0121 335 8036

Oxford Genetics Laboratory

For general enquiries: dutyscientist.oxfordgenetics@ouh.nhs.uk

Telephone: 01865 226001

Wessex Regional Genetics Laboratory

For general enquiries: shc-tr.WRGLdutyscientist@nhs.net

Telephone: 01722 429080

Central and South Genomics: GMSAAdmin@uhb.nhs.uk



Websites for more information/forms

- Birmingham [Referral forms](#)
- Oxford [Referral forms](#)
- Wessex [Referral forms](#)
- Central and South Genomics [For Healthcare Professionals](#)
[Central and South Genomics](#)



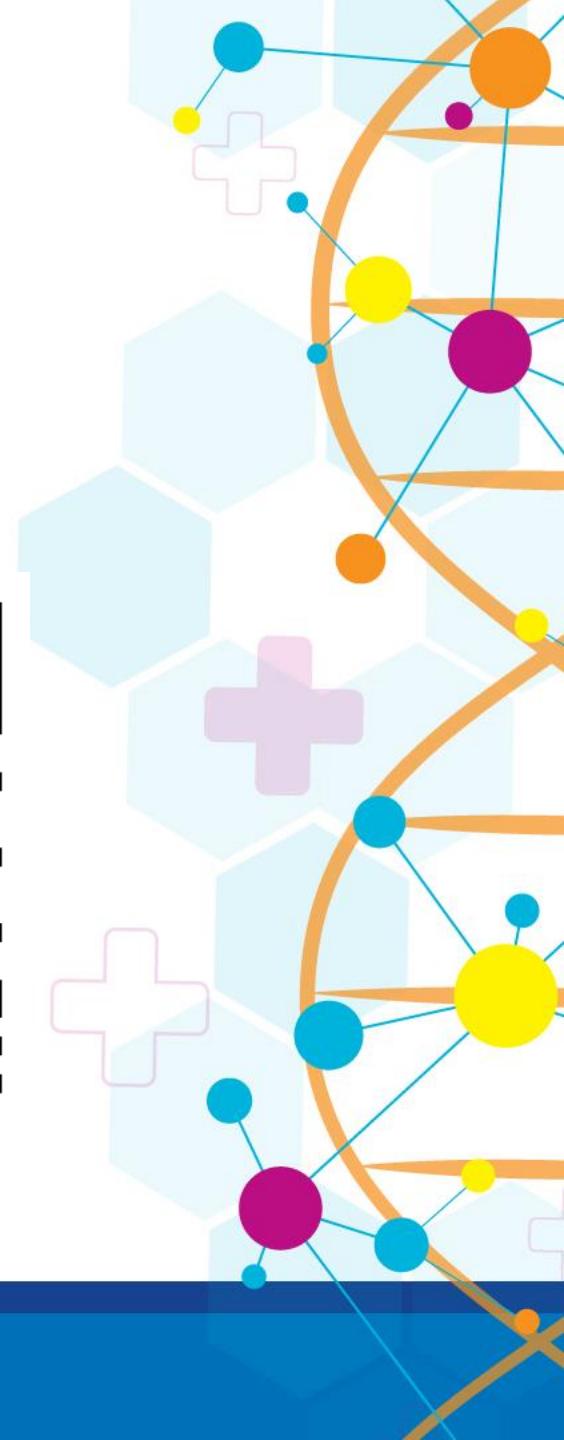
Conclusion

- *The primary test for rare disease clinical indications is whole genome sequencing (WGS)*
- *Test request forms should provide detailed information on how the patient/family meet the eligibility criteria for the test indication that is being requested including HPO*
- *Tests should only be requested where there is clear evidence that a result is highly likely to change clinical management for the patient or their family.*
- *Not testing in situations with limited or no clinical utility will ensure patients and families who will benefit from genomic testing have equitable access to it within a clinically relevant timeframe*

Thank you

Questions?

