Belfast Health and Social Care Trust

caring supporting improving together

GenOCEANIC
– creating a data infrastructure for precision clinical genomics

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Belfast HSC Trust

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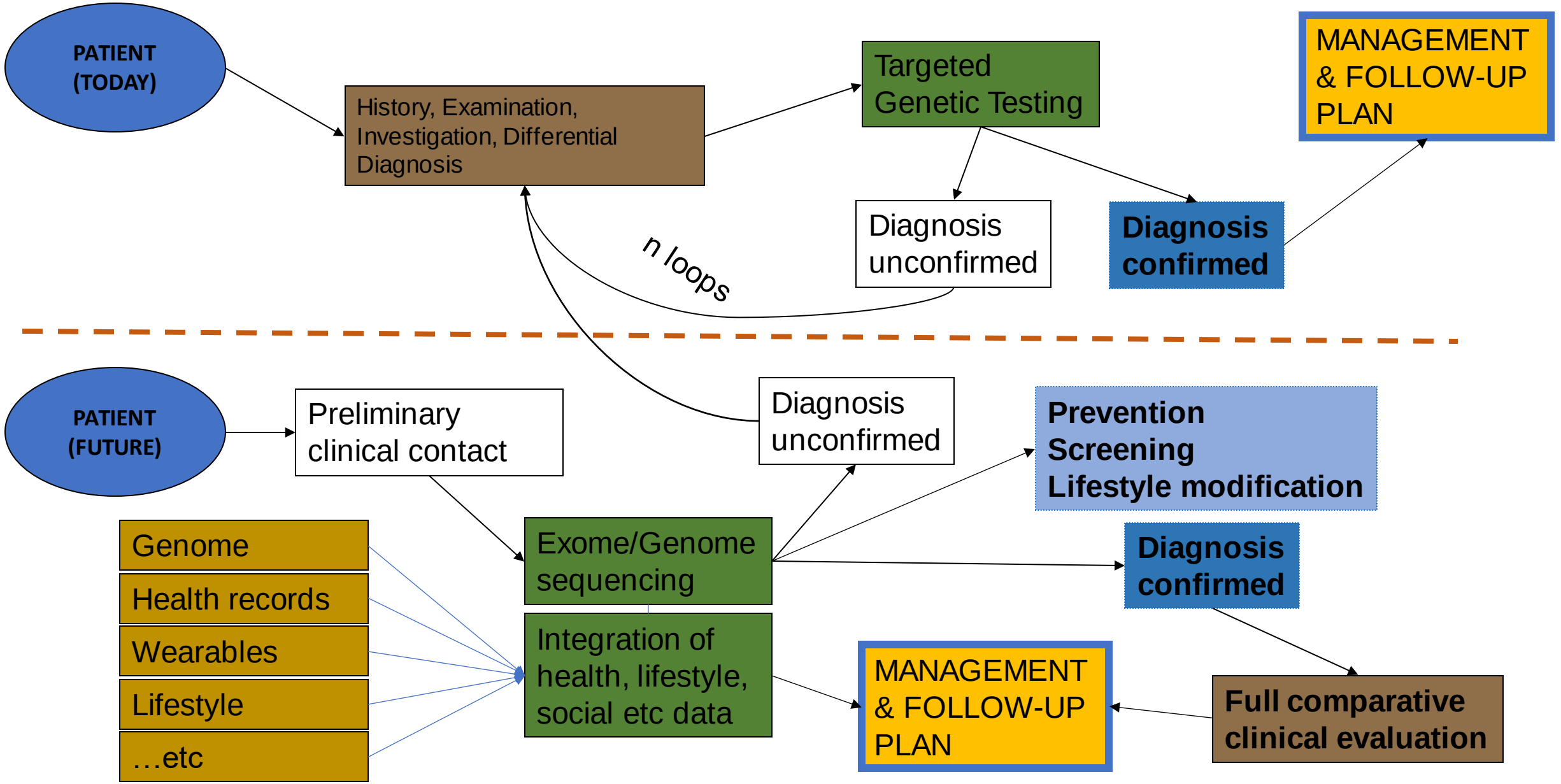
A patient with a problem…
… A clinician with a question
!
?

Precision medicine for rare diseases?

