18th International Conference of the
Research Institute of Ophthalmology

RIO 2026

22 -23 JANUARY 2026
ROYAL MAXIM PALACE KEMPINSKI, CAIRO

Genomics for ophthalmologists

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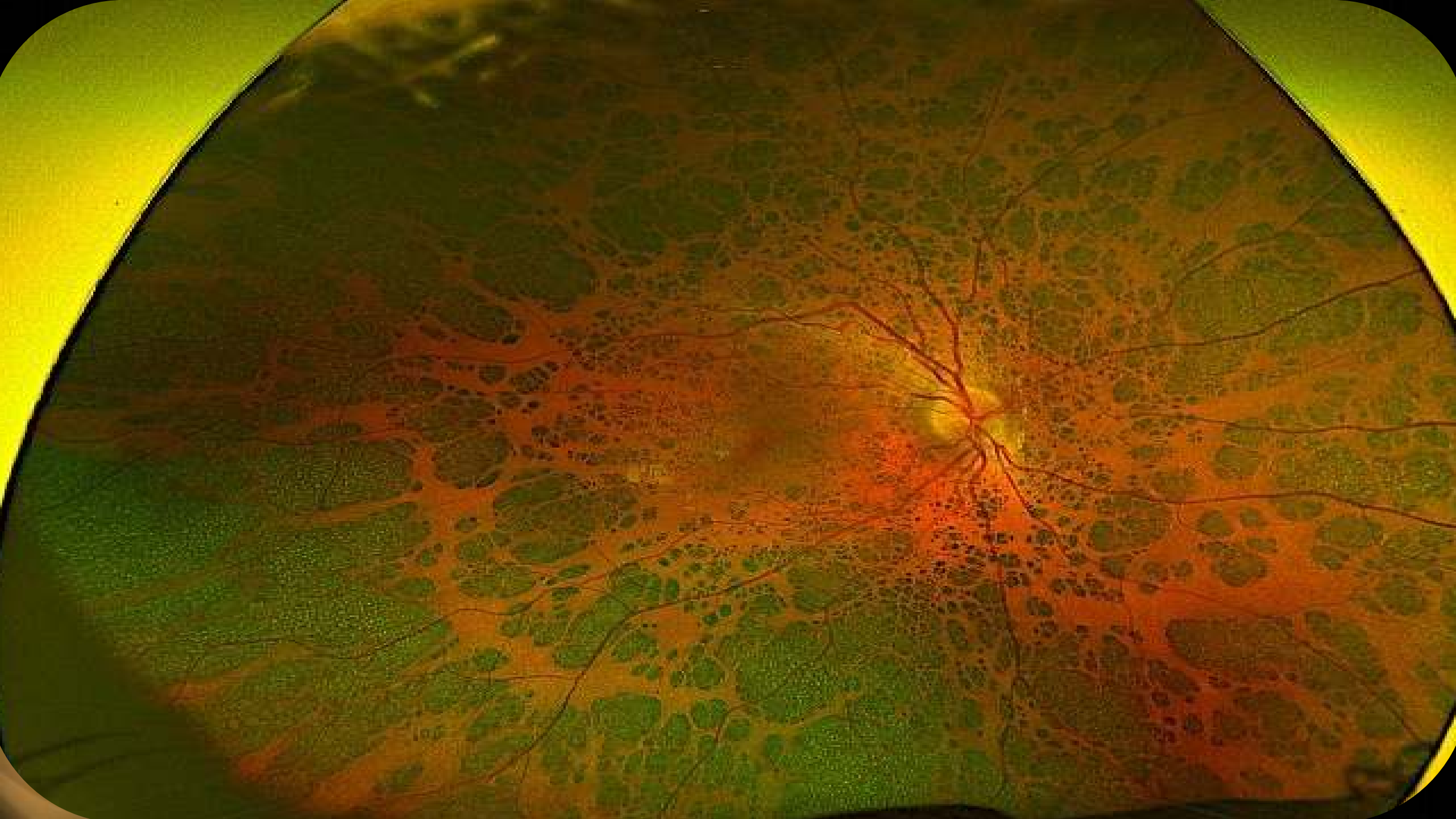
Southampton UK


