NHS
Central and South
Genomics

Right Test, Right Time: Essential Updates in Genomic Testing

Please complete pre-survey:

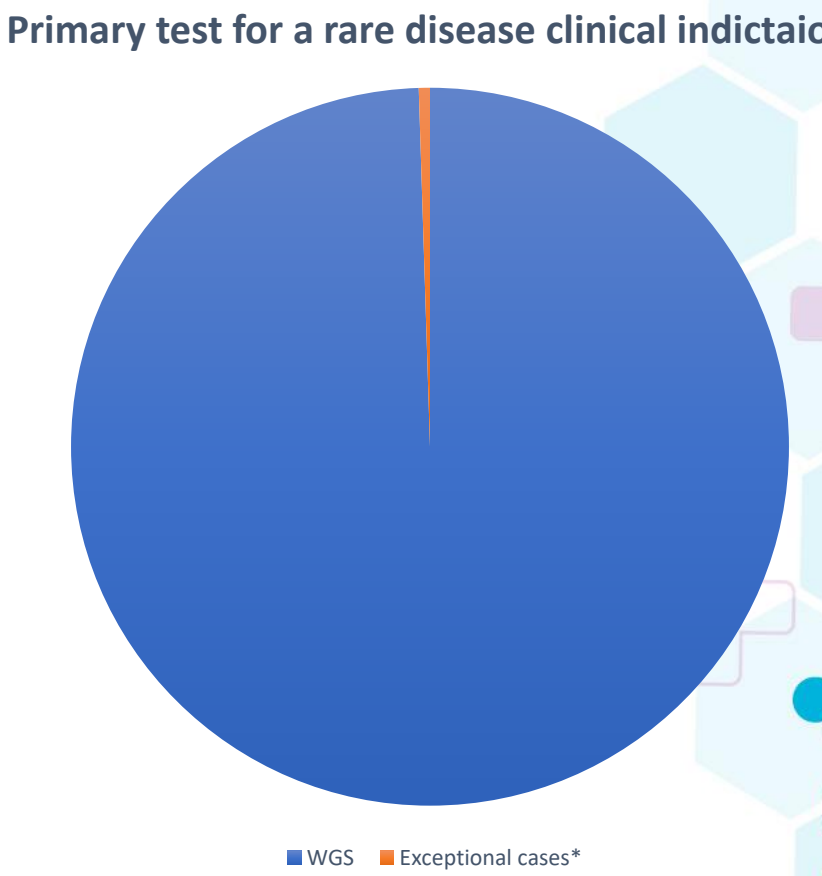
Objectives

- 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

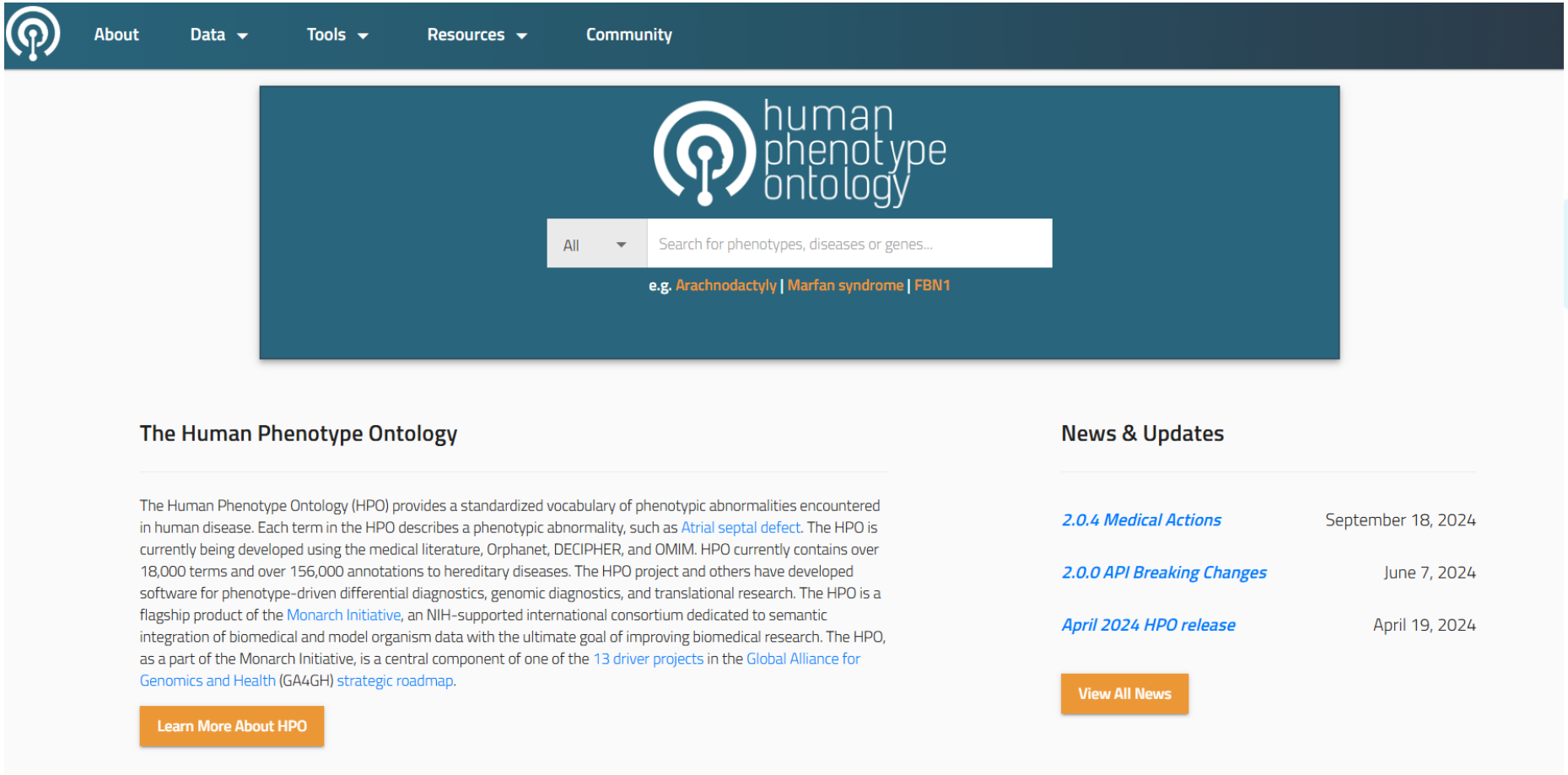
The main changes

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

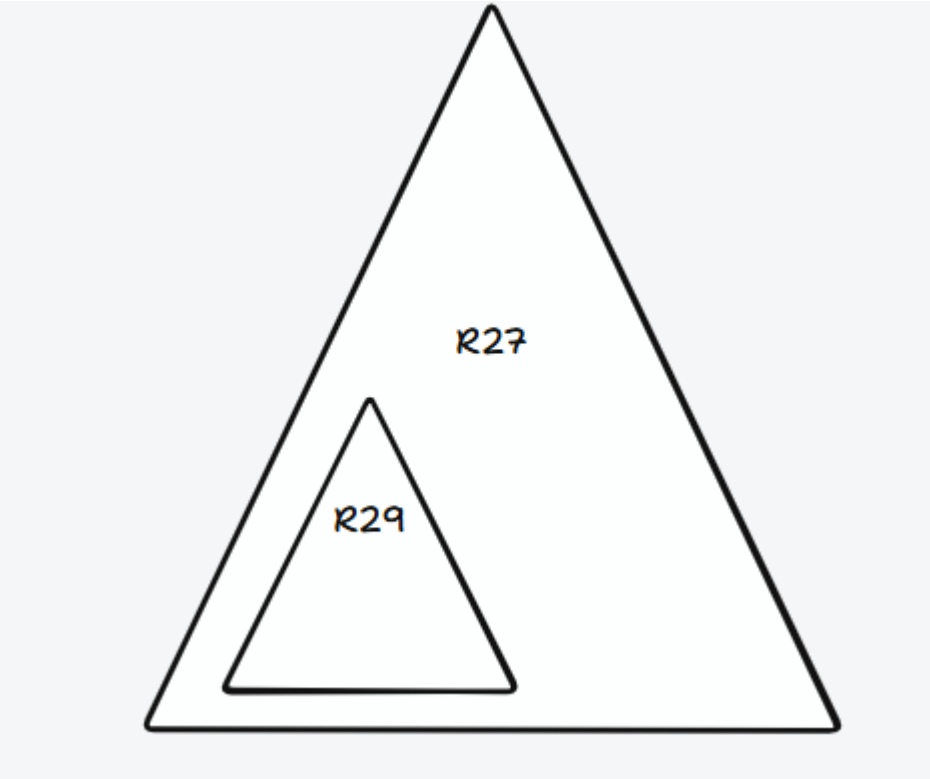


2. HPO



[HPO website]

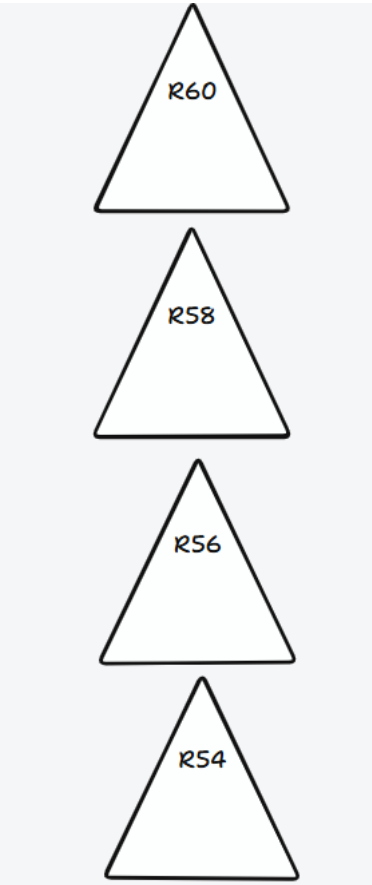
