Genomics Informatics Programme

27th June 2025

10:00 – 15:00

Opening thoughts – Deborah Porter

- We welcome you here today
- We thank techUK for continuing to be a valued partner in helping us engage industry
- We thank all of you for taking the time today for joining us
- We see the huge value of our continued industry engagement in shaping and informing our work
- Significant work on strategic budgets has been achieved to ensure the programme continues to accelerate
- Over to the team…

Welcome

- Since the last techUK session we have continued to be busy
- All projects within the programme have made significant steps forward
- We will focus on a programme-wide update, focusing on the three core projects
- Where relevant, we will provide indications to the next areas of focus
- As the programme accelerates our presence has continued to increase:

Introductions

Deborah Porter
Deputy Director
Genomics Service Transformation
NHS England

John Fraser
Joint Head of Informatics
Genomics Service Transformation
NHS England

Hamish Price
Programme Manager
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Ravi Natarajan
Technical Architect
NHS England

Beryl Cairns-Hockey
Project Manager
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Luisa Van Der End
Project Manager
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Michael Price
Business Analyst
NHS England

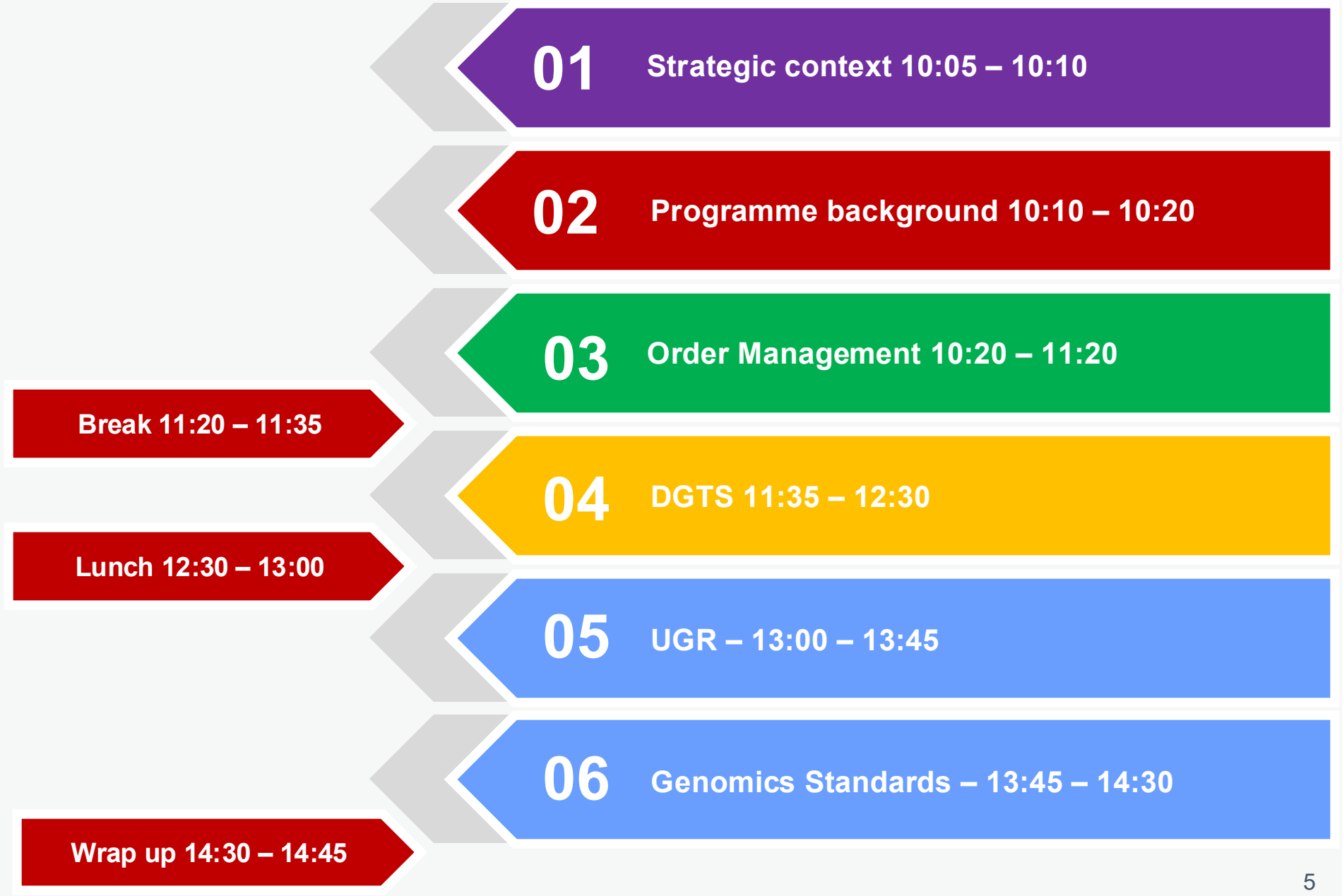
Adam Laurent
Business Analyst
NHS England

Jim Phelps
Business Analyst
NHS England

Omar Khan
Interop Standards
NHS England

Demetra Georgiou
Genomics Service
Imperial

Agenda


01 Strategic context 10:05 – 10:10
02 Programme background 10:10 – 10:20
03 Order Management 10:20 – 11:20
Break 11:20 – 11:35
04 DGTS 11:35 – 12:30
Lunch 12:30 – 13:00
05 UGR – 13:00 – 13:45
06 Genomics Standards – 13:45 – 14:30
Wrap up 14:30 – 14:45

The NHS Genomic Strategy

A 5-year strategy for delivering genomics in the NHS, including 36 commitments for genomics that are closely aligned with other health and life science strategies.