All Ophthalmologists should do genetic testing



Genomics – for ophthalmologists

1	2	3
Which test?	Consenting	Results

Improve the care of children

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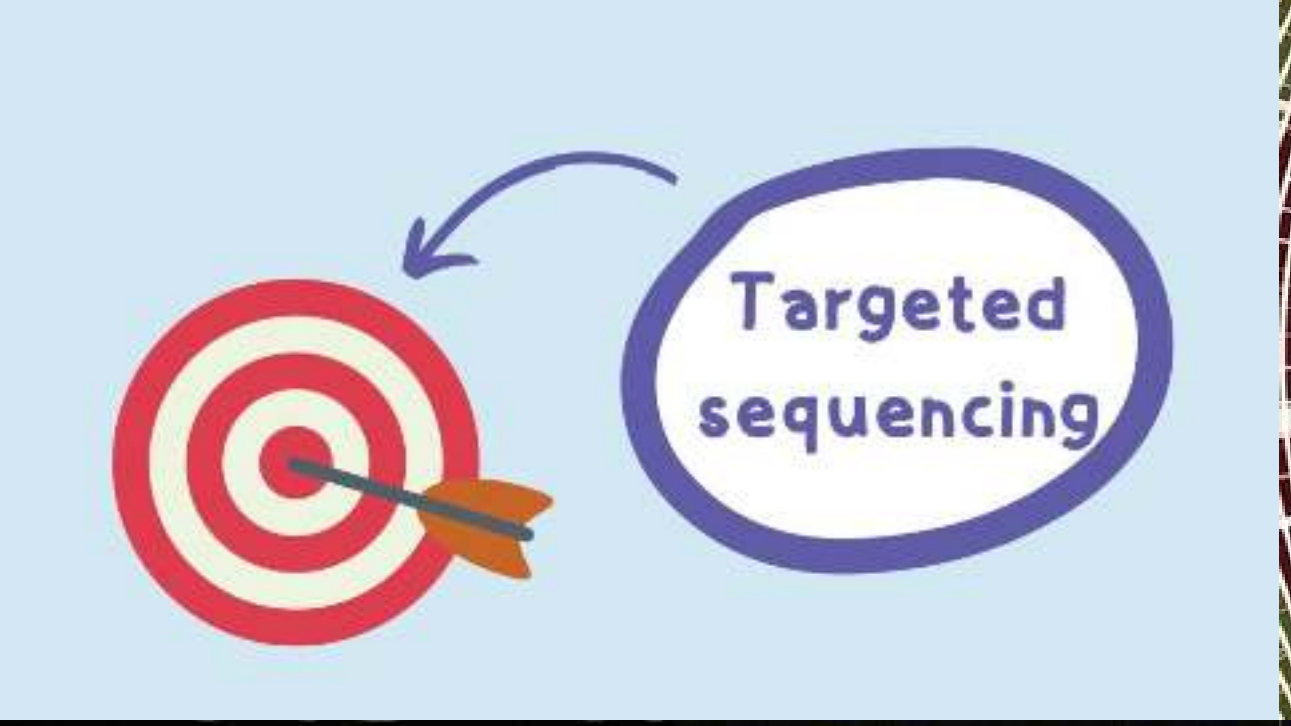
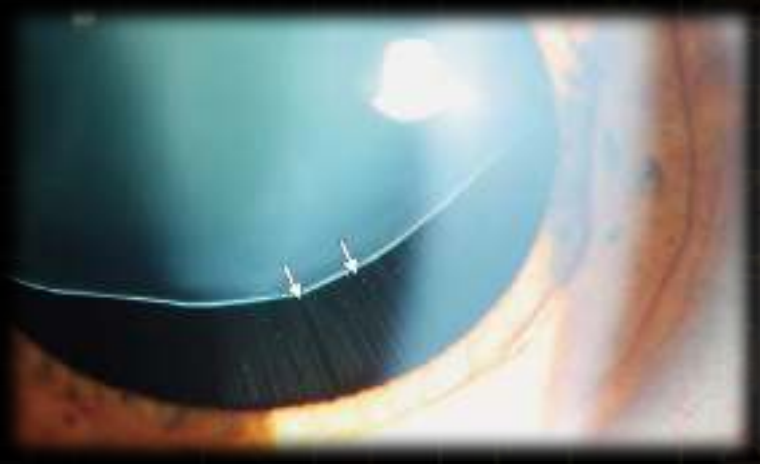

Targeted
sequencing


Whole Genome
Sequencing


CGH, ARRAY-CGH


Gene
Panel



- Do you target FBN1?
- Who do you test?
- Which gene panel?