3. No R29 – amalgamation



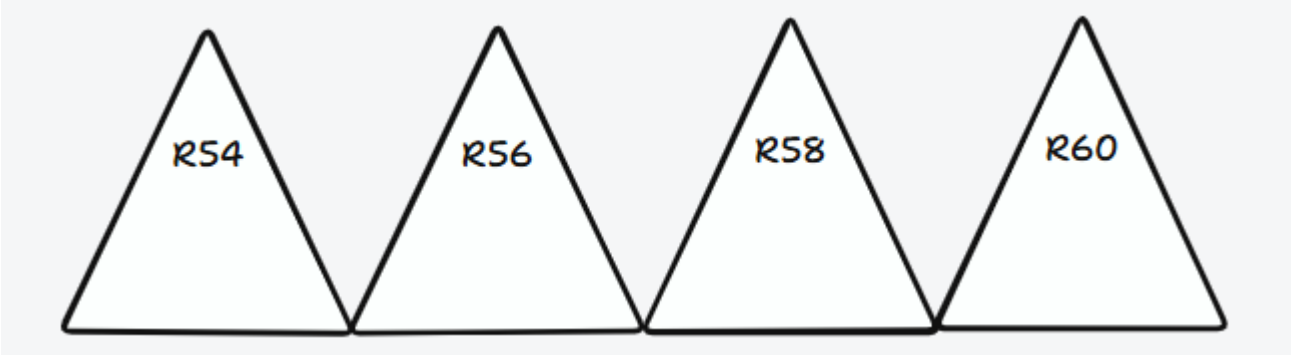
- **R27 (Paediatric disorders)**
- **This panel contains these 13 panels:**
- DDG2P
- **Intellectual disability (R29)**
- Early onset or syndromic epilepsy
- Likely inborn error of metabolism
- Skeletal dysplasia
- Monogenic hearing loss
- Paediatric disorders - additional genes
- Clefting
- Neurological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