Accelerating Genomic Medicine in the NHS (2022)

Vision	That the power of genomics in predicting, preventing and diagnosing disease, and targeting treatment is accessible to all, as part of routine care in the NHS
Ambition	Over the next five years, we will accelerate the use of genomic medicine across NHS, providing a world leading, equitable services to populations and individuals

Priority 1
Embedding genomics in the NHS, through a world leading, innovative service model

Priority 2
Delivering equitable genomic testing for improved prediction, prevention, diagnosis and precision medicine

Priority 3
Enabling genomics to be at the forefront of the data and digital revolution

Priority 4
Evolving the service through cutting edge science, research and innovation

Supporting the Health Mission, 3 Shifts & 10-year vision for Data Services

Priority 3 – The data & digital revolution

Digitised Test Directory

Digitised Order Management

Unified Genomic Record

Bioinformatics

Cross-NHS Genomic Data and Digital governance established, Framework developed, supporting enterprise architecture principles

Developing an interoperable informatic and data infrastructure

…that enables the NHS to use and share genomic data appropriately to improve patient care.

Putting the NHS at the forefront of using genomic data

…alongside other health data to drive health improvements for individuals and populations.

Enabling the NHS to use cutting-edge analytical tools and up to date variant databases

… to maximise diagnosis, increase access to precision medicine and efficiency.

NHS England Genomic Medicine Service

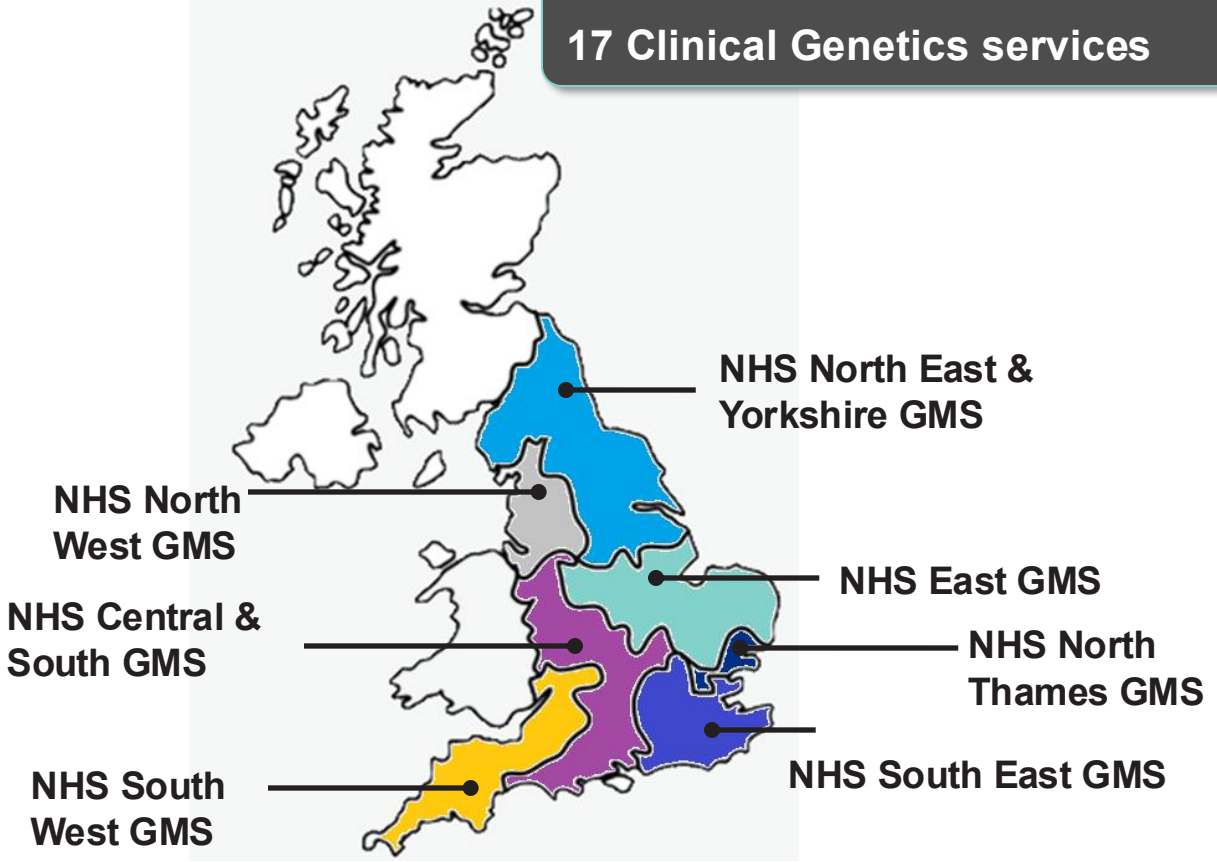
Working across the care continuum from primary to secondary and tertiary care.