GMS Panels | Panels | Genes and Entities

FBN1

fibrillin 1

OMIM: 134797

See this entity in PanelApp

Panel	Mode of inheritance
Filter panels	
Green in DDG2P Component of the following Super Panels: - Paediatric disorders Signed-off version 6.0	BOTH monoallelic and biallelic, autosomal or pseudoautosomal
Green in Ehlers Danlos syndrome with a likely monogenic cause R-numbers: **R101** Signed-off version 4.0	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
Green in Fetal anomalies R-numbers: **R21, R412** Signed-off version 6.0	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
Green in Lipodystrophy - childhood onset R-numbers: **R158** Signed-off version 4.0	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
Green in Pneumothorax - familial R-numbers: **R190** Signed-off version 3.0	MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown
Green in Rare syndromic craniosynostosis or isolated multisuture synostosis R-numbers: **R100** Signed-off version 6.0	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
Green in Skeletal dysplasia Component of the following Super Panels: - Paediatric disorders R-numbers: **R104** Signed-off version 8.0	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted

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Think about logistics carefully – (make friends with clinical genetics colleagues)

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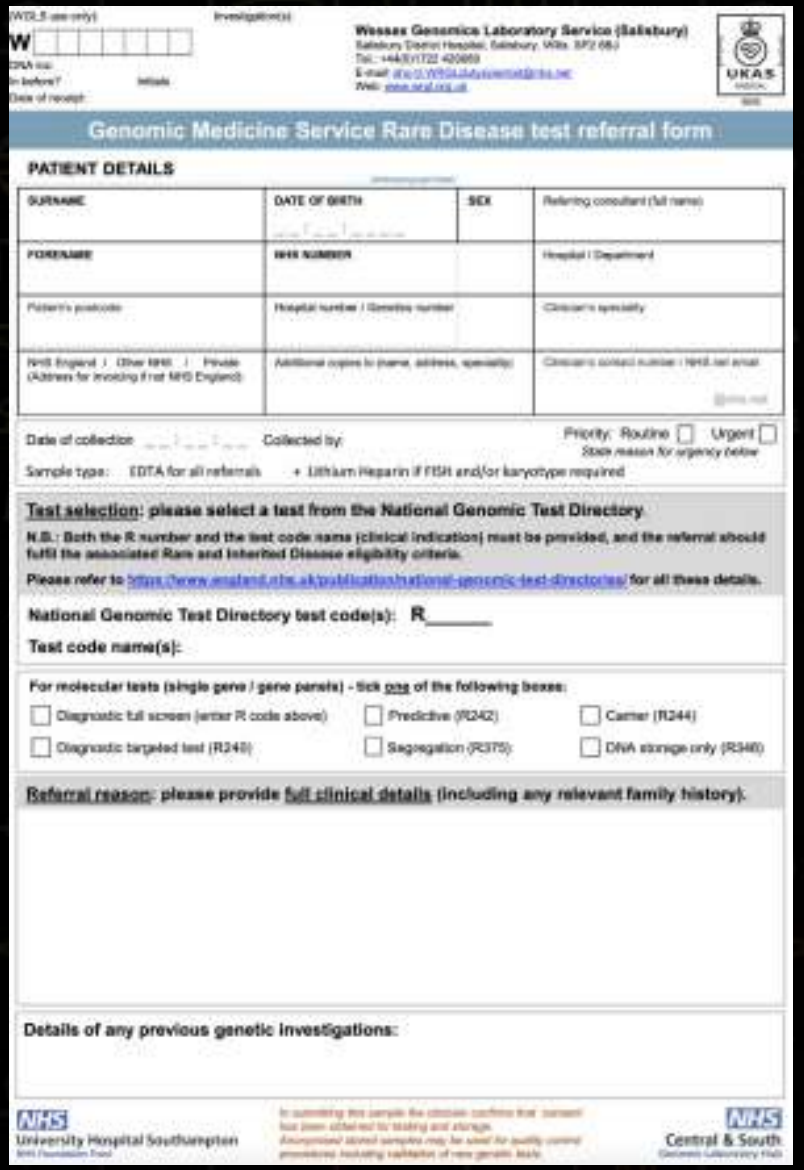
What about in my clinics?