Please take a moment to complete
the post-event survey >
Vicki.geddes@uhb.nhs.uk



Upcoming Webinars

[Right Test, Right Time: Essential Updates in Genomic Testing](#)



[Right Test, Right Time: Genomics for Neurologists](#)

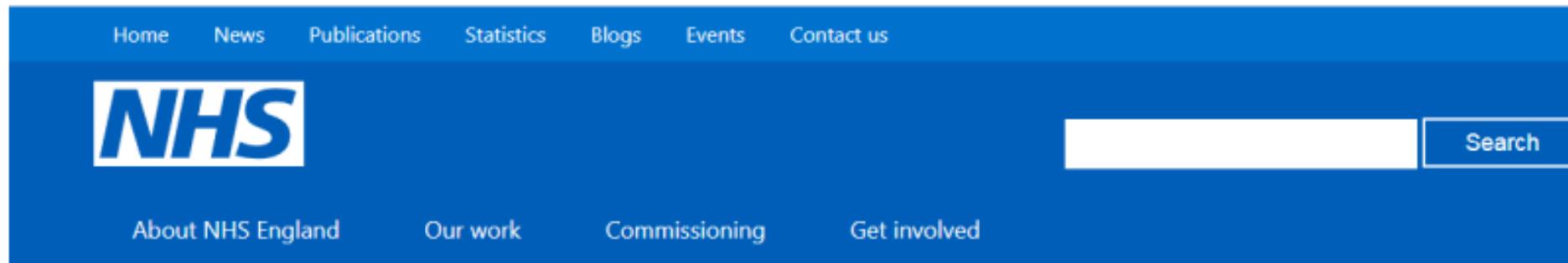


Further Guidance

- National Test Directory
- WGS Guide
- HPO Guide



NTD Eligibility Criteria



The image shows the header of the NHS England website. It features a blue navigation bar with white text. From left to right, the menu items are: Home, News, Publications, Statistics, Blogs, Events, and Contact us. Below the menu is the NHS logo. To the right of the logo is a search bar with a white input field and a blue 'Search' button. Underneath the search bar are four links: 'About NHS England', 'Our work', 'Commissioning', and 'Get involved'. The background of the header is a light blue color with a subtle hexagonal grid pattern.

National Genomic Test Directory

Document first published: 3 August 2018

Page updated: 20 March 2019

Topic: Commissioning, Genomics, Specialised commissioning

Publication type: Guidance

The 2019/2020 National Genomic Test Directory specifies which genomic tests are commissioned by the NHS in England, the technology by which they are available, and the patients who will be eligible to access to a test. The 2019/2020 National Genomic Test Directory for rare and inherited disorders and cancer can be accessed below.



- [NHS England » National genomic test directory](#)



National genomic test directory

Document first: 3 August 2018

published:

Page updated: 11 February

2025

Topic: Commissioning,
Genomics,
Specialised
commissioning

Publication type: Guidance

The national genomic test directory specifies which genomic tests are commissioned by the NHS in England, the technology by which they are available, and the patients who will be eligible to access to a test. The national genomic test directory for rare and inherited disorders and cancer can be accessed below.

If you have any questions about the genomic testing available in your area, please contact your local genomic laboratory hub.

Document[National genomic test directory for rare and inherited disease](#)

Microsoft Excel 171 KB

Summary

The national genomic test directory for rare and inherited diseases specifies the genomic tests commissioned by the NHS in England for rare and inherited disorders, the technology by which they are available, and the patients who will be eligible to access to a test.

Version 7.1 published 2 Jan 2025.

Document[Rare and inherited disease eligibility criteria](#)

PDF 3 MB 412 pages

Summary

This eligibility criteria document supplements the national genomic test directory by setting out which patients should be considered for testing under that indication, and the requesting specialties is a list of the clinical specialties who would be expected to request the test.

Version 7.1 published 2 Jan 2025.

- Available at;
<https://www.england.nhs.uk/publication/national-genomic-test-directories/>
- Describes **which genetics tests are commissioned** by NHS in England (WGS and non-WGS)
- Details the **technology used** for each test, e.g. WGS
- Lists **who can request** each test
- Gives **patient eligibility criteria** for each test
- 2 categories: **Rare Disease** and Cancer
- 2 formats: PDF or Excel spreadsheet (can filter test based on technology)
- **NB: Make sure you access the latest version online**

