

GenOCEANIC

– the voyage towards an open standards based platform for clinical genomic analysis

Shane McKee

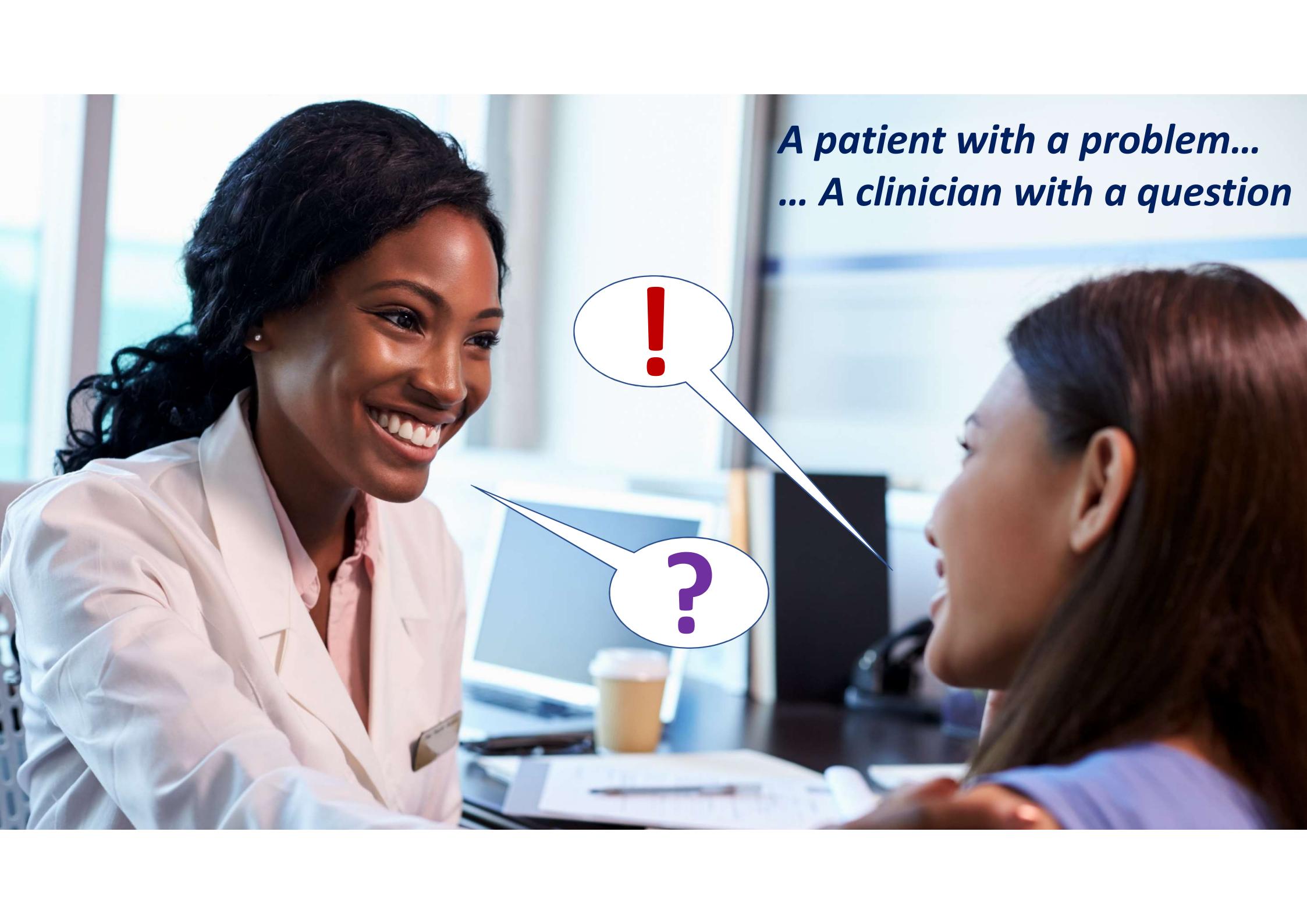
Consultant in Genetic & Genomic Medicine

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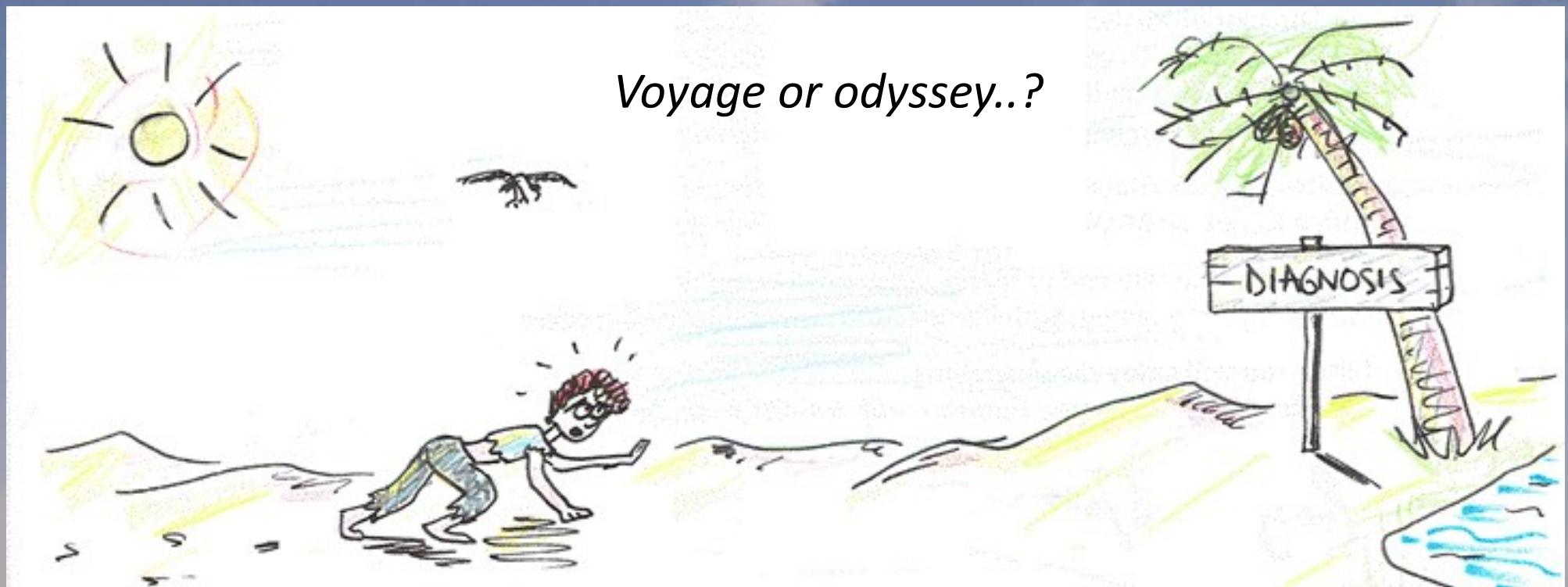
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A photograph of a medical consultation. On the left, a Black female clinician with dark curly hair, wearing a white lab coat over a pink collared shirt, is smiling warmly at a patient. On the right, a young woman with long brown hair, wearing a light blue top, looks towards the clinician. In the center, two thought bubbles extend from the clinician's head. One bubble contains a red exclamation mark (!) and the other contains a purple question mark (?). The background shows a clinical office with a computer monitor, keyboard, and a coffee cup on a desk.