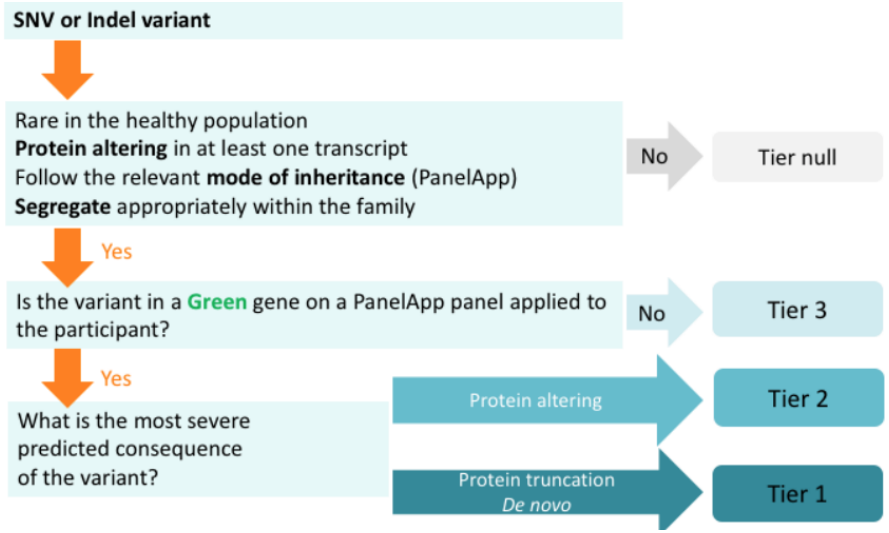


It should be obvious but...

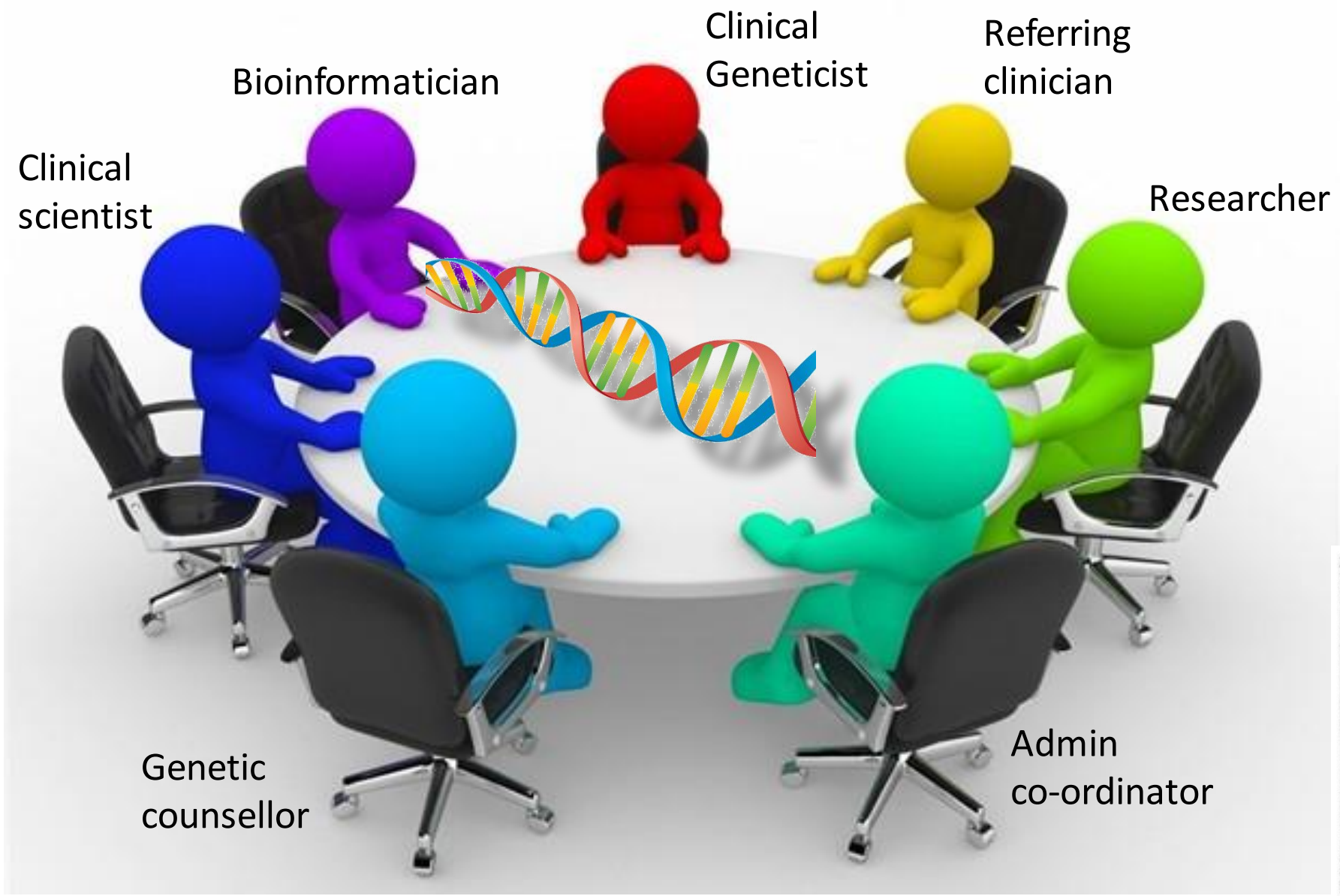
- Precision medicine needs to be precise
- It is not a blunderbuss approach
- Diagnosis is not the end of the process
- "Reading the genome" will not always give us the answer
- Not every rare disorder is "genetic"
- We are likely to need trained professionals for quite some time...