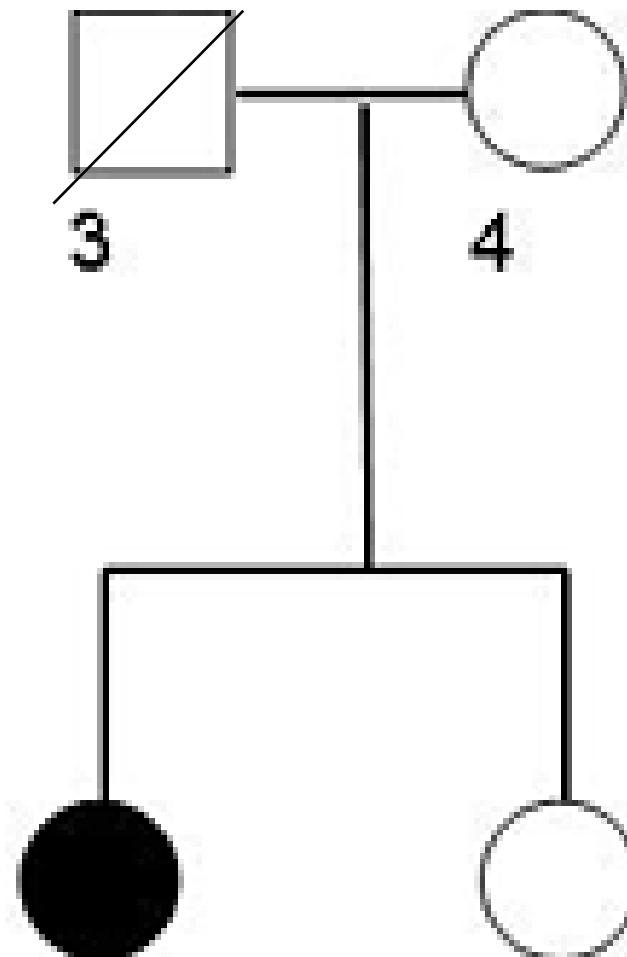
WGS success steps

WGS : 7 Steps for Success	
1	Arrange a trio for paediatric cases to help actionable results. Without DNA from both parents, scientists cannot fully analyse any rare variants that are detected
2	Store the patient sample correctly (eg blood samples in purple top EDTA). DNA will be damaged if it's not in the correct tube. Formalin will damage tissue DNA too.
3	Complete all forms electronically . Handwritten forms will be rejected.
4	Include the patient's ethnicity to improve accuracy of results. We need to compare results with population specific data to understand if variant could be causative
5	Explain why your patients meet the eligibility criteria & include ALL HPO terms (Human Phenotype Ontology). Even those that might not seem relevant, otherwise we might miss relevant findings. Find HPO terms on the HPO website & select terms that start with HPO.
6	Email the Record of Discussion & Test Order Form to the <u>relevant regional laboratory</u> . We can't start testing until we have ALL forms for family members being tested. One Record of Discussion per person. One Test Order Form per duo or trio.
7	Look out for an email from the WGS team. We'll email you if there is a problem. Testing will be cancelled after 16 weeks if the referral isn't complete.

- Copy of WGS : 7 steps

WGS – steps to success

- In pediatric cases trio testing remains the best route for diagnosis: Proband/Mother/Father
- Where possible the test request should be made as a **trio** (proband and both parents) or **duo** (proband and one parent)
- Clearly label all 3 samples
- Obviously a trio or duo are not always possible – record on the TOF where and why it isn't possible



Use Human Phenotype Ontology (HPO) terms on referral forms

- Universal standardised terminology



The Human Phenotype Ontology

The Human Phenotype Ontology (HPO) provides a standardized vocabulary of phenotypic abnormalities encountered in human disease. Each term in the HPO describes a phenotypic abnormality, such as *Renal vesicular defect*. The HPO is currently being developed using the medical literature, DiGeorge, DECIPHER, and OMIM. HPO currently contains over 18,000 terms and over 750,000 annotations to hereditary diseases. The HPO project and others have developed software for phenotype-driven differential diagnostics, genomic diagnostics, and translational research. The HPO is a flagship product of the Monarch Initiative, an NIH-supported international consortium dedicated to semantic integration of biomedical and model organism data with the ultimate goal of improving biomedical research. The HPO, as part of the Monarch Initiative, is a central component of one of the 13 driver projects in the Global Alliance for Genomics and Health (GA4GH) strategic roadmap.