*A patient with a problem...
... A clinician with a question*



Voyage or odyssey..?



#NazBike22

Northern Ireland

- 1.9M population (3% of UK)
- 5 Trusts (Acute, Social Care, Mental Health)
- 11 Council areas
- Legacy of Troubles, political instability
- Relatively poor infrastructure outside Belfast
- Small – max 2h drive to Belfast from anywhere



*Everything is
changing...*



Epic

NIPACS+

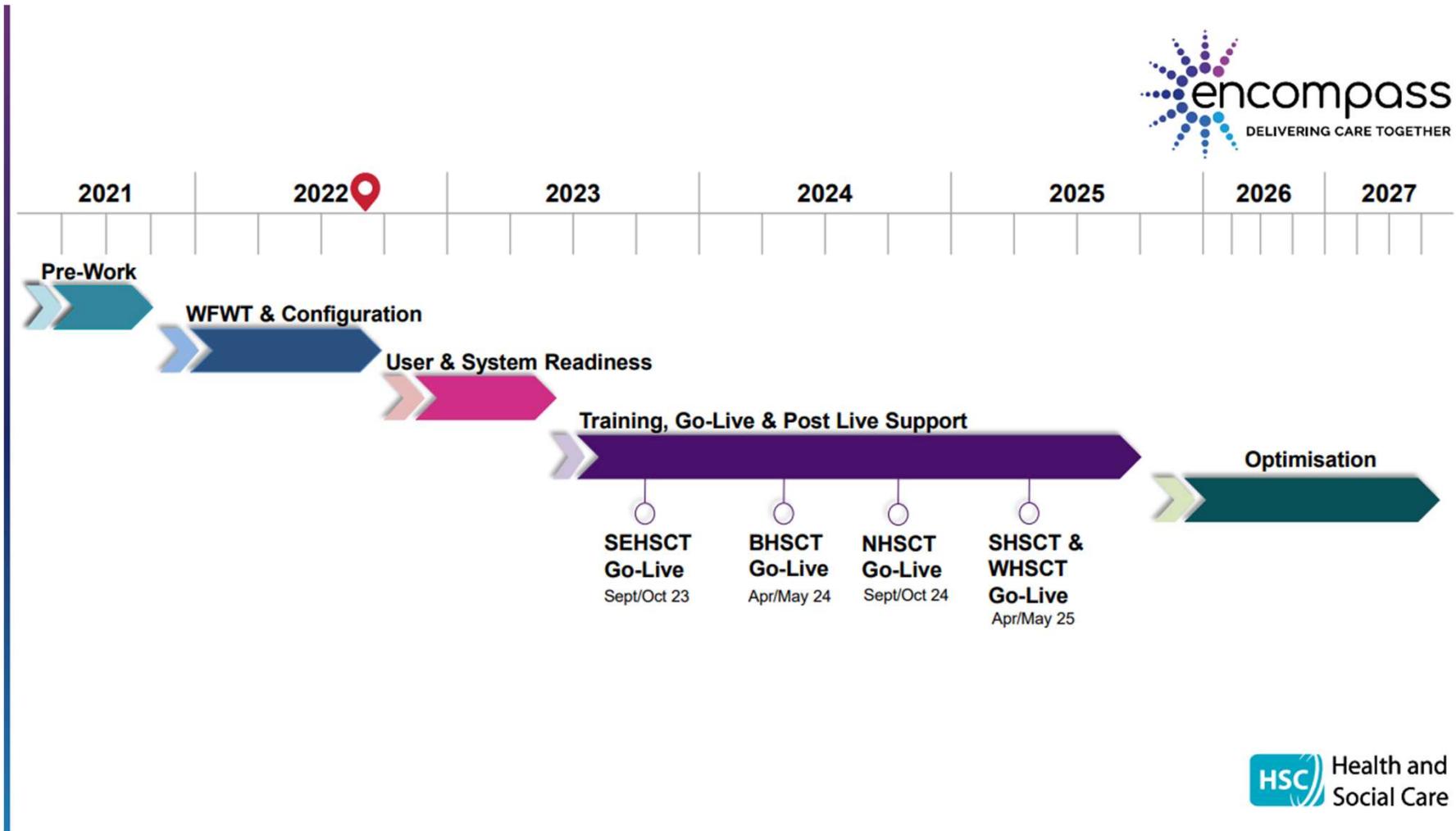
Northern Ireland Picture
Archive & Communication System