Northern Ireland:
UK 100,000 Genomes Cohorts

- **Cohort 1: 402 probands**
  455 Tier1/Tier2 variants returned
  - 243 selected for classification
  - 22 PATHOGENIC; 37 LIKELY PATHOGENIC (14.7%)
  - (not including Tier3/Untiered)
- **Cohort 2: 39 probands**
  105 variants returned (T1/T2)
  - 56 selected for classification
  - 7 PATHOGENIC; 10 LIKELY PATHOGENIC (43.5%)
- **Current:** "deeper dive" ongoing
  - Pathogenic/Likely Pathogenic: **25%**
- Better phenotyping → more diagnoses -
  i.e. better definition of the clinical question

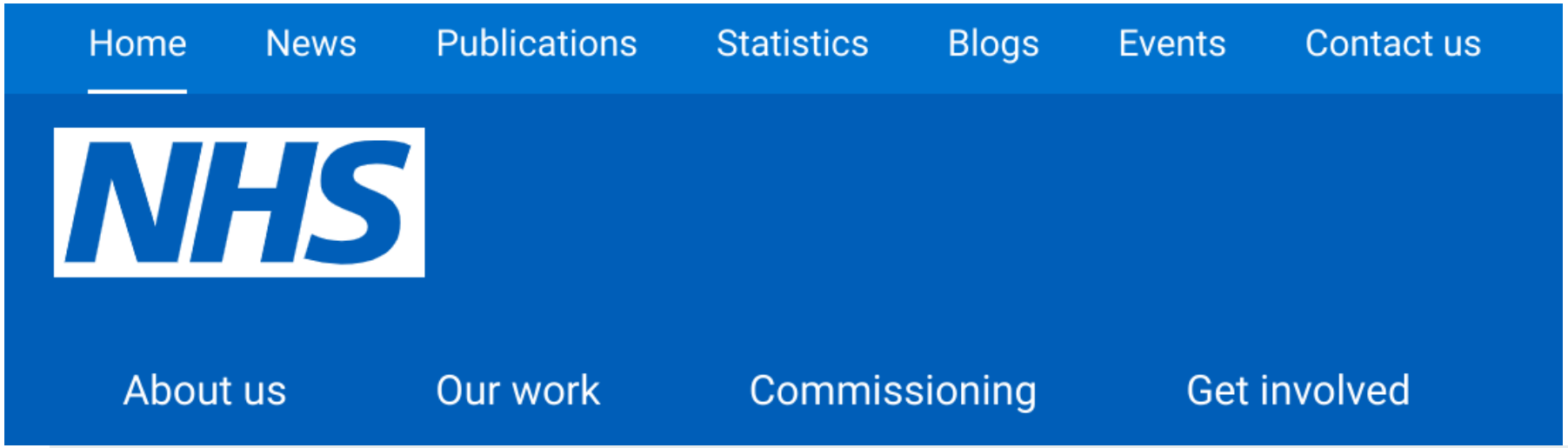


The Genomics Multidisciplinary Team



It takes a village to interpret a genome…

UK National* Test Directory



Document

.pdf

Rare and inherited disease eligibility criteria

PDF 3 MB 391 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.