Wessex Genomics Laboratory Service (Salisbury)

Genomic Medicine Service Rare Disease test referral form

PATIENT DETAILS

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

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

N.B.: Both the R number and the test code name (clinical indication) must be provided, and the referral should fulfil the associated Rare and Inherited Disease eligibility criteria.

Please refer to https://www.england.nhs.uk/publication/national-genomic-test-directories/ for all these details.

National Genomic Test Directory test code(s): R_____

Test code name(s):

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 (R243) ☐ Segregation (R375) ☐ DNA storage only (R346)

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

Details of any previous genetic investigations:

NHS University Hospital Southampton

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 processes including validation of new genetic tests.

NHS Central & South Genomics Laboratory Hub

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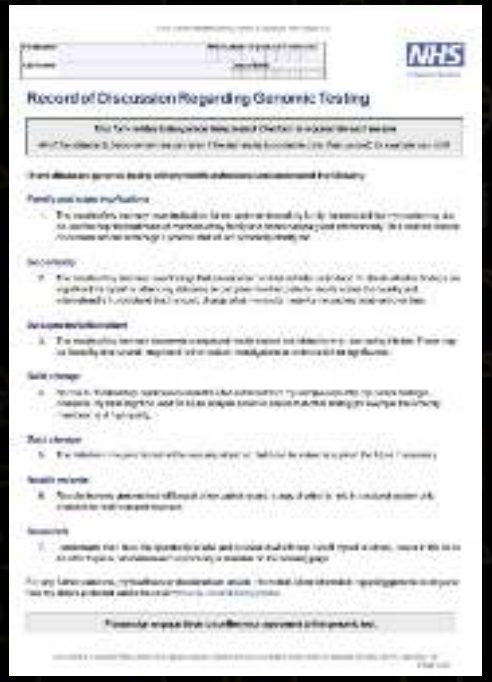
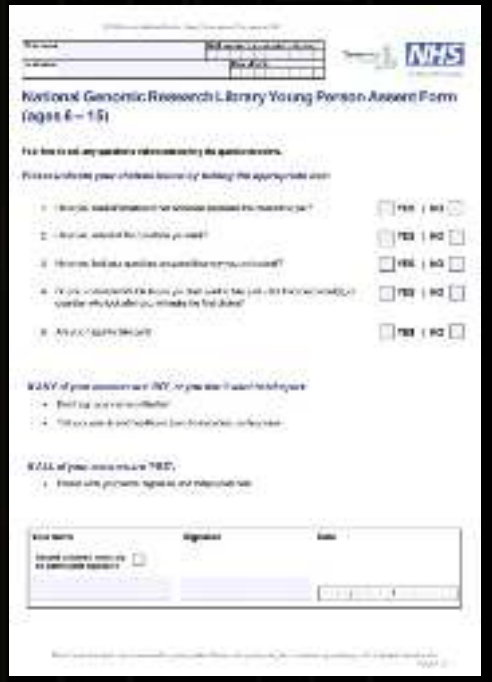
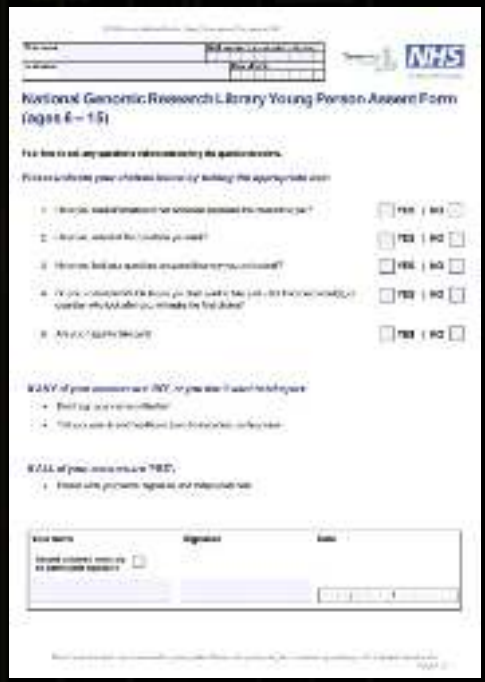
What about in my clinics?