NIPIIMS

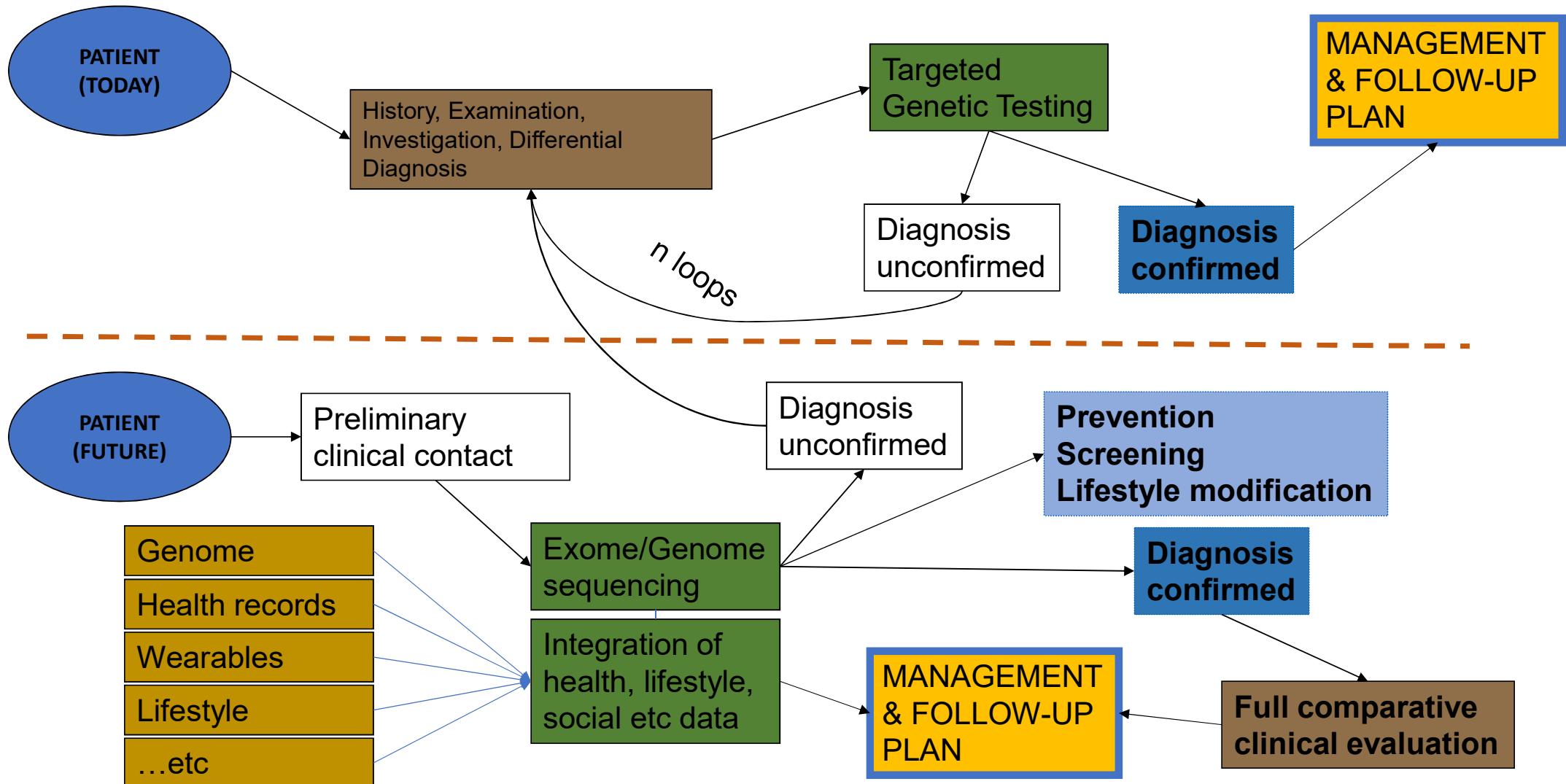
Northern Ireland Pathology
Information Management System

Google

Timeline: #encompassNI



Precision medicine for rare diseases?



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**

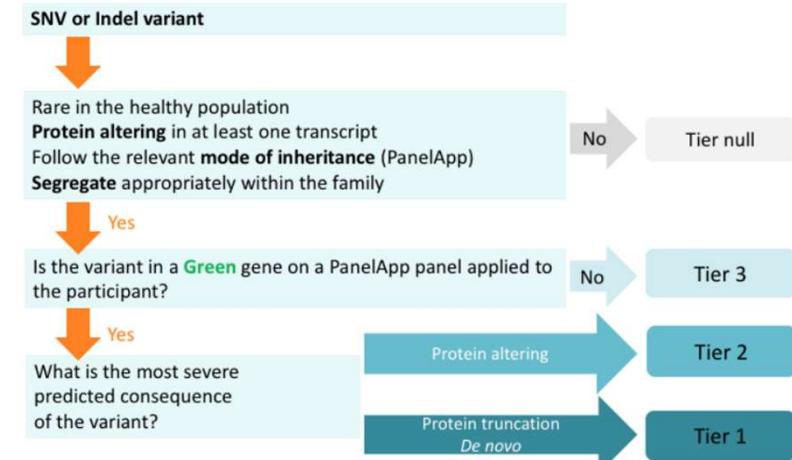
105 variants returned (T1/T2)

- 56 selected for classification
- 7 **PATHOGENIC**; 10 **LIKELY PATHOGENIC** (43.5%)