Geography	Patient Population	NHS Trusts	NHS ICSs
North East & Yorkshire	8 million	34 NHS Trusts	4 ICSs
North West	7 million	34 NHS Trusts	4 ICSs
Central & South	10 million	45 NHS Trusts	14 ICSs
East	8 million	32 NHS Trusts	13 ICSs
North Thames	7 million	36 NHS Trusts	11 ICSs
South East	8 million	29 NHS Trusts	8 ICSs
South West	4 million	18 NHS Trusts	7 ICSs

7 Genomic Laboratory Hubs

7 Genomic Medicine Service Alliances established from 2021

17 Clinical Genetics services



Continued Engagement

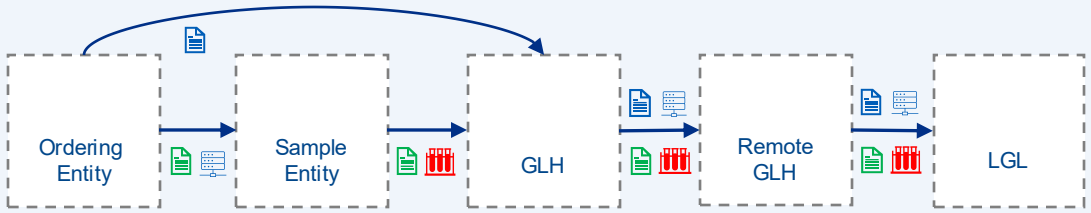
- To shape and understand the complexity of the programme and its requirements we have engaged with over 600 internal stakeholders and many external
- We are also reinforcing internal NHS knowledge with the voice of the patient
- Through the regional Patient & Public Voice (PPV) Leads, we are running a national patient engagement session on July 11th

Digital Order Management

Beryl Cairns-Hockey
Michael Price (remote)
Ravi Natarajan
Demetra Georgiou (Imperial)

Digital Genomics Order Management

CMO
Current Mode of Operation



Paper based test order form

Paper based sample linkage form

Sample(s)

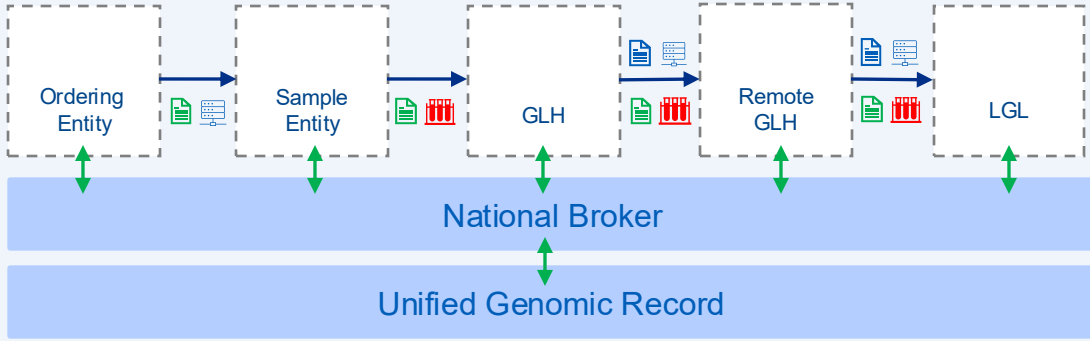
Some local integrations

Challenges

- Variation in how genomic tests are ordered nationally
- Paper based challenges
- Duplication in process
- Lack of visibility
- Inefficiencies

Scalable

FMO
Future Mode of Operation



Opportunities

- Streamlined ordering of genomic tests across organisations
- Request, Process & Track digitally orders, samples & results
- Native interoperability with Electronic Patient Systems (EPRs) & Laboratory Information Management Systems (LIMS)
- Rich operational data for continual improvement