West Midlands Genomics Laboratory
Birmingham Women's Hospital
Mindelsohn Way | Edgbaston | Birmingham | B15 2TG
Tel: 0121 335 8036
Email: bwc.genetics.lab@nhs.net

NHS Central & South Genomic Laboratory Hub

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)

- ☐ 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

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

If a specimen is known to present an i
labeled **'DANGER OF INFECTION'** and

Sample type: (please circle)	Venou
	Tissue (p
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): 15mm² in tissue culture medium or sterile saline.
Fetal or postnatal tissue biopsy (e.g. skin, muscle, cord): 5mm² 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 **Email:** bwc.genetics.lab@nhs.net	**SHIPPING ADDRESS** Birmingham Women's Hospital Genetics Laboratory – Ground Floor Mindelsohn Way | Edgbaston | Birmingham | B15 2TG	**LABORATORY OPENING TIMES** Monday to Friday: 07:00 - 18:00 Saturday: 09:00 - 14:00

TRIAGE DETAILS (FOR INTERNAL GLH LAB USE ONLY)

Priority	Prenatal	Urgent	Presymptomatic	Priority	Routine
B number indication			**Culture**	Culture	No culture
Additional information					
D Indication 1			**Reason**		
			Reason		
			Reason		

	Time

West Midlands

Oxford Genetics Laboratories
Oxford University Hospitals NHS Foundation Trust
The Churchill Hospital
Oxford OX3 7LE
Admin office: 01865 226001
Email: dutyscientist.oxfordgenetics@ouh.nhs.uk

NHS
Central & South
Genomic Laboratory Hub
Oxford Genetics Laboratories

PATIENT DETAILS *(Printed label if available)*

Family name:

First name(s):

Date of birth: Sex*: M F U
*Please state if karyotypic and/or phenotypic sex differ from given sex.

NHS number:

Hospital number:

Address: Ethnic Origin:

Case / Family number:

Postcode: NHS Private Please supply the name and address for

REFERRER DETAILS

Referring Clinician: Job Title:

Consultant:
(if different from above. Mandatory)

Contact Name:
(if different from above)

Additional copies to:

Email for report: Tel No:
(PTO for more information)

Hospital address:

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:

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)**

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.

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

(Clinician – Please tick which tube is required)
For Chromosome analysis, Fluorescence In Situ Hybridization (FISH): **Blood in LITHIUM HEPARIN (1-5ml)**

For gene sequencing, specific mutation tests, dosage, SNP array: **Blood in EDTA (1-5ml)**

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.

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. **Name of person taking sample:**

For Lab use only: Date of receipt: Initials: Sample cond./Vol.:

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): Investigation(s):

W

DNA loc:
In before? Initials
Date of receipt:

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

UKAS MEDICAL 9665

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 speciality
NHS England / Other NHS / Private (Address for invoicing if not NHS England):	Additional copies to (name, address, speciality)		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, parents, 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)
- ☐ Diagnostic targeted test (R240)
- ☐ Predictive (R242)
- ☐ Segregation (R375)
- ☐ Carrier (R244)
- ☐ 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:

NHS 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 s
classification and packaging instruction P650, to arrive
post, courier service or hospital transport) and **within 4**
be clearly labelled '**PATHOLOGICAL SAMPLE FOR D**

Opening hours are 9 am - 5.30 pm Mon - Fri; please inf
weekend or bank holiday, or of anything sent by courie
s 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 epi[illegible] history

D) Personal history of telangiectasia, and refractory or [illegible] transfusions) *

*Pulmonary AVM only if confirmed by cross sectional imagin[illegible] therapeutic angiography/surgery. Do not diagnose if only su[illegible] ("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

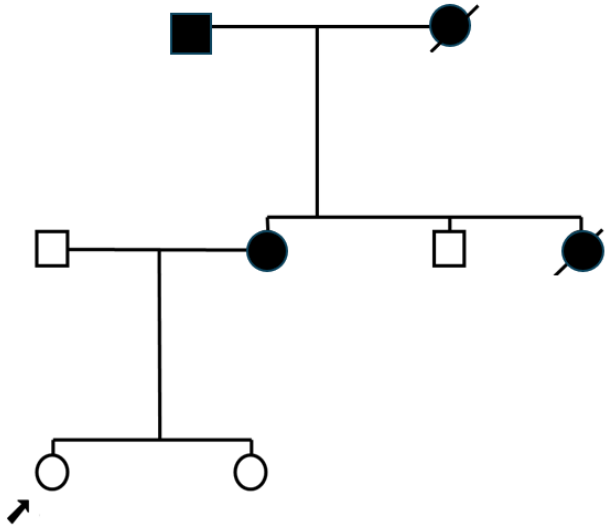
Is the patient or their partner pregnant?	If YES: gestation at sampling by scan?

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

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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)