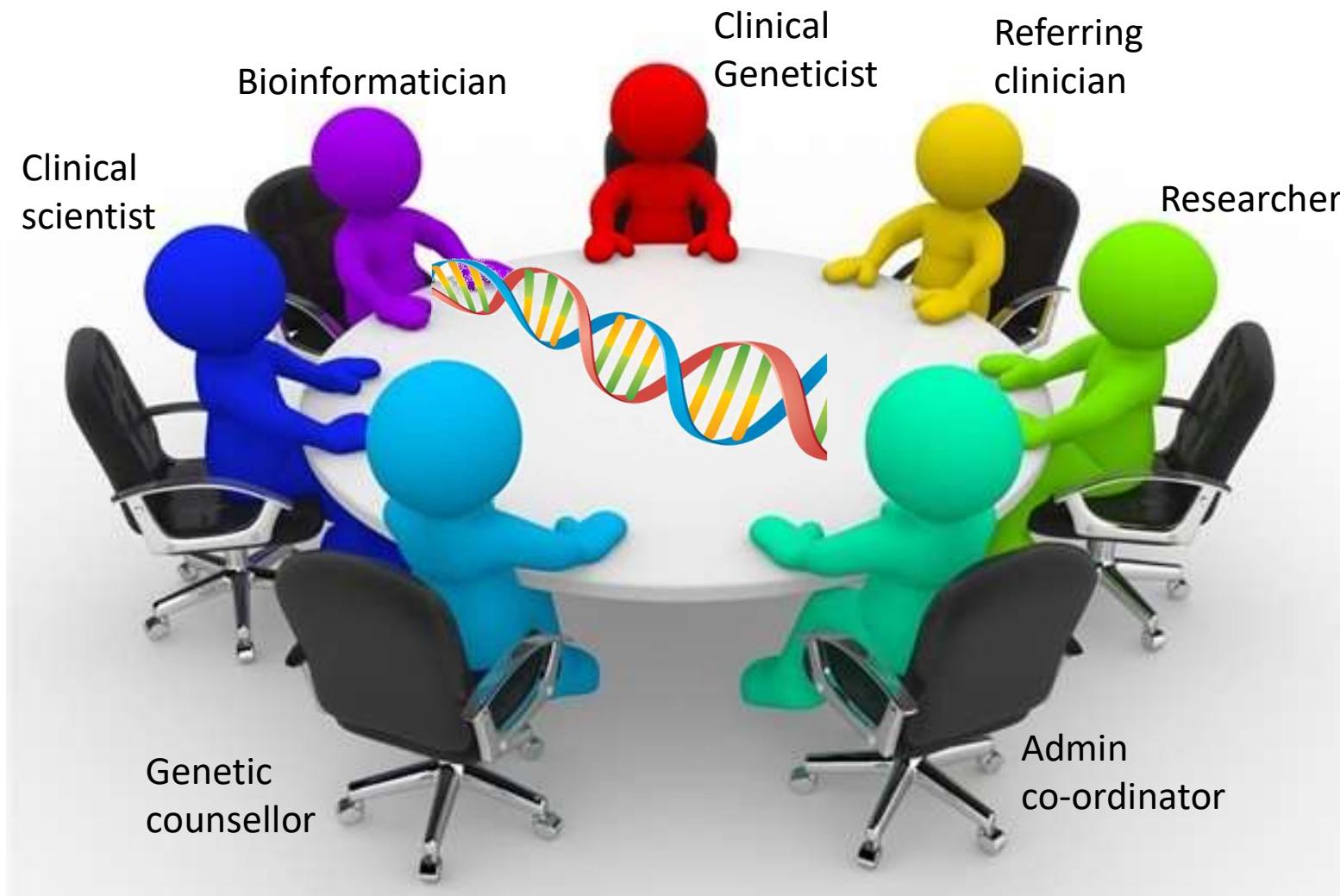
- **Current:** “deeper dive” ongoing

- 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...**



Current “Genetics” software

- **StarLIMS** – handles lab & much of clinical (appointments, patient contacts, family chart management, samples)
- **Progeny** – pedigree charts (largely Genetic Counsellors)
- **Hospital PAS** – letters (PatientCentre), appointments, episodes
- **NIECR** – portal for 2ry care letters, **labs**, GPOOH, appointments, **radiology** – NI-wide
- **Office365** – Teams, SharePoint
- **General on-line**, eg DECIPHER, VarSome, CanRisk

The image displays three windows of genetic software:

- StarLIMS (Live) - Site: Belfast - User: SHANE - Role: Lims_Admin**: A screenshot of the StarLIMS interface showing a patient record for David TESTTCS (Patient ID: G0000000635). The record includes demographic information (DOB: 10/04/1950, NHS Number: N/A), a pedigree number (PED015004), and a notes section. The "Actions" menu includes options like Add, Search, Patient Split, and Patient Merge.
- Progeny**: A pedigree chart showing a family tree. Generation I consists of male 1.3 (Hecto Gram) and female 1.4 (Penta Gram nee Gone). They have seven children in Generation II: II.2 (Mono Type nee Gram, 24 yrs), II.3 (Nano Gram), II.4 (Micro Gram), II.5 (Milli Gram), II.6 (Bari Tone), and II.7 (Poly Tone nee Gram). The pedigree includes symbols for males (square), females (circle), and affected individuals (shaded). A legend at the bottom right specifies: Genetic No: AB123, Created by: [redacted], Date: 12-Nov-2025, Information supplied by: [redacted].
- HSC Health and Social Care**: A screenshot of the NIECR patient record for TEST Gary Test (Male / 50 years). The record shows a summary of clinical information, including key information, assessments, and interventions. It also displays a combined encounter history table with columns for Admission, Discharge, Diagnosis, Visit Type, Location, Clinician, and Status. The table lists several visits, mostly categorized as Ambulatory Emergency (AE) visits to Daisy Hill A&E or DAU.