Record of Discussion Regarding Genomic Testing


Consultee declaration regarding whole genome sequencing


National Genomic Research Library Young Person Assent Form (ages 6 – 15)

What about in my clinics?



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What about in my clinics?












KEEP CALM BECAUSE EVERYTHING IS AWESOME

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delivering RESULTS

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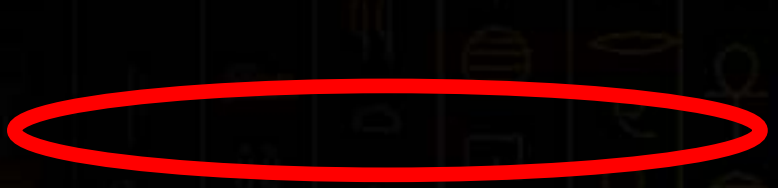
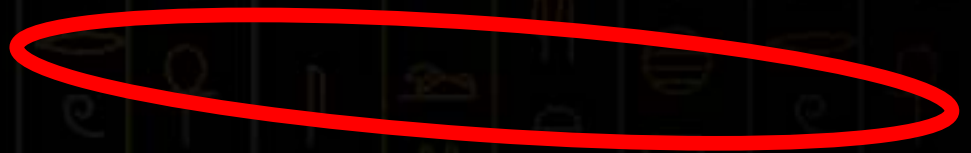
How will you deal with uncertainty?





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What about multiple causes?

RETINAL DYSTROPHY MUTATION ANALYSIS REPORT

NAME: [redacted]
DATE OF BIRTH:
SEX:
NHS No:
YOUR REF:
OUR REF:

ADDRESS: [redacted]

DATE: [redacted]

REASON FOR REFERRAL: [redacted]. RD NGS testing has been requested.

RESULTS:

NAME (DoB)	CLINICAL STATUS	RESULT
[redacted]	[redacted]	ABCA4 c.1609C>T p.(Arg537Cys) Homozygous, ABCA4 c.5881G>A p.(Gly1961Arg) Homozygous & CRB1 c.2714G>A p.(Arg905Gln) Homozygous (see **comments** and additional findings)

Additional variants: *The unclassified variants listed below have been identified during this screening. These variants have been scientifically assessed and currently are of unknown clinical significance either due to the absence of any evidence for or against pathogenicity, or the variant has been seen in control populations and there is no evidence to support pathogenicity however the minor allele frequency is below 1% therefore we cannot rule out that the variant is pathogenic.*

Sequence change
GPR98 c.13792C>A p.(Pro4598Thr) het
ELOVL4 c.699G>A p.(Thr233Thr) het
ADAM9 c.583C>G p.(Pro195Ala) het
MYO7A c.4782G>A p.(Val1594Val) hom

Sequence change
GPR98 c.18353C>T p.(Thr6118Ile) het
IMPDH1 c.921C>T p.(Ile307Ile) het
VPS13B c.8145T>C p.(Val2715Val) het
FZD4 c.1589G>A p.(Gly530Glu) het

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How to deal with expected findings

Aug 2025

GENOMIC LABORATORY REPORT

Dr Jay Self
Consultant Ophthalmologist
Southampton General Hospital
Tremona Road
Southampton
Hants. SO16 6YD

fi'r bcr edacl

Patient Name: [redacted]
Date of Birth:
Sex:
NHS No.:
Your ref.:
Family No:

cc: WGS rare disease Results, West Midlands Regional Genetics Laboratory, BWC NHS Foundation Trust, Birmingham.

Reason for testing

Diagnostic: to investigate the cause of [redacted] retinal dystrophy. Further to our previous report dated 23/01/2024, a gene agnostic analysis of the whole genome sequencing data has been undertaken by the Genomics England Clinical Interpretation Partnership (GeCIP) and a candidate variant identified.