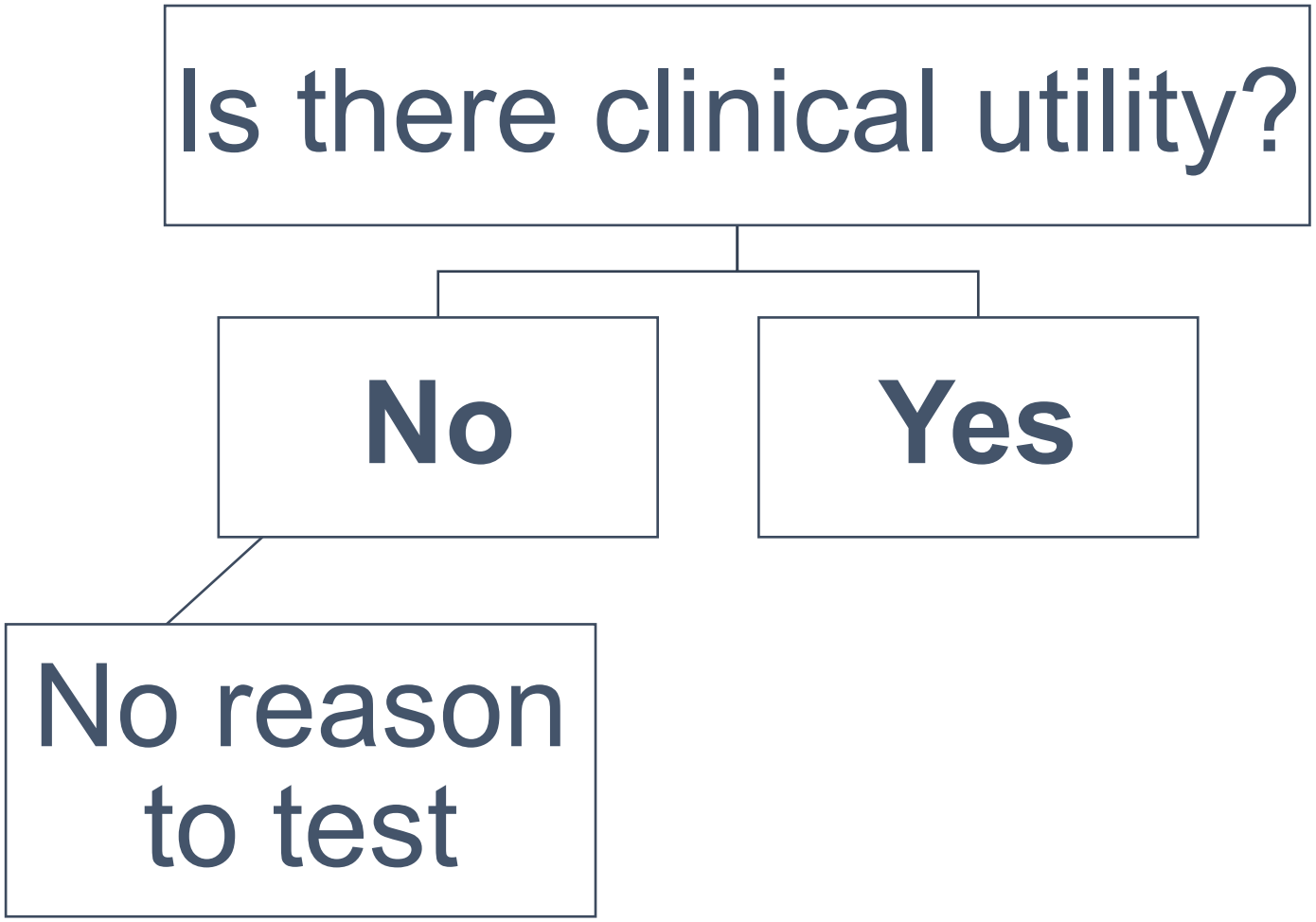
- ☐ Patient management (determining therapeutic decisions and/or clinical investigations and/or surveillance programme).
- ☐ Patient, parents, or adult relative reproductive decision making.
- ☐ Unaffected relatives are seeking predictive testing.

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

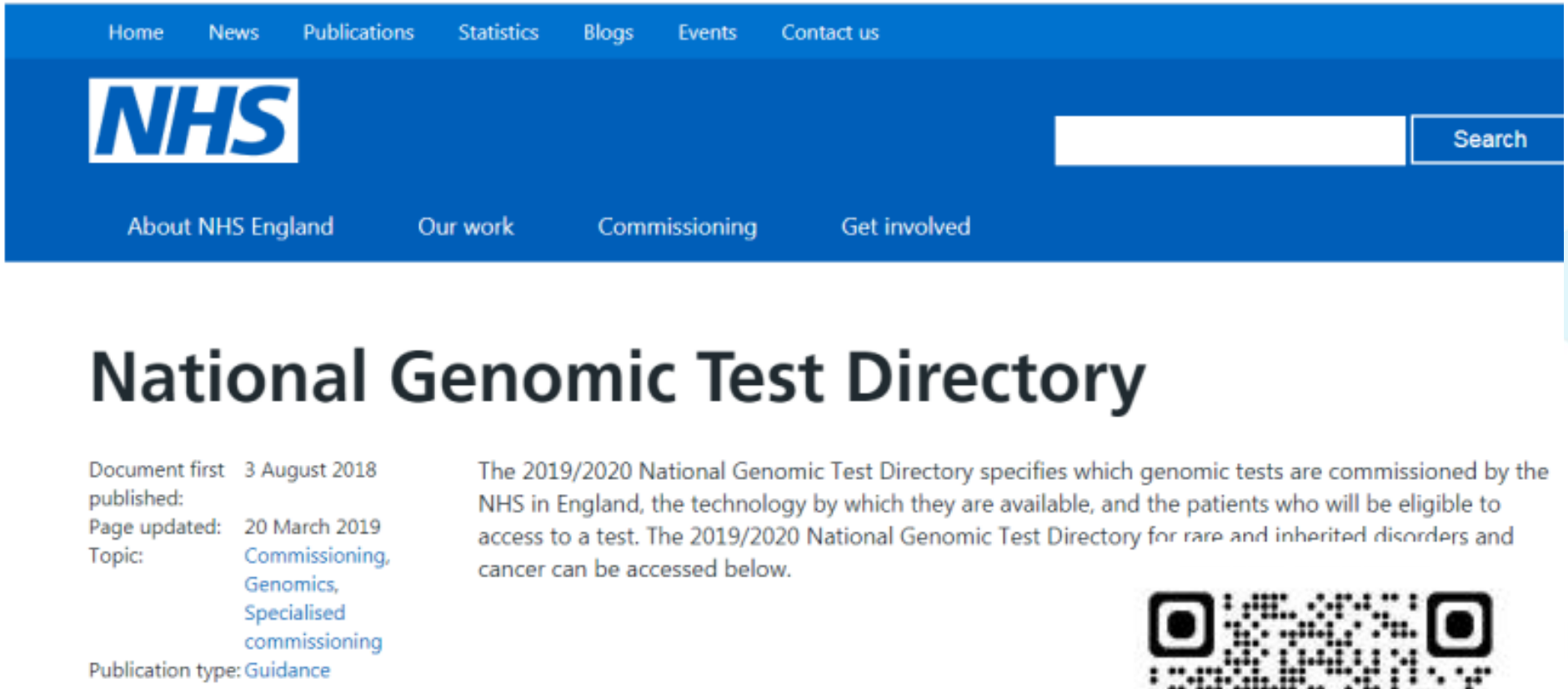
Right Test, Right Time: Essential Updates in Genomic Testing