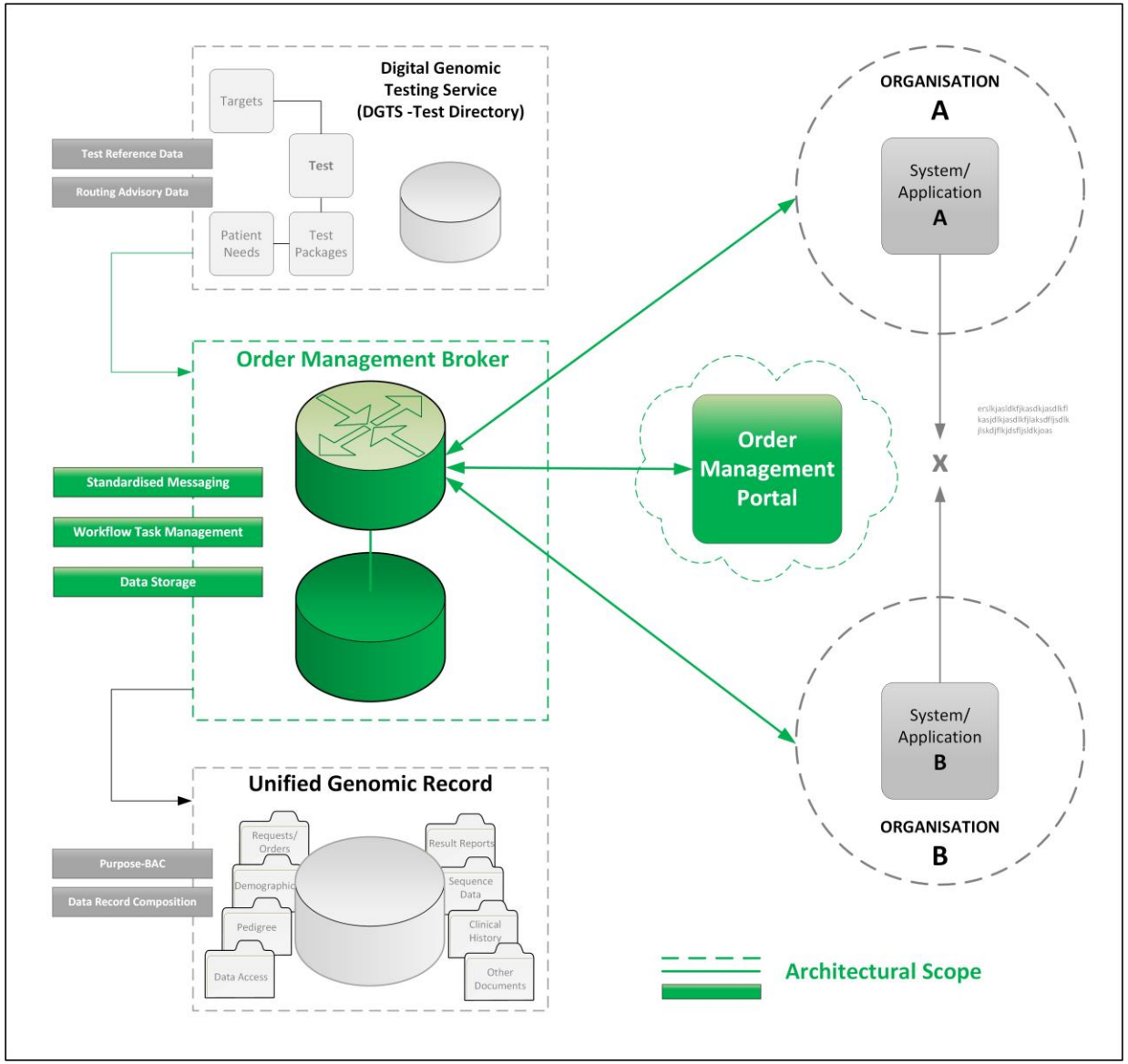
Architecture – Order Management Scope

OM broker infrastructure

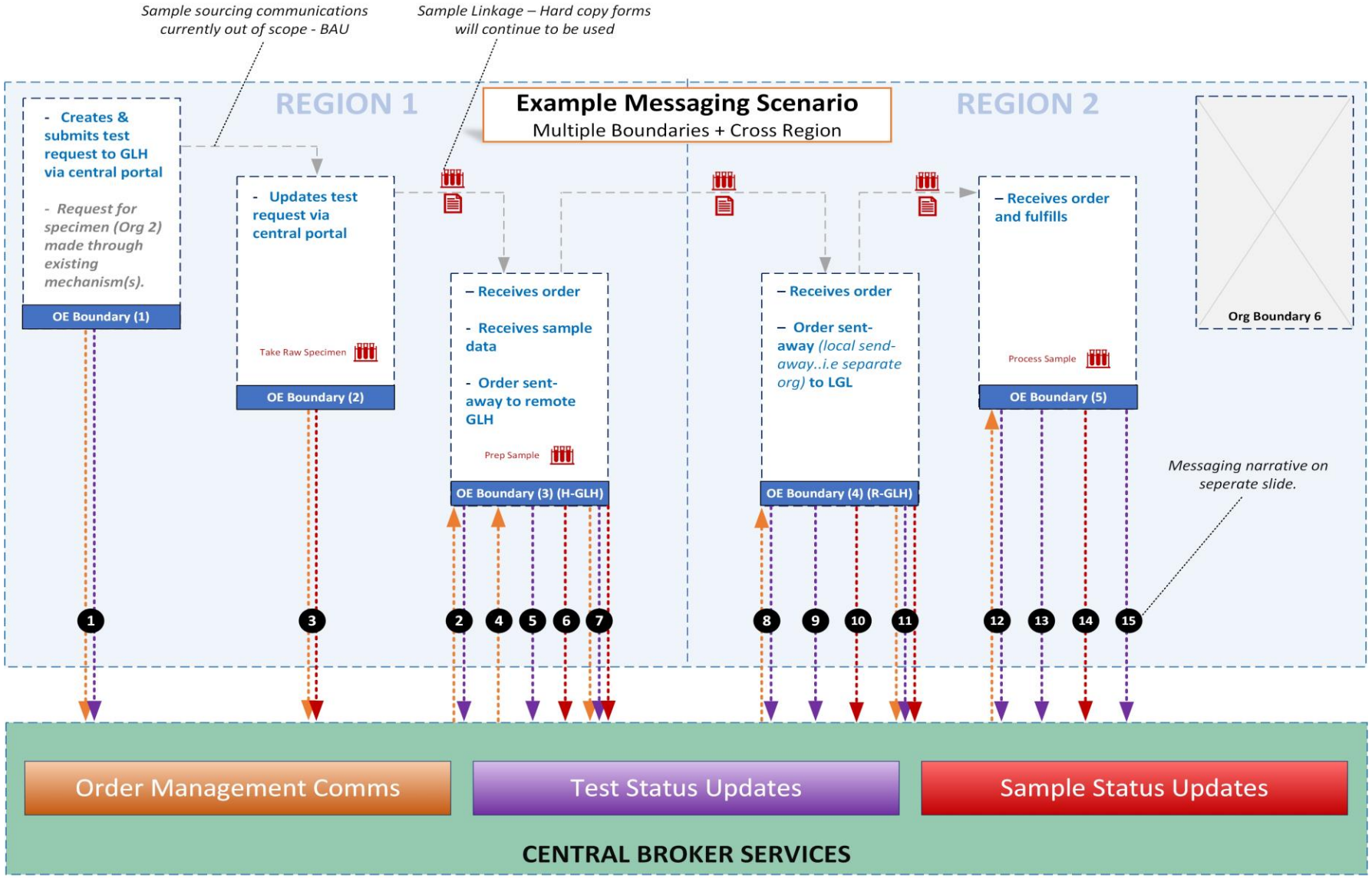
- Facilitate standardised messaging between organisational boundaries
- Provide a workflow task management capability
- Provide the data storage needed to support the movement of data between organisations throughout the workflow
- Consume the DGTS reference data
- Foundation component for UGR