Core needs for Clinical Genetics

PATIENT MANAGEMENT – Demographics, Coding, Episodes, Referrals

FAMILY MANAGEMENT – Pedigree clustering, Pedigree graphics

REGISTRY FUNCTIONALITY – Surveillance, Follow-up, Case-finding

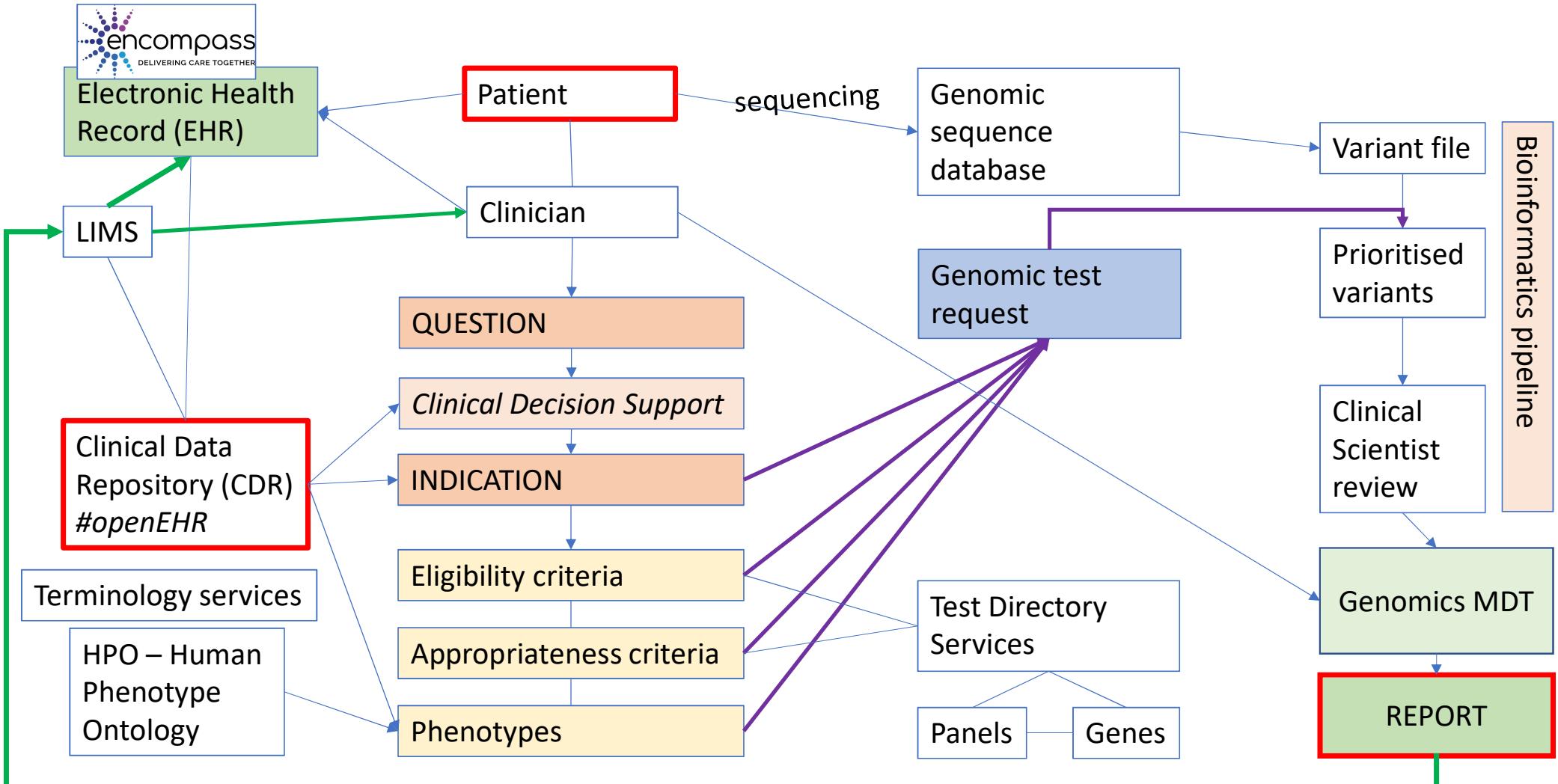
ORDERCOMMS & LAB INTERFACE – Test ordering & resulting