Updated 21 April 2022.

* England

R60 Adult onset hereditary spastic paraplegia

Testing Criteria

Unexplained spastic paraplegia of likely monogenic aetiology with onset in adulthood

STR testing of spinocerebellar ataxia loci will be included as a component test where spinocerebellar ataxia is considered plausible clinically.

Referrals for testing will be triaged by the Genomic Laboratory; testing should be targeted at those where a genetic or genomic diagnosis will guide management for the proband or family.

Where in Pathway

At presentation following assessment by a Neurologist or Clinical Geneticist

Requesting Specialties

- Clinical Genetics
- Neurology

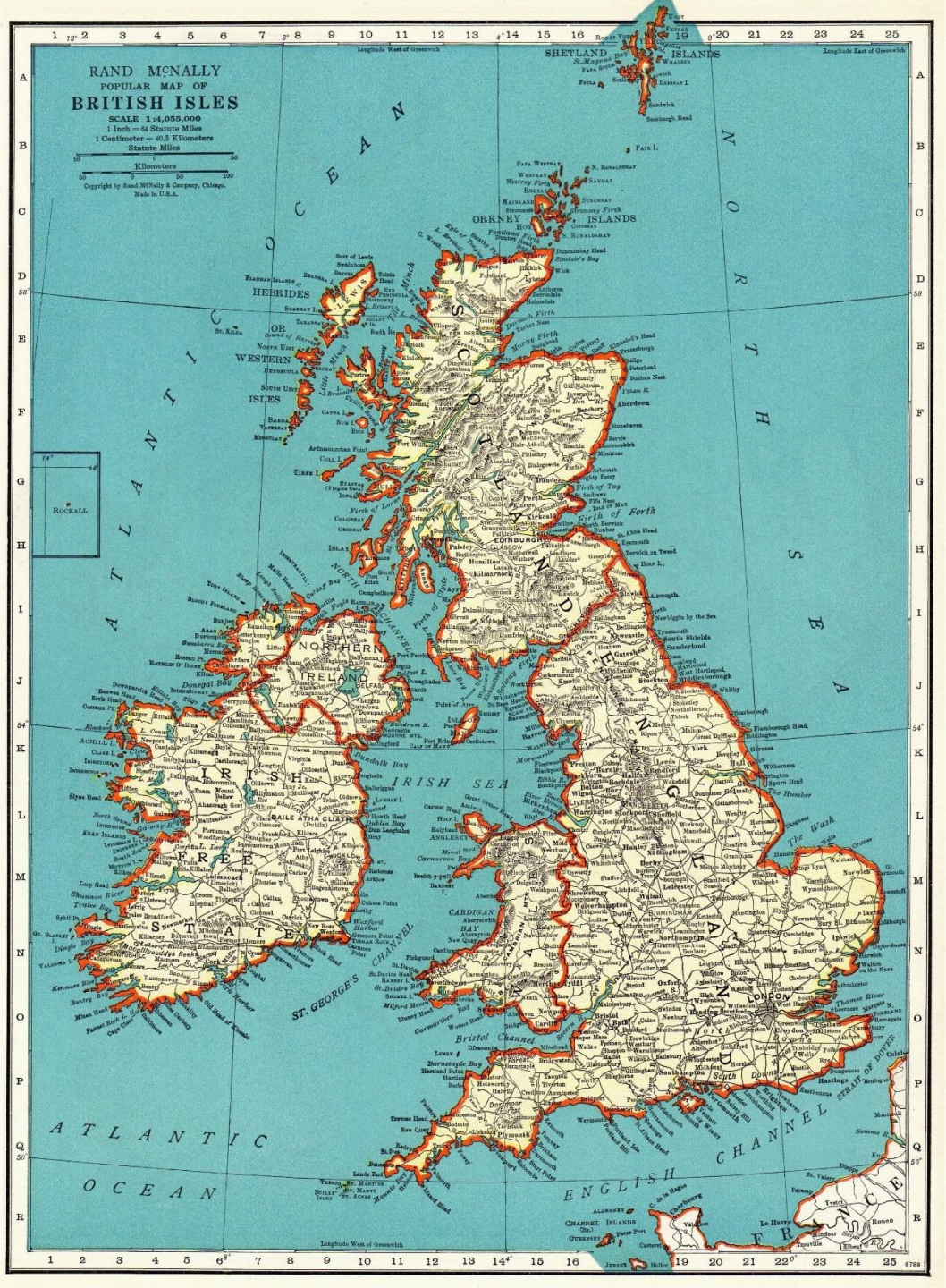
Specialist Service Group

- Neurology

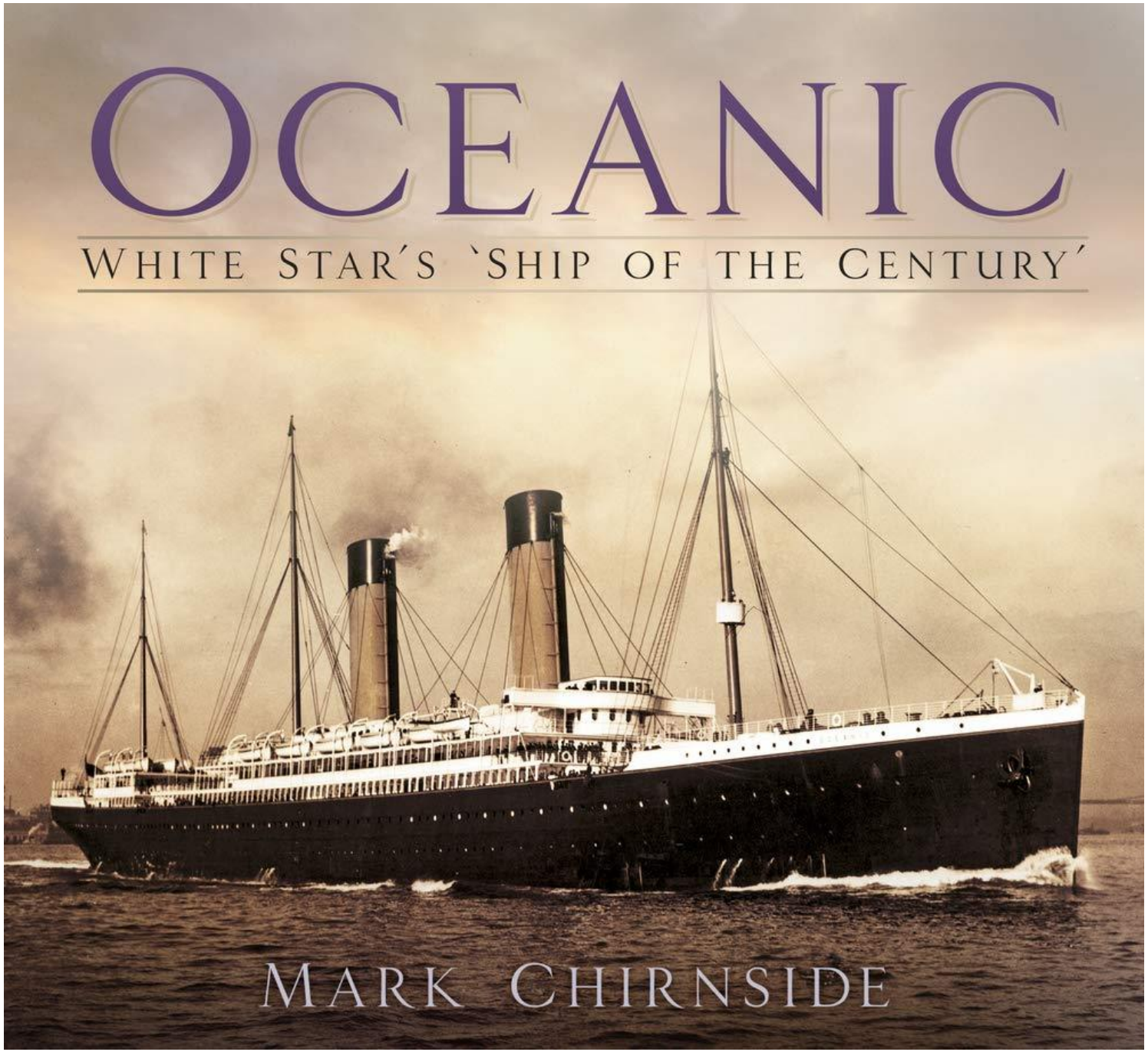
Associated Tests

Please note that initially only WGS testing will be undertaken for R60 Clinical Indication requests, unless clinical presentation and/or initial results indicate all tests are necessary. Whilst this includes testing of all STRs in the gene panel, analysis is currently not optimal and therefore if a specific STR is suspected this should be stated at referral to prompt additional testing where necessary.