OM portal

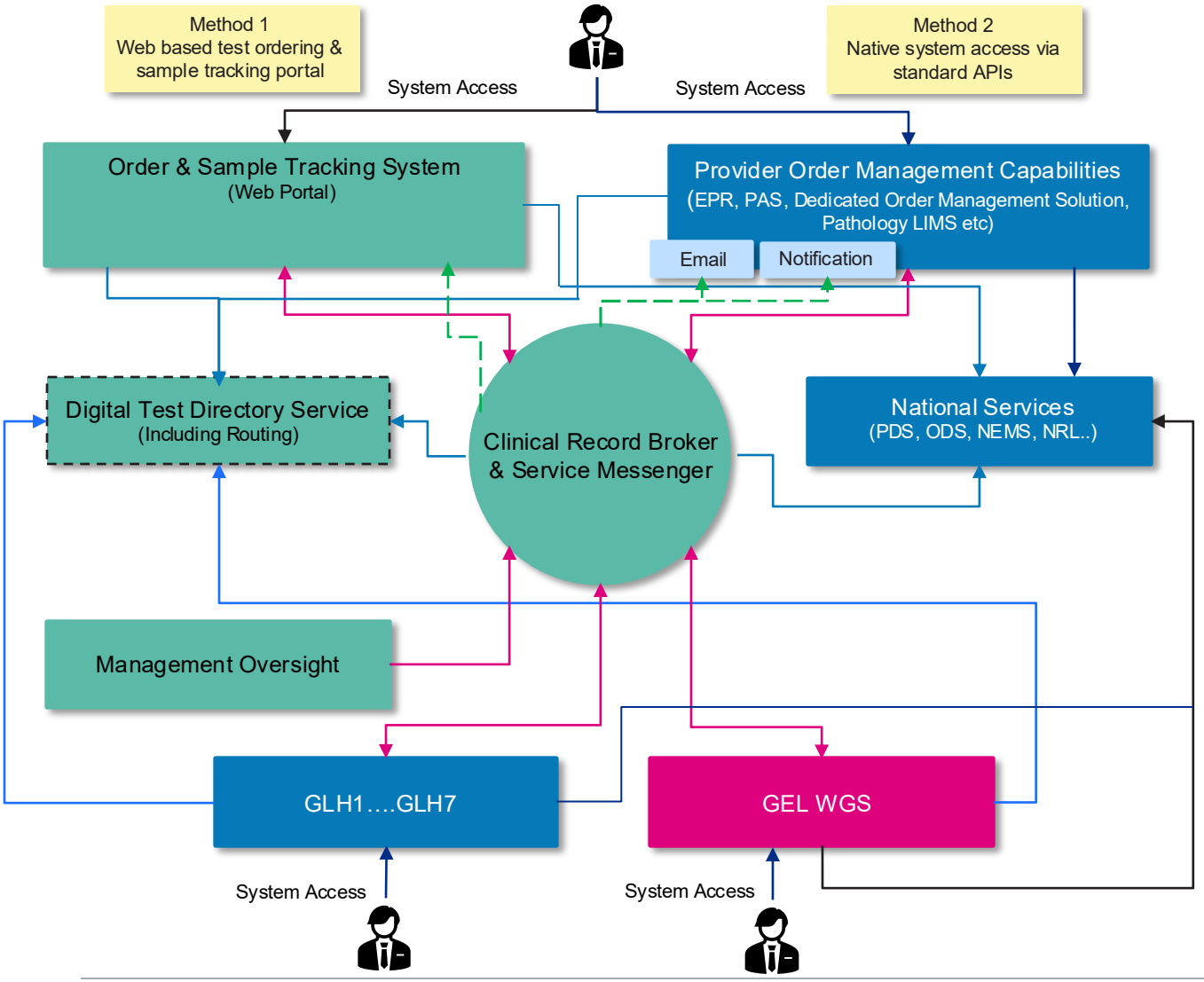
- Provides a user interface for ordering and managing genomic test requests, leveraging all functional capabilities of the broker



Architecture – Systems interaction



Architecture



- Adheres to the GMS architecture vision & Data Digital Framework
- Delivered to meet the needs of the baselined business requirements
- With a central core broker with end-to-end orchestration features utilising FHIR.
- Designed to connect to frontend systems, including:
  - Native interoperability with EPRs and LIMS
  - Integrations with Trust Integration Engines (TIEs)
  - An Alpha National Genomic Web Portal
- In a non-production environment using synthetic patient data
- Testing complex scenarios, including:
  - familial (duo/trio tests)
  - failure routes
  - fetal
- Designed with future Unified Genomics Record (UGR) in mind

Connecting to the Core Broker – 3 Options

Native Interoperability with EPR and LIMS

EPR and LIMS Integrations

National Web Portal

Strategic end goal

What's currently being tested

North Thames GLH (NTGLH) Alpha integration

- NTGLH is made up of 7 lab hubs, 36 NHS trusts across 11 Integrated Care Systems (ICSs) & 7 million patients
- Digital Order Management has partnered with:
  - Imperial, a part of the North-West London ICS Acute Provider Alliance
  - Great Ormond Street Hospital NHS Foundation Trust (GOSH), hosting the NTGLH
  - the Royal Marsden Hospital (RMH), hosting The Cancer Laboratory Hub
- GOSH and RMH share an instance of EPIC Beaker but with different workflows for cancer and rare disease. RMH also has an external facing ordering portal
- Both Trusts have their individual Trust Integration Engines (TIEs)
- Imperial use Cerner as their EPR and Win-path and Co-path as their LIMS and their own TIE

NHS Imperial College Healthcare NHS Trust

The ROYAL MARSDEN NHS Foundation Trust

NHS Great Ormond Street Hospital for Children NHS Foundation Trust

As a part of the Alpha, all three partners have successfully connected to the national infrastructure.