VARIANT REPOSITORY – Bioinformatic pipelines

CLINICAL DECISION SUPPORT – Guidelines, Algorithms, AI

MDT MANAGEMENT – Synchronisation, Tasks

Building a diagnostic genomics architecture



UK National* Test Directory

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NHS

About us Our work Commissioning Get involved

Document



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

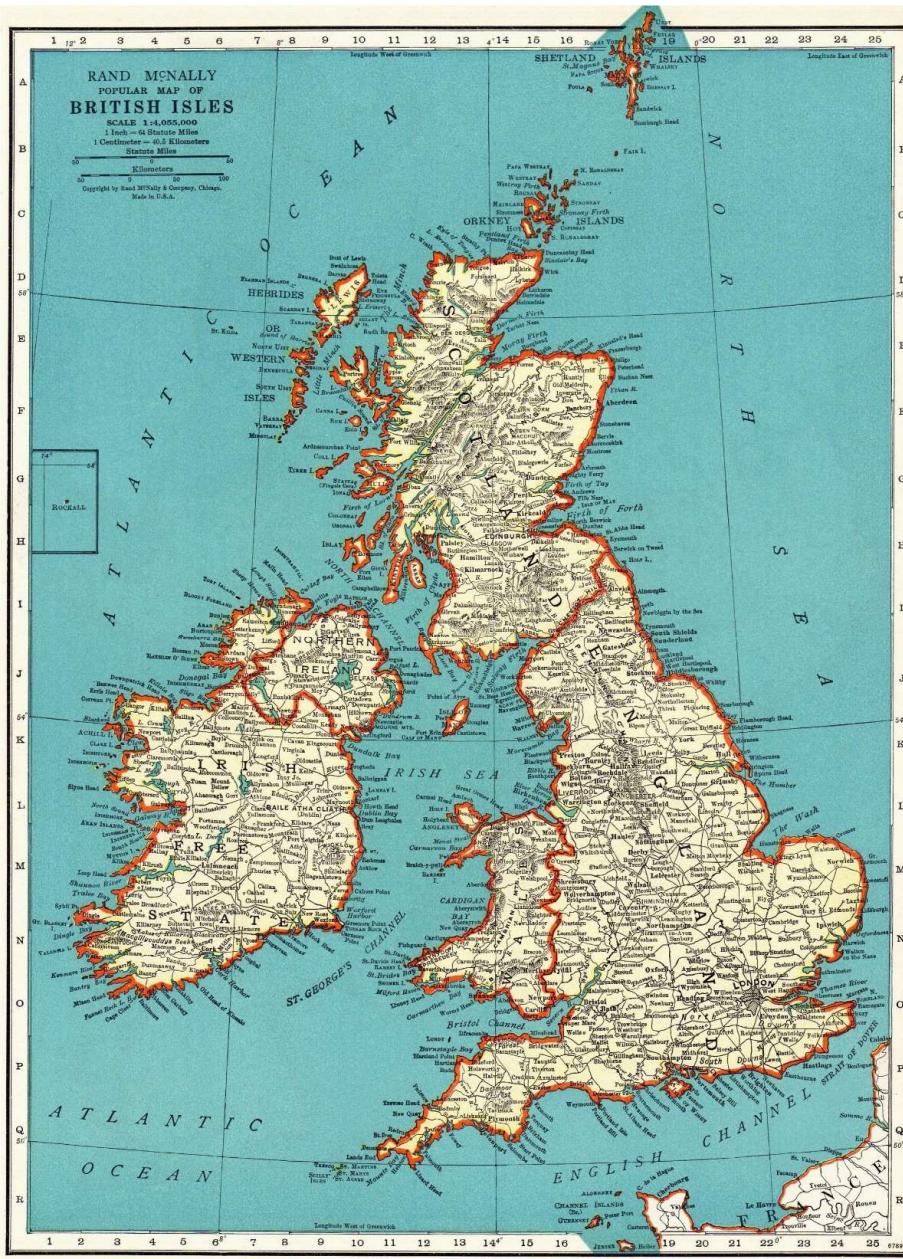
Specialist Service Group

- 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.

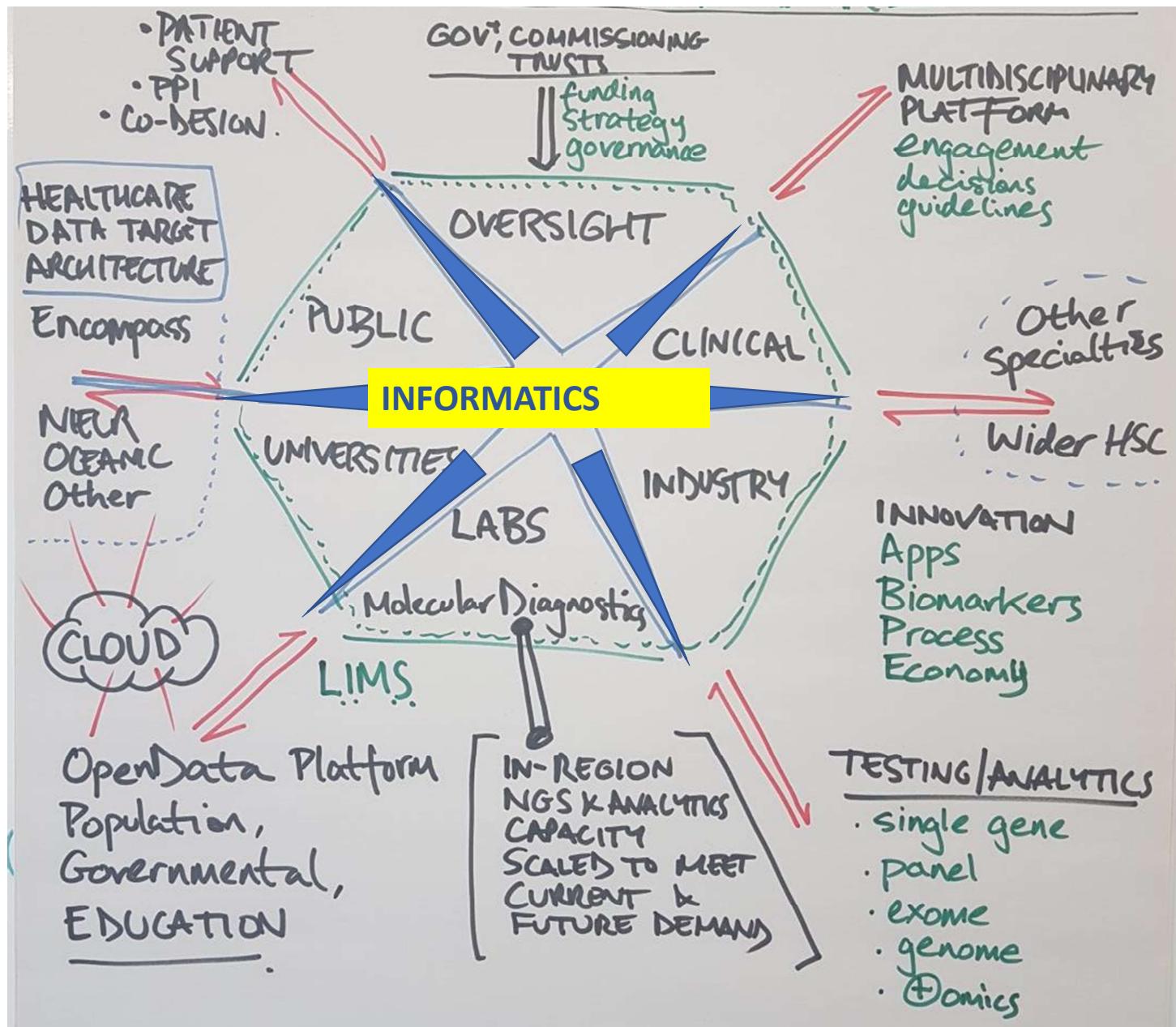
Code	Name	Optional Family Structure	Scope(s)	Target Type	Target Name	Method
R60.2	Hereditary spastic paraplegia - adult onset STR testing	Singleton	STRs	Panel of genes or loci	Hereditary spastic paraplegia - adult onset (567)	STR testing
R60.3	Adult onset hereditary spastic paraplegia WGS (phase 2)	Trio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