Right Test, Right Time: Genomics for Neurologists

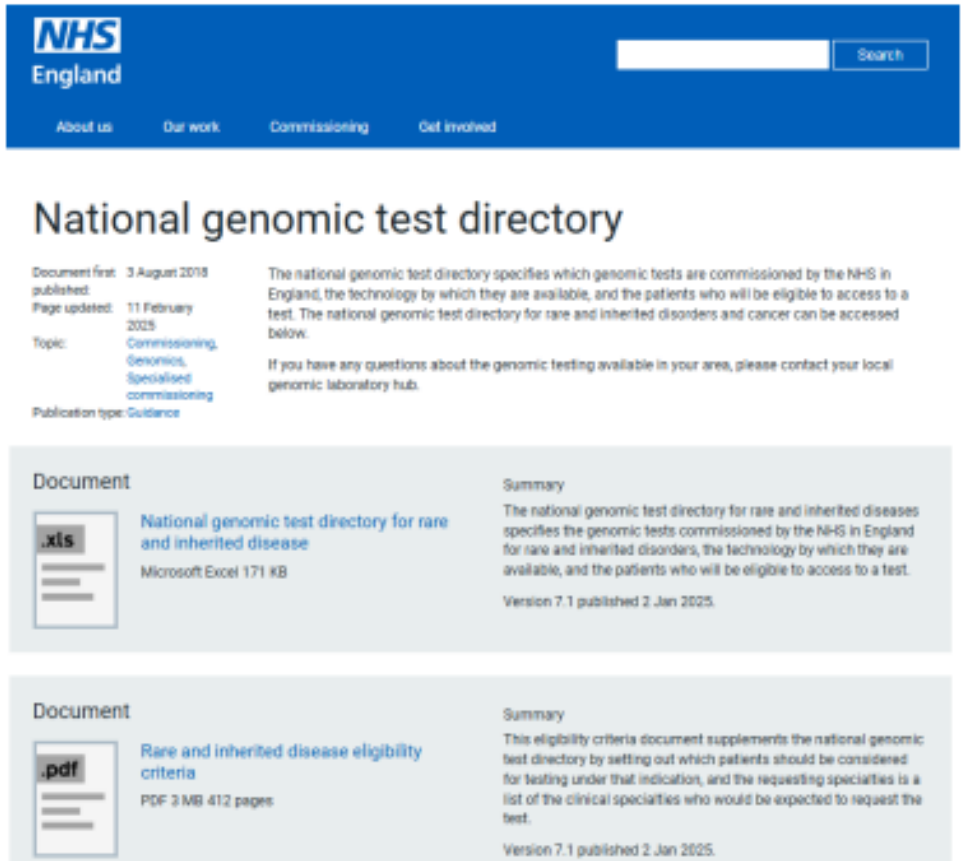
Further Guidance

- National Test Directory
- WGS Guide
- HPO Guide

NTD Eligibility Criteria



- [NHS England » National genomic test directory]



- 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

NHS
Central and South
Genomics

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 relevant regional laboratory. 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.

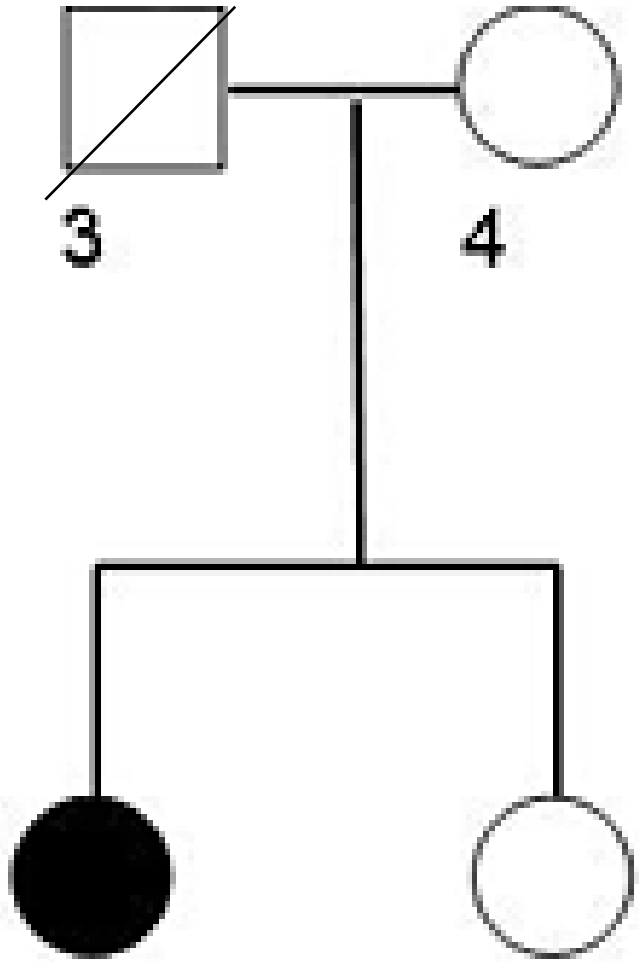
- Copy of WGS : 7 steps

WGS – steps to success

- In pediatric cases trio testing remains the best route for diagnosis: Proband/Mother/Father