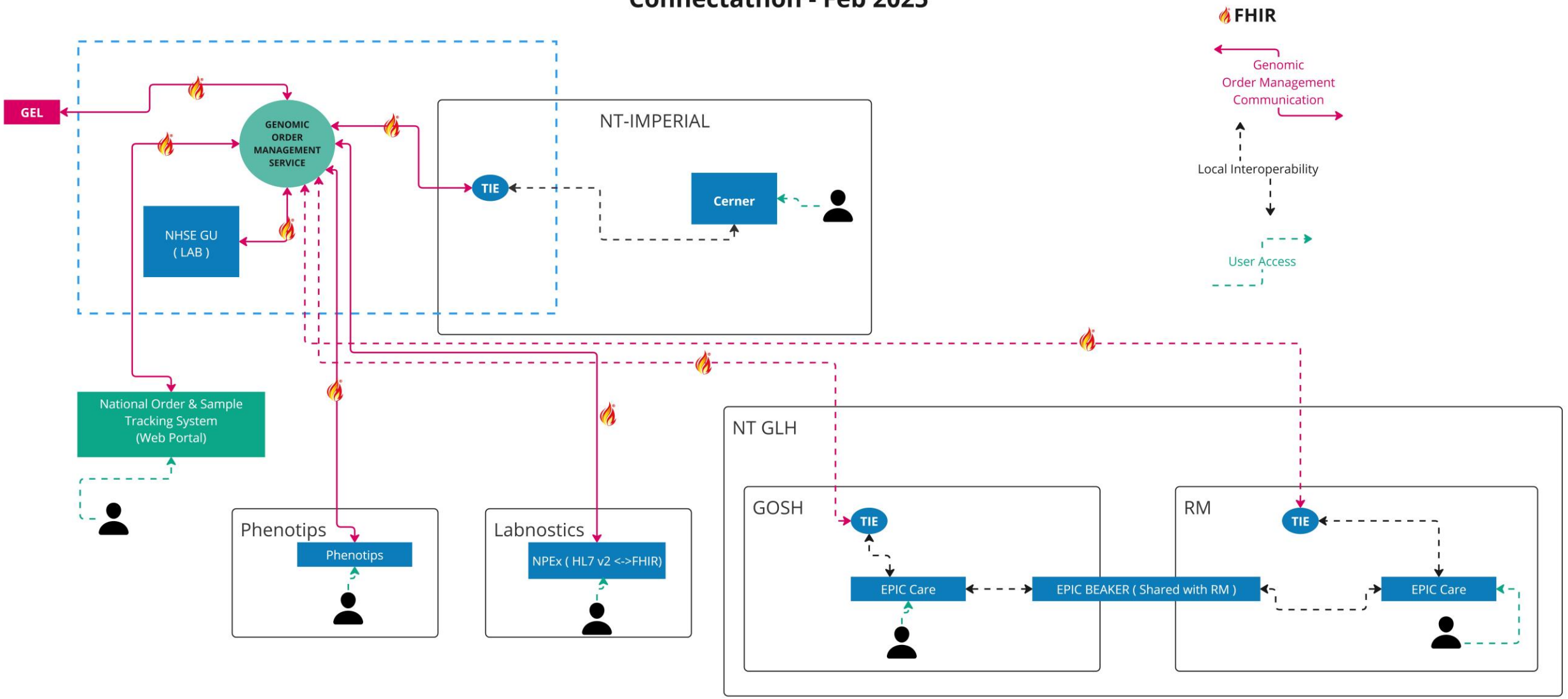
It validates high-level architecture and patterns and exposes real life limitations and challenges.

The Alpha integration will generate a blueprint for other trusts to adopt for their infrastructure.