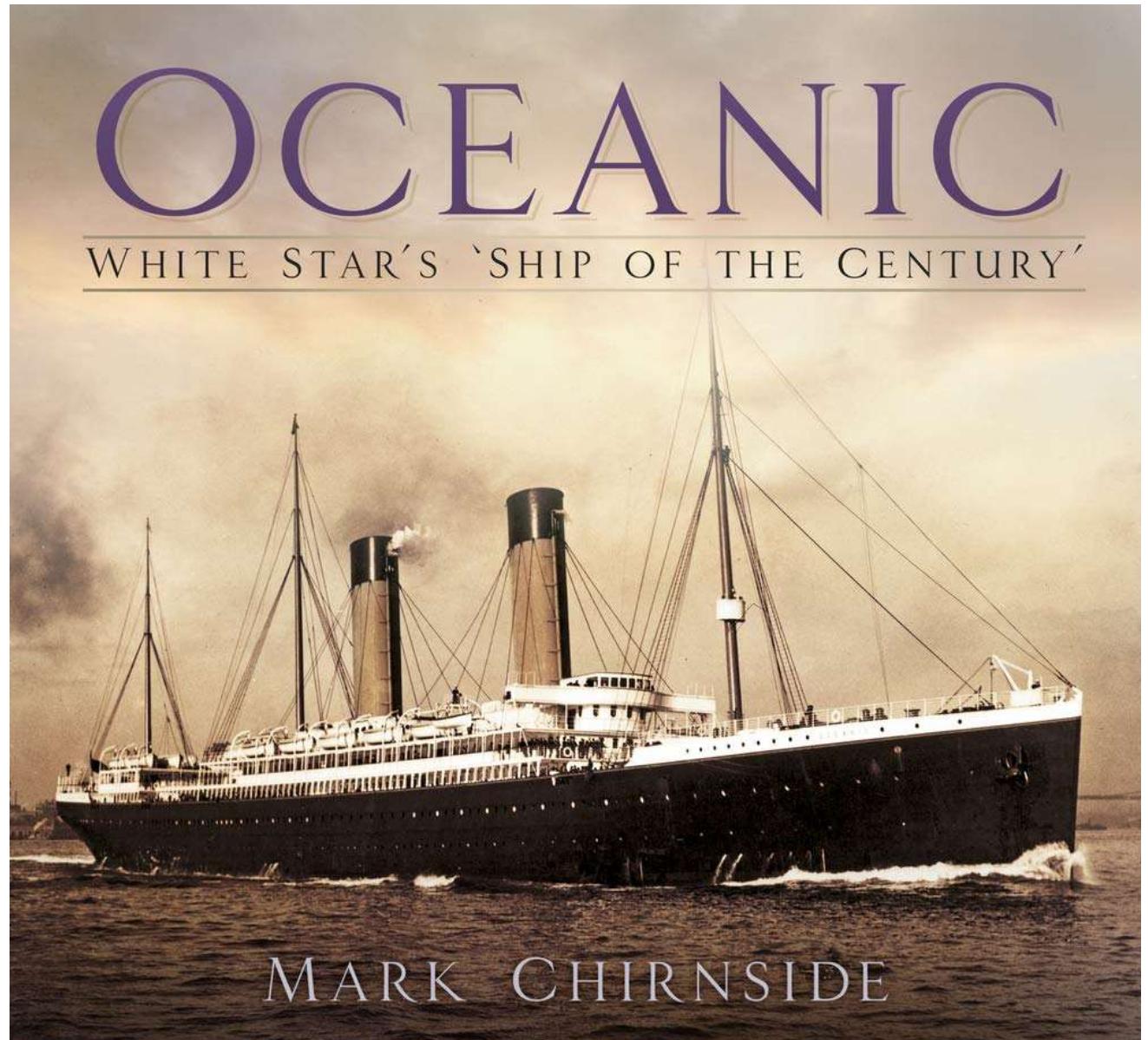


Eligibility vs Appropriateness

- Is this a good idea?



Genomics
Open
Core
Enabling
Architecture
for Northern
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)