Code	Name	Optional Family Structure	Scope(s)	Target Type	Target Name	Method
R60.2	Hereditary spastic paraplegia - adult onset STR testing	Singleton	STRs	Panel of genes or loci	Hereditary spastic paraplegia - adult onset (567)	STR testing
R60.3	Adult onset hereditary spastic paraplegia WGS (phase 2)	Trio or singleton	Exon level CNVs, Small variants, STRs	Panel of genes or loci	Hereditary spastic paraplegia - adult onset (567)	WGS



Genomics
Open
Core
*E*nabling
*A*rchitecture
for **N**orthern
Ireland
Care



GenOCEANIC

- Platform for PHENOTYPE data linked to GENOMIC data
- Open standards platform - *#openEHR* by design
- FHIR, HL7 from multiple existing sources
- Clinical data available for re-use
- Vendor-neutral, technology-agnostic
- Supporting Agile, DevOps, wide scope
- Synergistic with #encompassNI (Epic)

CAMBIO HEALTHCARE SYSTEMS

FUTURE PERFECT HEALTHCARE

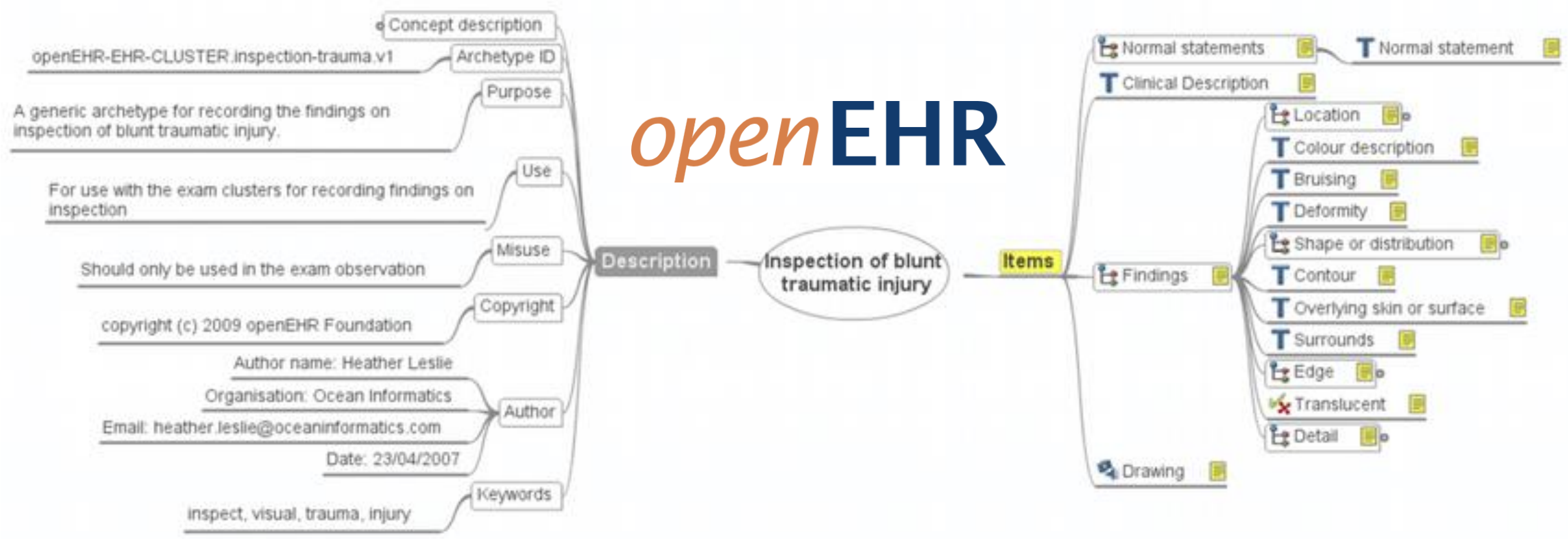
better

ZettaGenomics

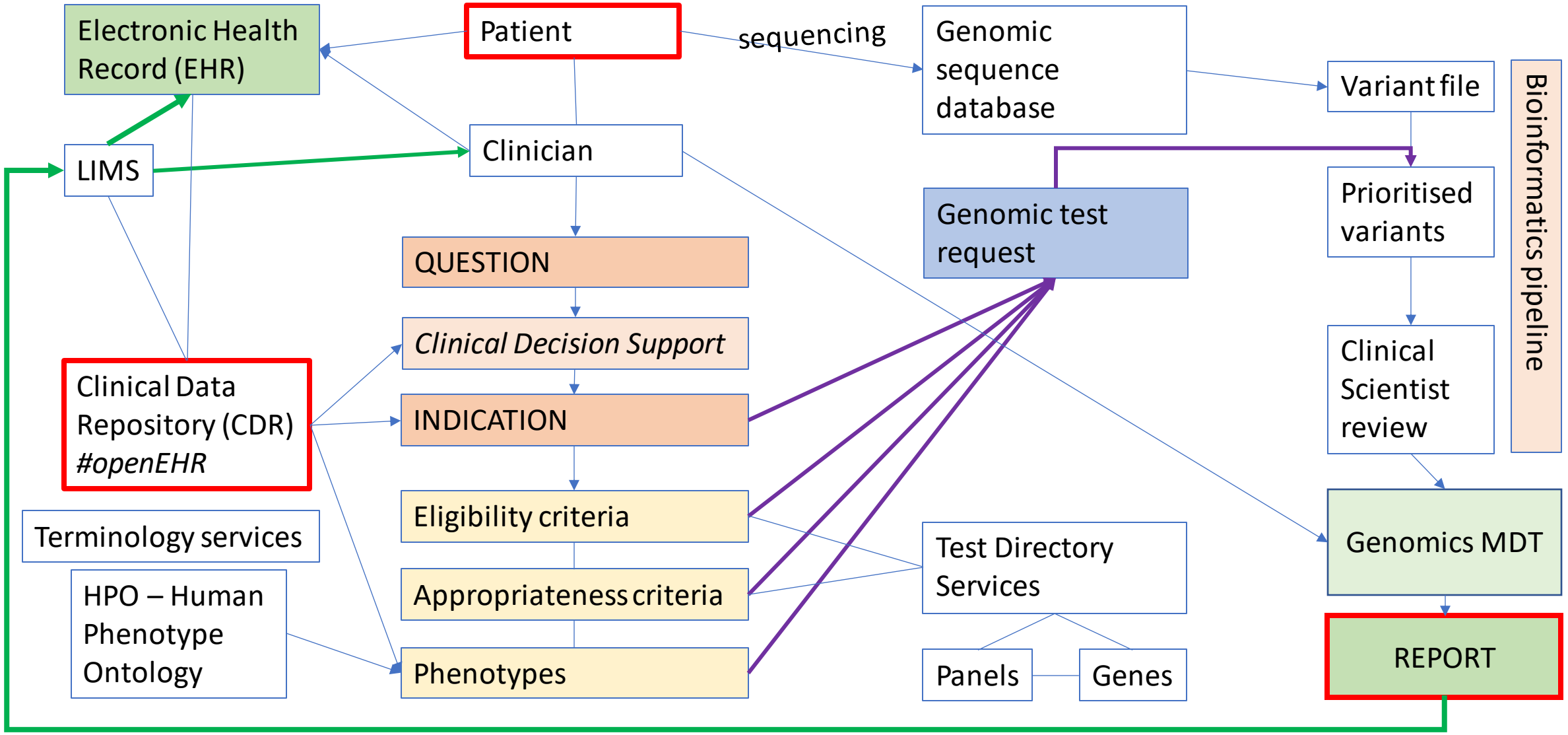
openEHR

www.openehr.org / CAMBIO UK / FuturePerfect / Better Healthcare / Zetta Genomics

#openEHR – open standards, clinically reviewed archetypes & templates – vendor-neutral clinical data repository



Building a diagnostic genomics architecture



Dashboard
Register Patient
Patient Search
Lists
HPO Browser
Form Designers
Tasks
User Management
Administration
About
Audit Log
Log Out

KNIFE, Stanley

Born **31 October, 1943 (79 Years)**

Gender **MALE**

NHS No. **351 063 5132**

ODYSSEY | LAB RESULTS | DISEASE DIAGNOSIS | PHENOTYPES | GENE PANEL REQUEST

Name: SARS-CoV-2 Ag Test (POCT)
Specimen Type: Respiratory
Test Date: 24/11/2022
Laboratory ID: V32000586 MVMLAB MV_RL...
Specimen Recieved: 24/11/2022
Test Status: Final