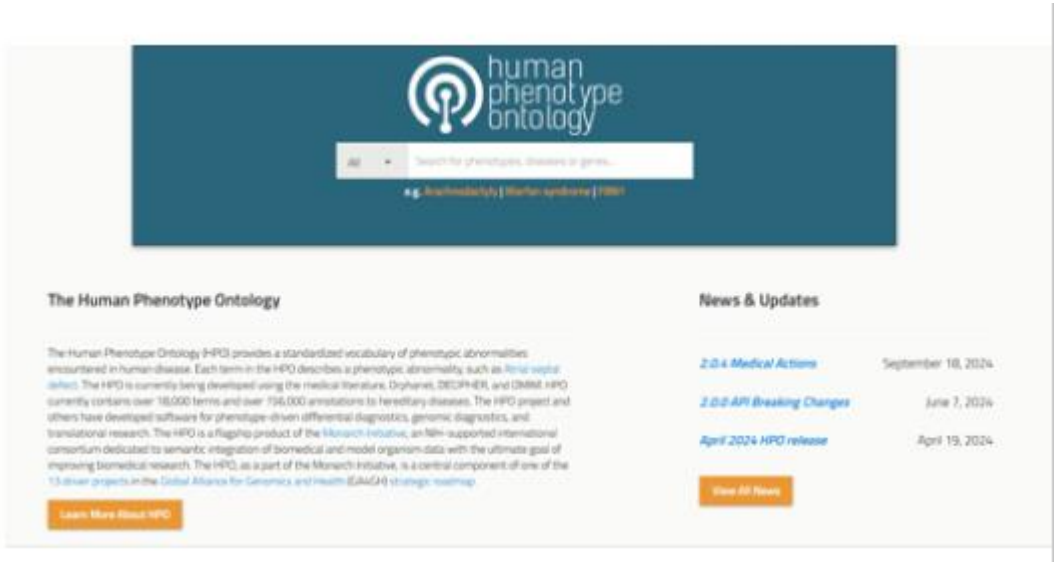
- Where possible the test request should be made as a **trio** (proband and both parents) or **duo** (proband and one parent)
- Clearly label all 3 samples

- 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



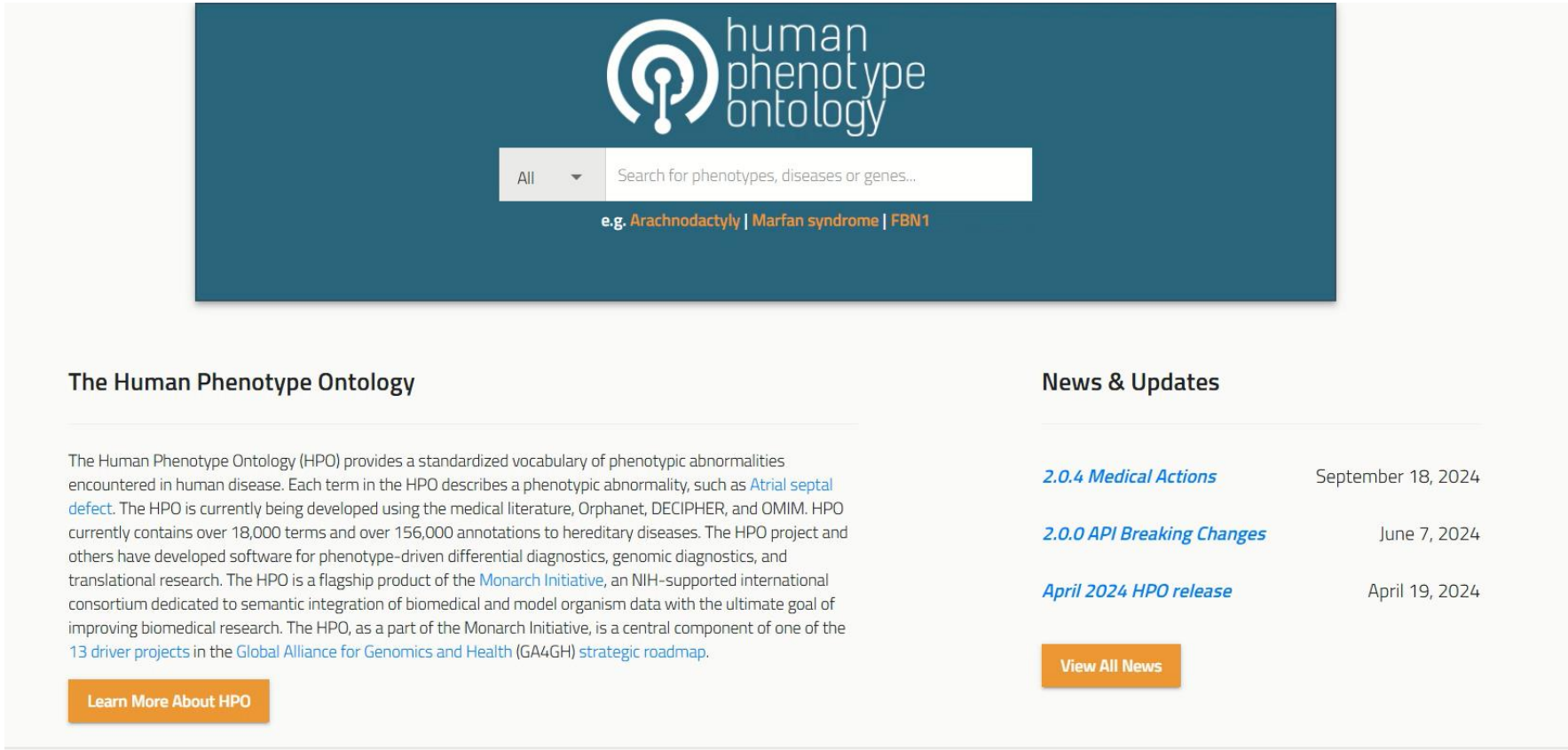
- Human Phenotype Ontology –website
- NHS Guide to HPO – short video