Learn More About HPO

News & Updates

2.0.4 Medical Actions September 18, 2024

2.0.2 API Breaking Changes June 7, 2024

April 2024 HPO release April 19, 2024

[View All News](#)

- [Human Phenotype Ontology](#) – website
- [NHS Guide to HPO](#) – short video



Case Study

- 2-year-old patient
- Birthweight was above the 90th percentile
- Port wine stain on forehead
- Large tongue
- Unusually folded earlobes



HPO Website

Port wine stain
on forehead



The Human Phenotype Ontology

The Human Phenotype Ontology (HPO) provides a standardized vocabulary of phenotypic abnormalities encountered in human disease. Each term in the HPO describes a phenotypic abnormality, such as [Atrial septal defect](#). The HPO is currently being developed using the medical literature, Orphanet, DECIPHER, and OMIM. HPO currently contains over 18,000 terms and over 156,000 annotations to hereditary diseases. The HPO project and others have developed software for phenotype-driven differential diagnostics, genomic diagnostics, and translational research. The HPO is a flagship product of the [Monarch Initiative](#), an NIH-supported international consortium dedicated to semantic integration of biomedical and model organism data with the ultimate goal of improving biomedical research. The HPO, as a part of the Monarch Initiative, is a central component of one of the [13 driver projects](#) in the [Global Alliance for Genomics and Health \(GA4GH\) strategic roadmap](#).

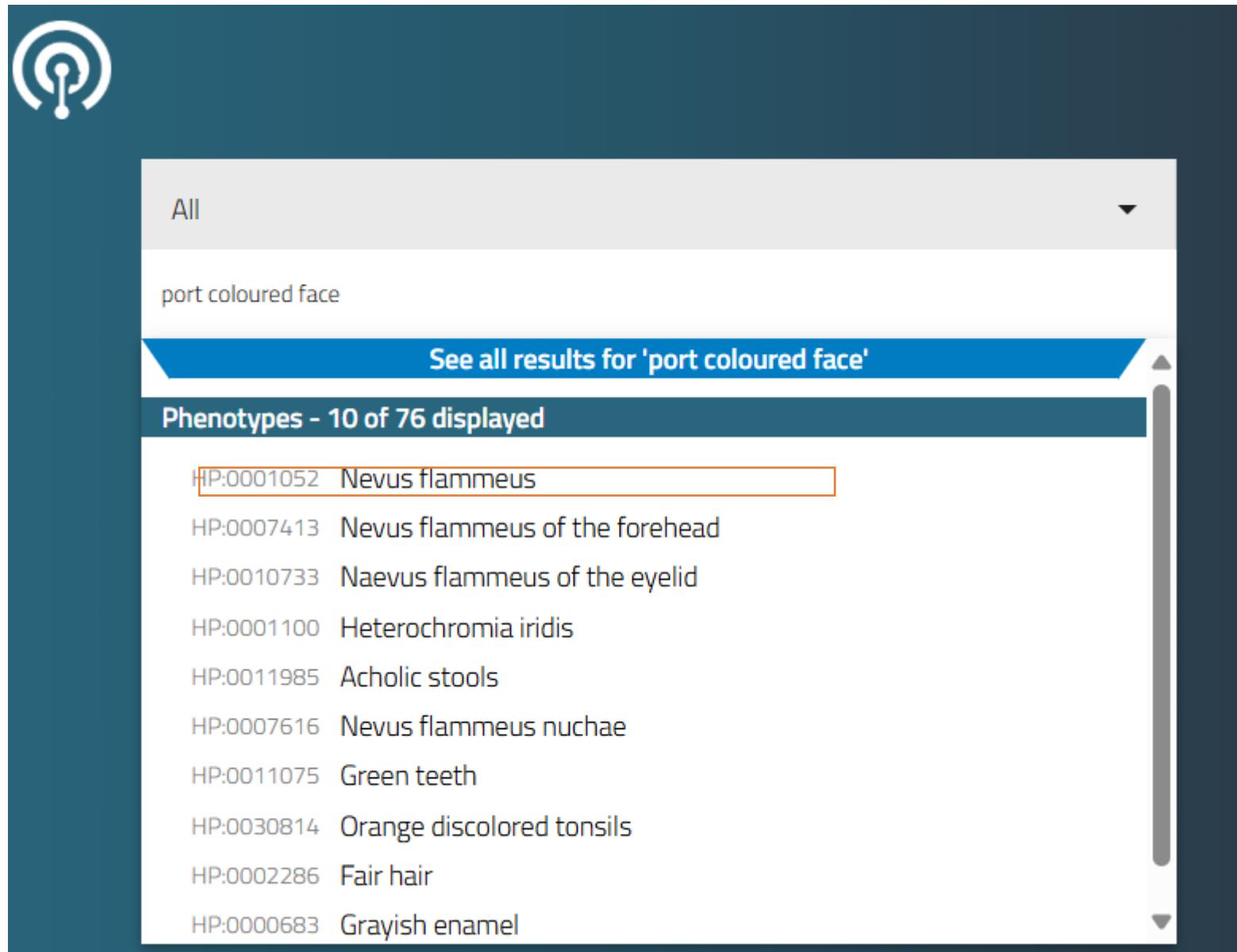
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Port coloured face