Name: Flu A,B & RSV PCR
Specimen Type: Respiratory
Test Date: 24/11/2022
Laboratory ID: V32000587 MVMLAB MV_RF...
Specimen Recieved: 24/11/2022
Test Status: Final

Name: SARS-CoV-2 Rapid Ag (POCT)
Specimen Type: Respiratory
Test Date: 23/11/2022
Laboratory ID: V32000584 MVMLAB MV_RL...
Specimen Recieved: 23/11/2022
Test Status: Final

Fanacea

Epic encompass
DELIVERING CARE TOGETHER

Primary care

Private sector

All-island

Out-of-Area

PanOCEANIC

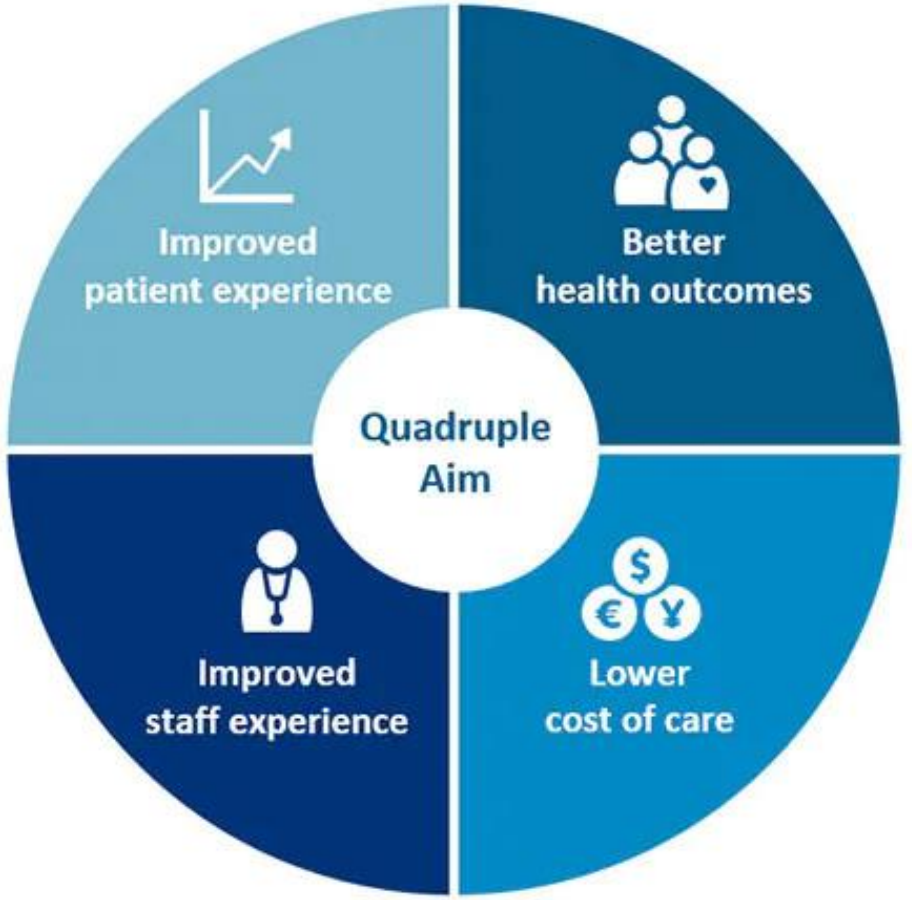
- GenOCEANIC
- PathOCEANIC
- OncOCEANIC
- PharmOCEANIC
- EtcOCEANIC...

- Apps & innovation
- Out-of-scope services
- Registries, Reporting
- PROMS, PREMS
- Upstreaming / civics
- Research

Belfast Health and Social Care Trust
caring supporting improving together

The odyssey continues…

- NI Regional Genetics team
- Cheryl Flanagan – Project Manager
- Shirley Heggarty - NI Regional Genetics Laboratories
- AJ McKnight – Queen's University Belfast & NI Rare Disease Partnership
- Ryan Wilson & Finola McGrady – NI Dept of Health
- Mark Thornton – Cambio Healthcare & partners
- Regional clinicians
- Patients & families



CAMBIO HEALTHCARE SYSTEMS openEHR ZettaGenomics better FUTURE PERFECT HEALTHCARE