Case Study

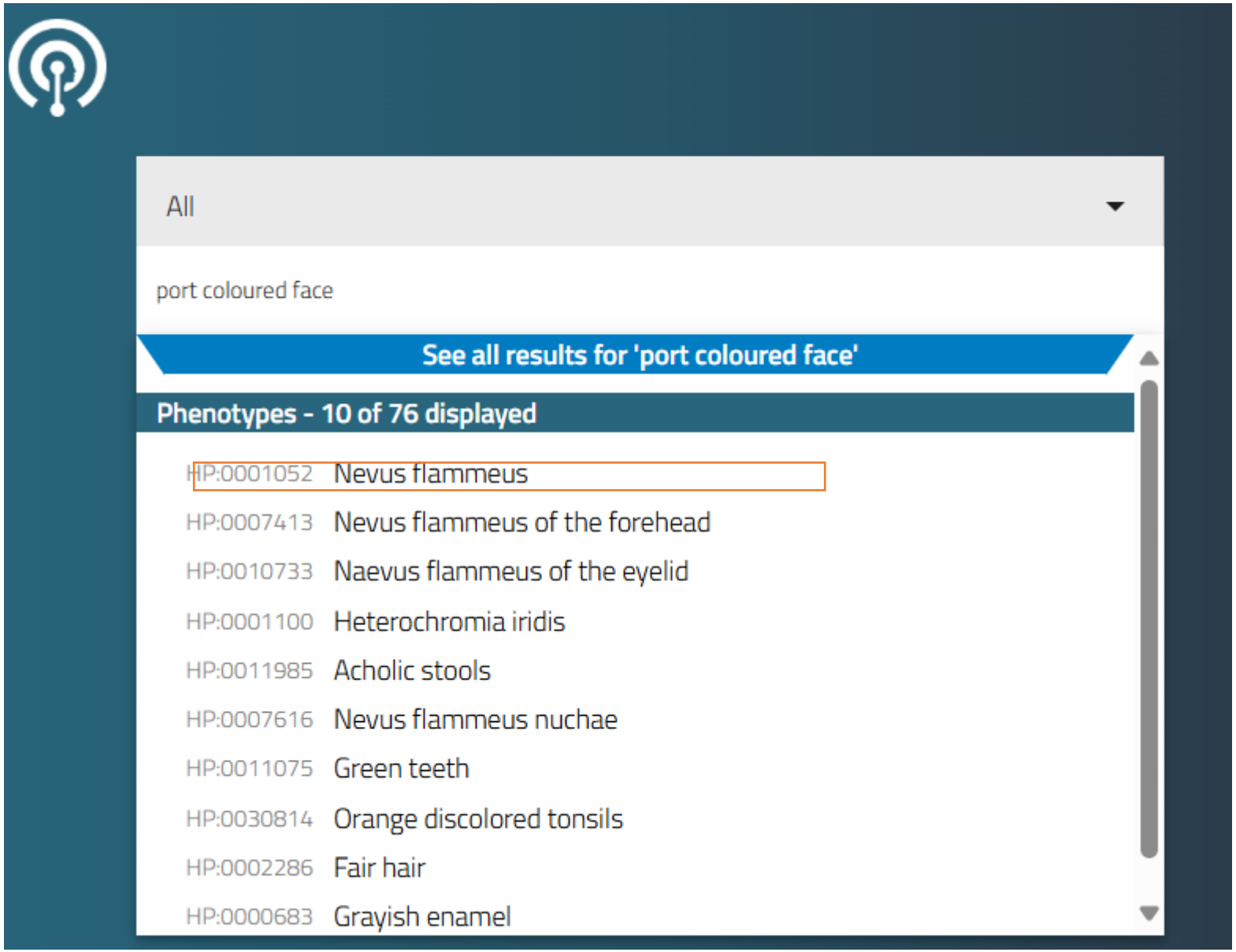
- 2-year-old patient
- Birthweight was above the 90th percentile
- Port wine stain on forehead
- Large tongue
- Unusually folded earlobes