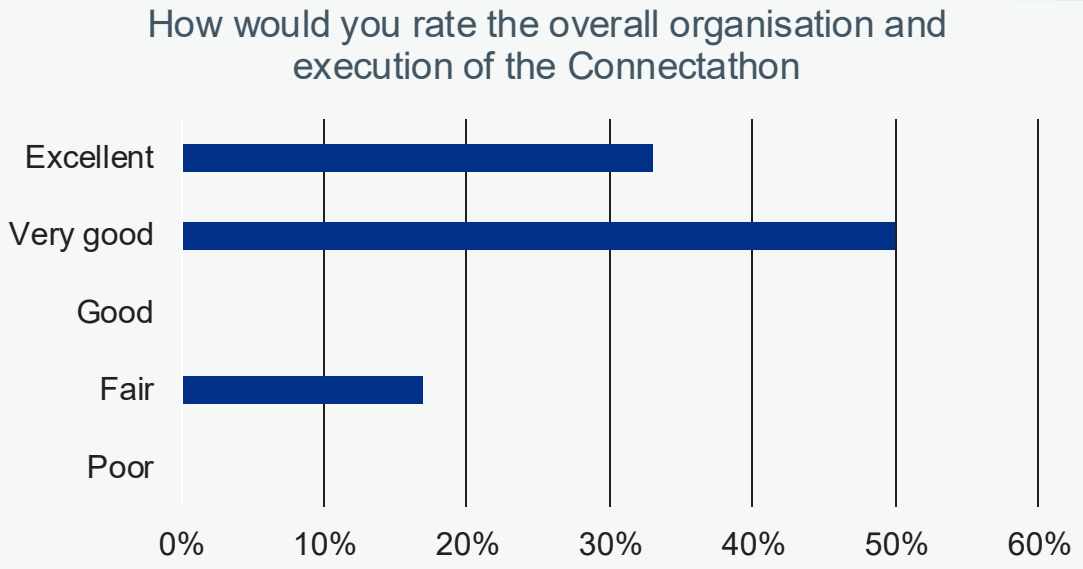
Connectathon

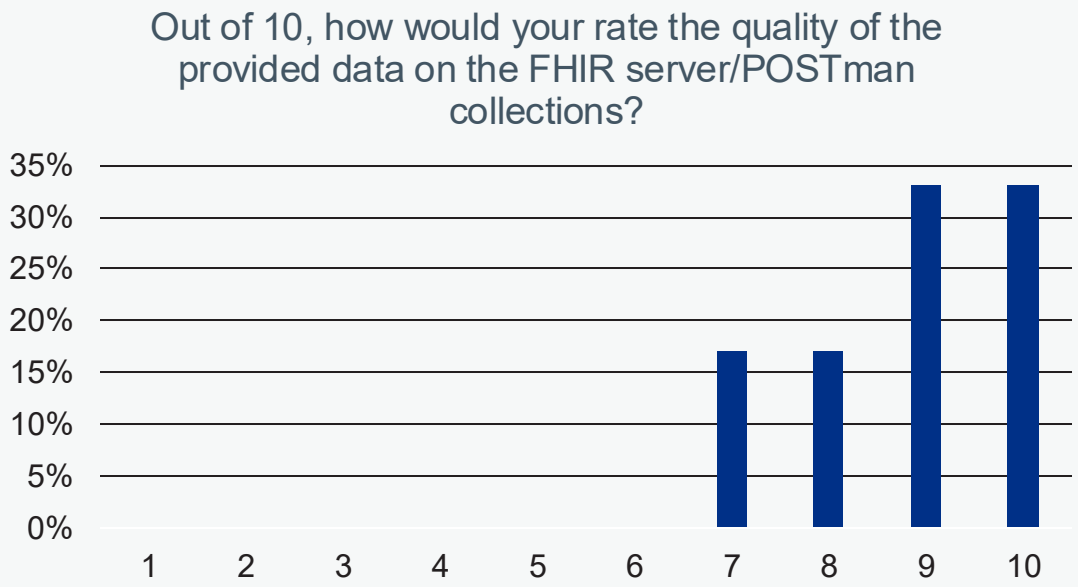
- 2 sessions held at TechUK in London – opportunity for suppliers to test workflows connected to the genomic order management service
  - 15th January
  - 21st February
- Approximately 60 people attended across the 2 dates, including Imperial, Royal Marsden, GOSH, GEL, broker developer Aire Logic, web portal developer Kainos, Phenotips, Labgnostics, GEL, Meditech, Cerner and EPIC
- Scenarios Tested
  - Rare Disease Singleton Non WGS & Rare Disease Singleton WGS
- Rare Singleton Non-WGS – proved - Imperial TIE handles HL7 v2-to-FHIR transformations, order transactions, and report retrieval between Cerner, Order Management API, and NHE GU, aligning with genomics FHIR IG specifications
- Rare Singleton WGS – proved - The user could submit a test request via the national portal, triggering FHIR-based processing through the Order Management API broker, NGIS, and NHE GU, with data integrated into TOMS and core services.

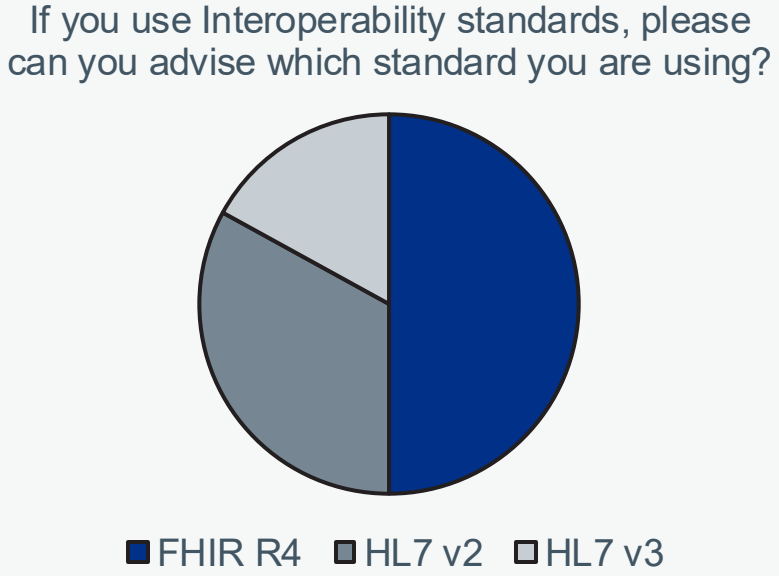
Connectathon - Feb 2025



Survey responses







Survey feedback

Question - What improvements/suggestions would you make to enhance future connectathon events?
Question - What could have been done to improve the documentation?

Connectathon

- Pre-allocated NHS numbers for connectathon testing
- Contents list of all documentation and their purpose to make it easier to locate information