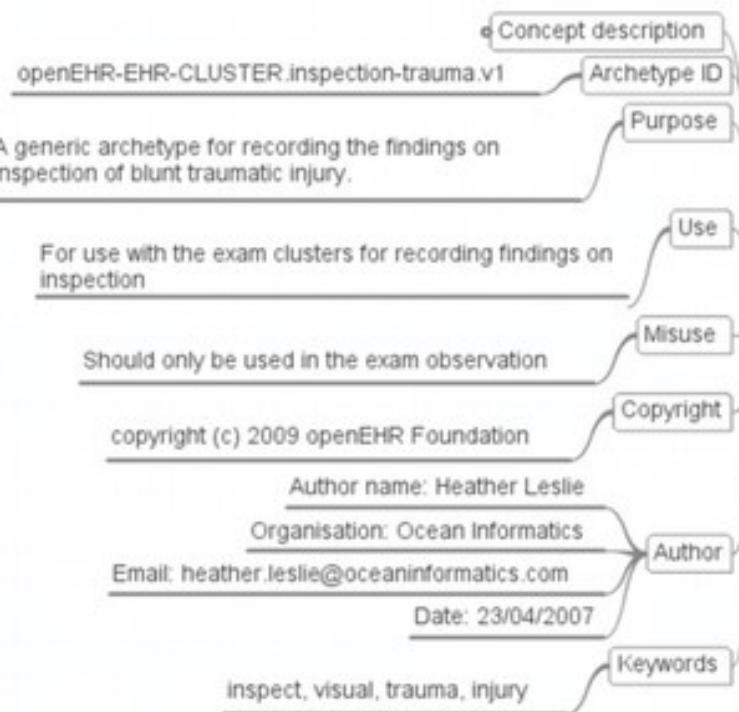


ZettaGenomics

openEHR

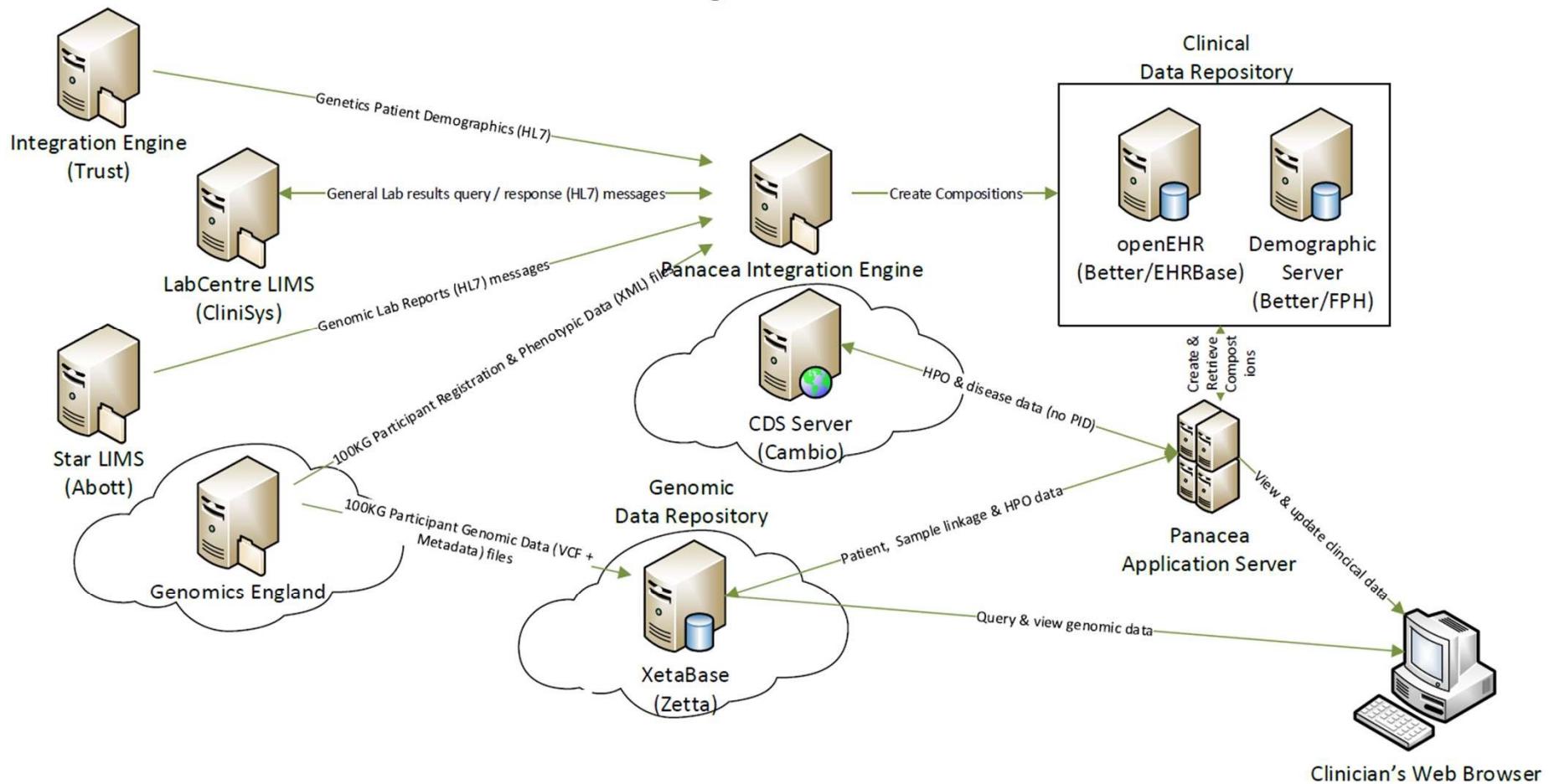
#openEHR – open standards, clinically reviewed archetypes & templates – vendor-neutral clinical data repository

openEHR



GenOCEANIC Data Flow Diagram – Showing Data Flows into openEHR CDR and XetaBase surfaced by the Panacea Platform

Stage 2



* All I.T. components are on site at BHCT apart from where shown in cloud



GenOCEANIC



Dashboard

+ Register Patient

Patient Search

Lists

HPO Browser

Form Designers

Tasks

User Management

Administration

About

Audit Log

Log Out

GenOCEANIC Clinical Data Repository

Welcome to GenOCEANIC , shane.mckee@belfasttrust.hscni.net!

Future Perfect (Healthcare)

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 Received:** 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 Received:** 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 Received:** 23/11/2022**Test Status:** FinalThe logo for Fanacea features a stylized green lowercase letter 'f' with a complex, branching, tree-like or network pattern inside it. To the right of the 'f', the word "fanacea" is written in a bold, dark green sans-serif font.



encompass

DELIVERING CARE TOGETHER

PanOCEANIC

GenOCEANIC

PathOCEANIC

OncOCEANIC

EtcOCEANIC...



Primary care

Private sector

Out-of-Area

Apps & innovation

Out-of-scope services

Registries, Reporting

PROMS, PREMS

Upstreaming / civics

Research

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