Result summary

Possible genetic diagnosis of autosomal recessive *CLN3*-related neuronal ceroid lipofuscinosis; additional evidence required to confirm or refute this (see below)

Result

[redacted] is heterozygous for a pathogenic *CLN3* deletion and a *CLN3* missense variant of uncertain clinical significance (details below).

Implications of result

Biallelic pathogenic *CLN3* variants cause autosomal recessive neuronal ceroid lipofuscinosis type 3 (MIM204200); characterised by retinal degeneration and variable neurologic manifestations[1]. As a diagnosis is not yet confirmed, these variants should not be used in isolation for clinical decision making.

fi'r edacl bcr

am, B15 2TG

A genetic cause of this individual's clinical presentation has not been identified

Result

This analysis[1] did not identify any underlying genetic cause of his clinical presentation. This result does not exclude a genetic diagnosis in this individual due to the limitations of the analysis.

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Never do anything yourself that you can ~~hire~~ MDT someone else to do, especially if they can do it better.

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Only do genomic testing with clinical genetics support

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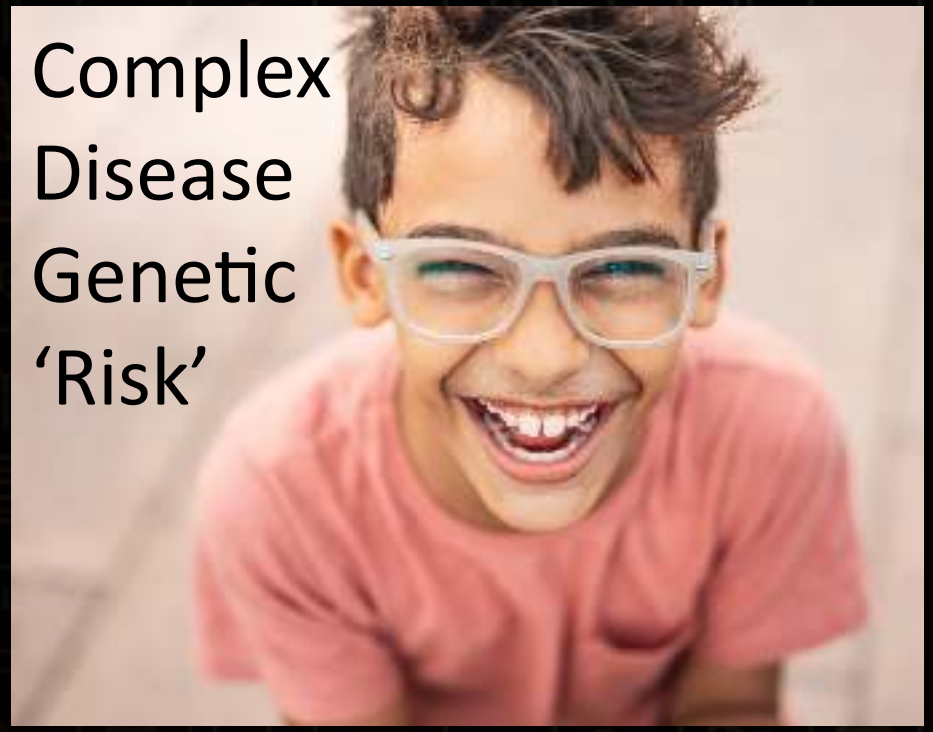
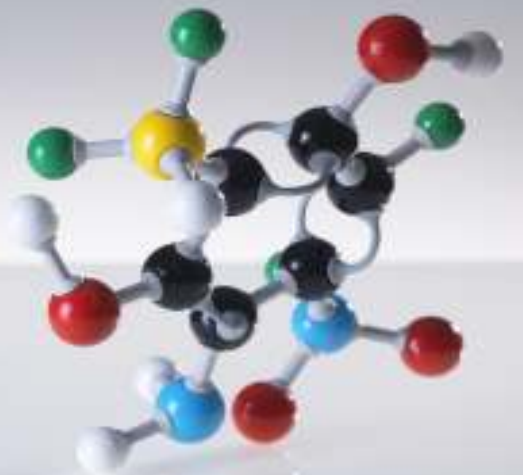
ONE LAST THING

.....it's only going to get harder!

Complex Disease Genetic 'Risk'



Pre-marital Genetic Screening



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