The image shows a screenshot of a web-based search interface for the Human Phenotype Ontology (HPO). The search term 'port coloured face' is entered into the search bar. Below the search bar, a blue button reads 'See all results for 'port coloured face''. A dark blue header bar displays the text 'Phenotypes - 10 of 76 displayed'. The main content area lists ten phenotype entries, each consisting of a code (e.g., HP:0001052) and a description (e.g., 'Nevus flammeus'). The first item in the list, 'Nevus flammeus', is highlighted with a red border.

HP:0001052	Nevus flammeus
HP:0007413	Nevus flammeus of the forehead
HP:0010733	Naevus flammeus of the eyelid
HP:0001100	Heterochromia iridis
HP:0011985	Acholic stools
HP:0007616	Nevus flammeus nuchae
HP:0011075	Green teeth
HP:0030814	Orange discolored tonsils
HP:0002286	Fair hair
HP:0000683	Grayish enamel

Purple/red face mark above eyebrows

All

purple/red face mark above eyebrow

Search Results For "purple/red face mark above eyebrow"

Not seeing what you're looking for? [Contribute a term](#)

< Term Results [76] Disease Results [0] Gene Re >

.....

Filter

Term Identifier	Term Name	Matching String	Synonym Match
HP:0001052	Nevus flammeus		Yes
HP:0007413	Nevus flammeus of the forehead		Yes

Reddy purpley stain on child's head

All

reddy purpley stain on child's head

[See all results for 'reddy purpley stain on child's head'](#)

Phenotypes - 10 of 100 displayed

- HP:6001220 Positive Blastomyces dermatitidis sputum fungal **stain**
- HP:0007413 Nevus flammeus of the forehead
- HP:0010733 Naevus flammeus of the eyelid
- HP:0007616 Nevus flammeus nuchae
- HP:6000583 Positive synovial fluid gram **stain**
- HP:0001052 Nevus flammeus
- HP:0025156 Dependency **on** intravenous nutrition**on**
- HP:0025812 Seborrheic scales **on** scalp
- HP:0007516 Redundant skin **on** fingers
- HP:0030874 Oxygen desaturation **on** exertion**on**

Referral Form

HPO term (copy and paste) =
Nevus flammeus of the
forehead HP:0007413

Nevus flammeus of the
forehead HP:0007413



Panel app

<https://nhsgms-panelapp.genomicsengland.co.uk/panels>

Genomics England PanelApp

A crowdsourcing tool to allow gene panels to be shared, downloaded, viewed and evaluated by the Scientific Community

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PanelApp and the NHS Genomic Medicine Service (GMS) Panels Resource

Here on the PanelApp website you can find all the gene panels that relate to genomic tests listed in the [NHS National Genomic Test Directory](#), as well as the virtual gene panels that were used in the [100,000 Genomes Project](#). Not all the genes and panels shown here are available to be chosen for testing as part of the NHS Genomic Medicine Service (GMS).

NHS users should view the set of genes/panels that are available for testing as part of the GMS on the [NHS GMS Panels Resource](#) website. These are the panels that can be chosen when requesting a genomic test. The green (diagnostic evidence level) genes shown on these panels are those that are analysed as part of the diagnostic pathway*. The panel versions are periodically updated from PanelApp.