HPO Website

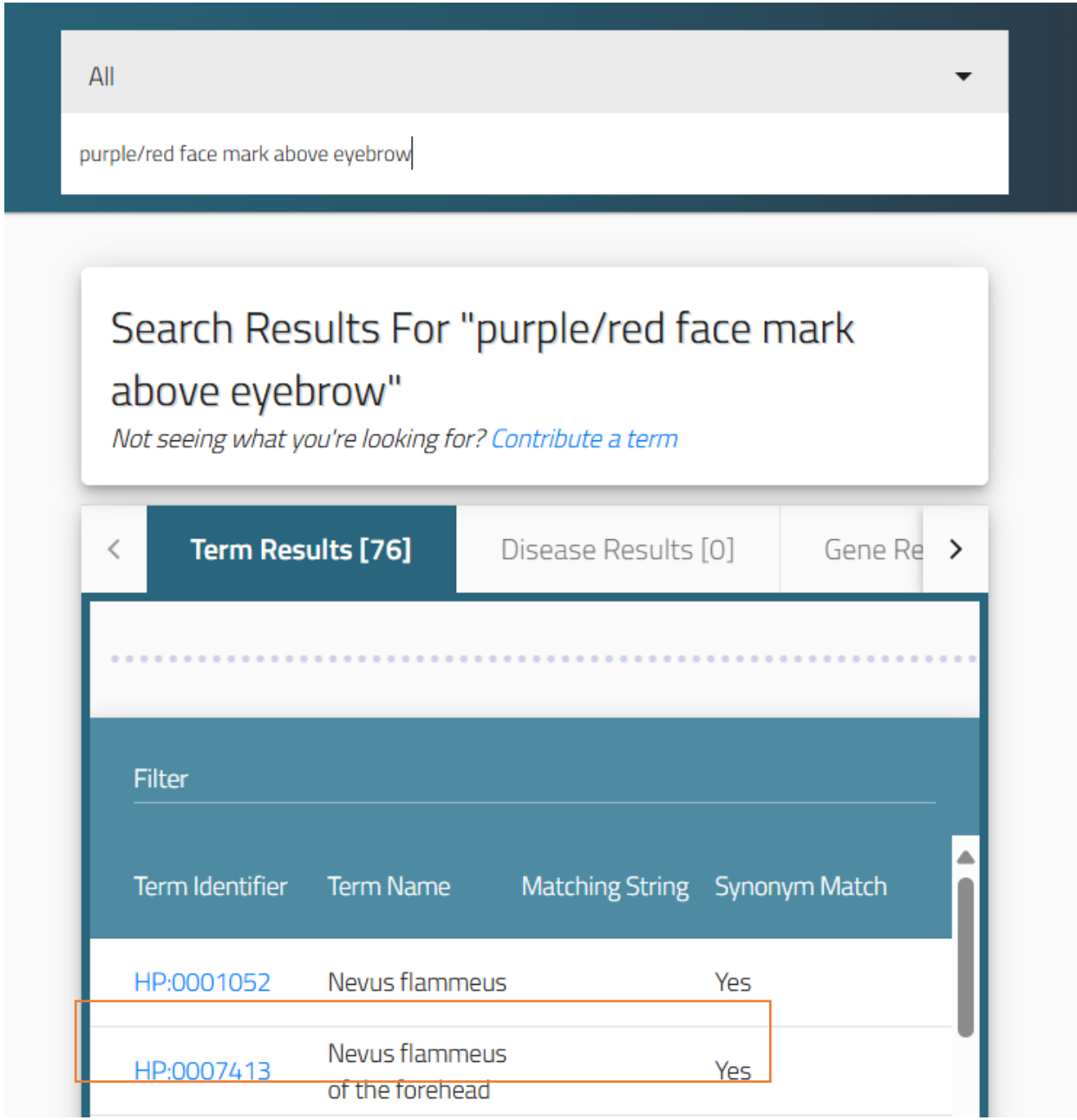
Port wine stain on forehead



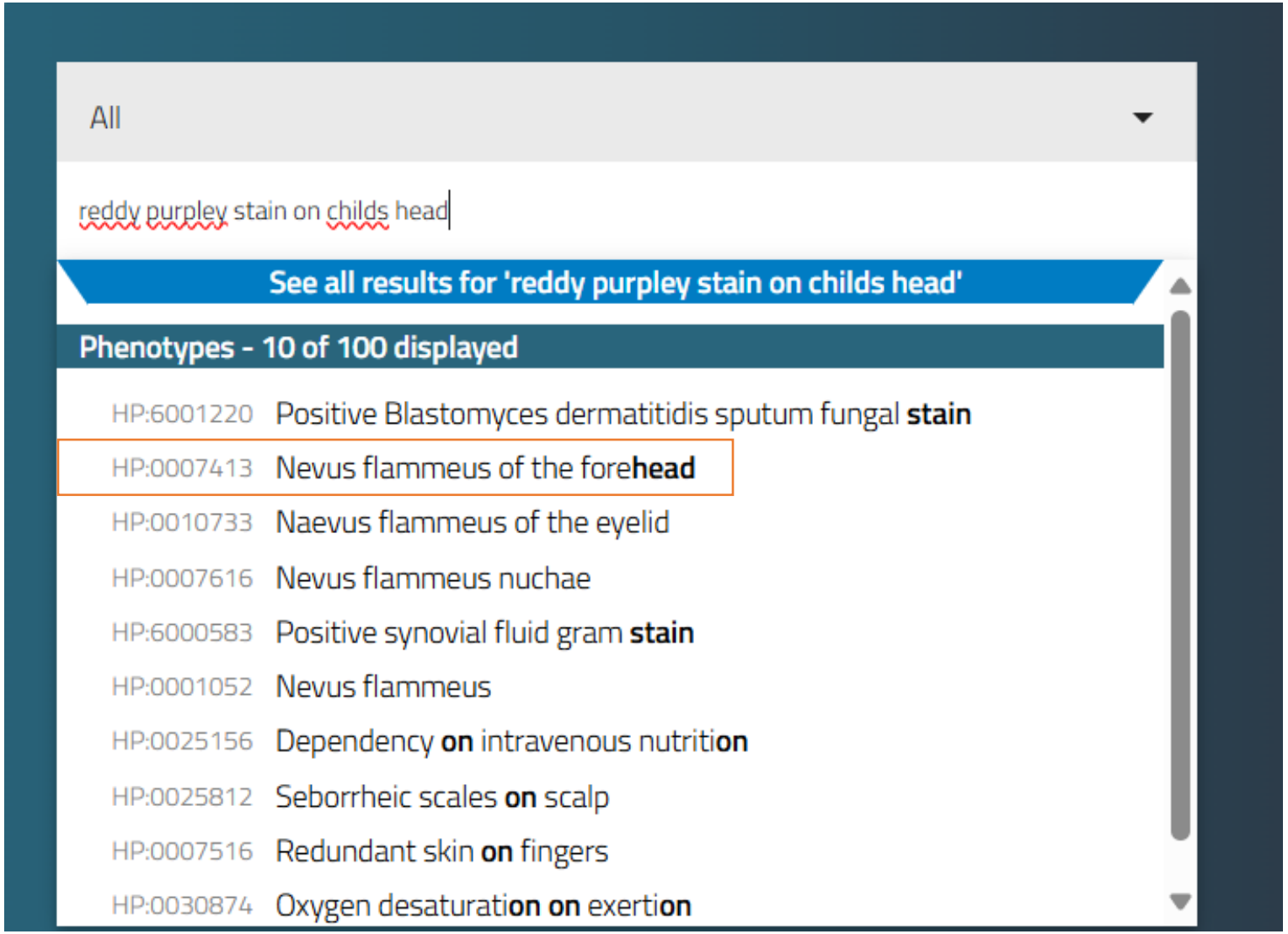
Port coloured face



Purple/red face mark above eyebrows



Reddy purpley stain on child's head



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