Technical

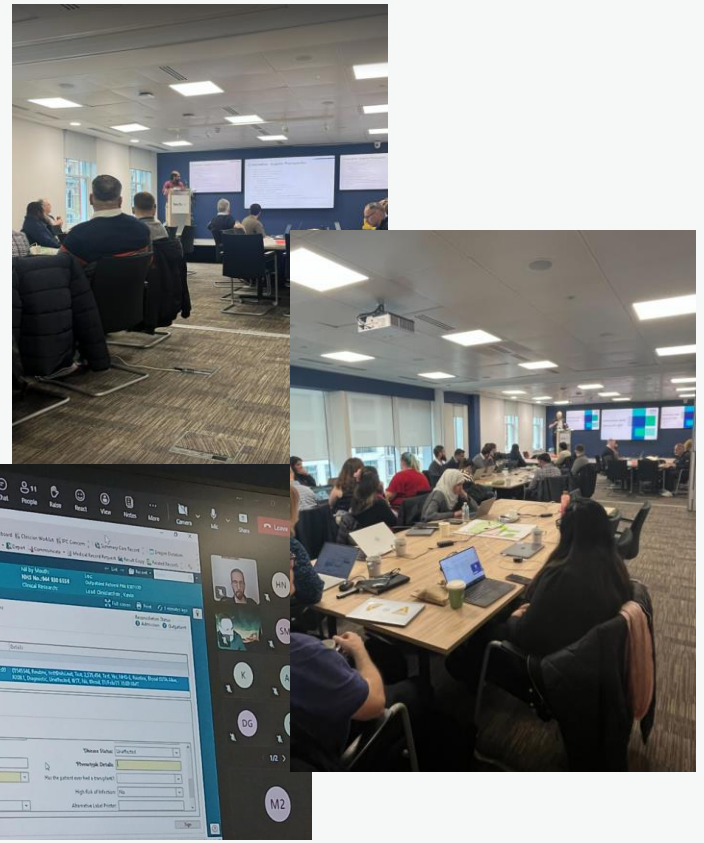
- Resolvable canonical URLs for FHIR assets

FHIR IG

- Structured reporting guidance

Follow-up

- Published roadmap/future plans
- Contacts for participants to follow up discussions



Developer Community Forum Comments

FHIR IG issues

- Correction to reason for testing mapping
- Inclusion of Condition resource in reasonCode
- Submission of data to central service vs. referencing local data

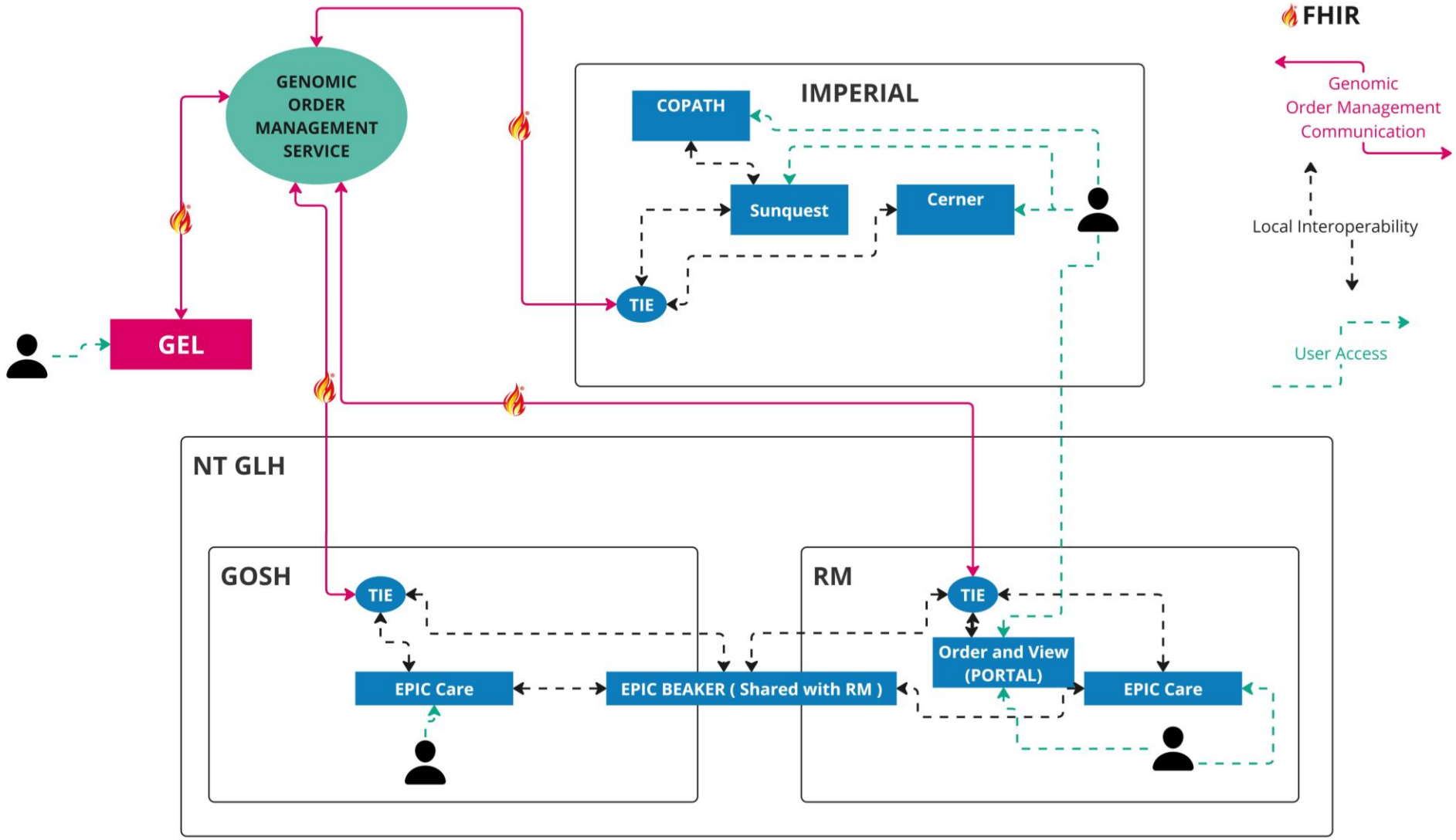
Broker capability

- if-match support – process for checking for existing resources (conditional updates)
- Transaction endpoint trailing slash
- Support of additional resource types outside MDS
- Response sizes when using embedded PDFs

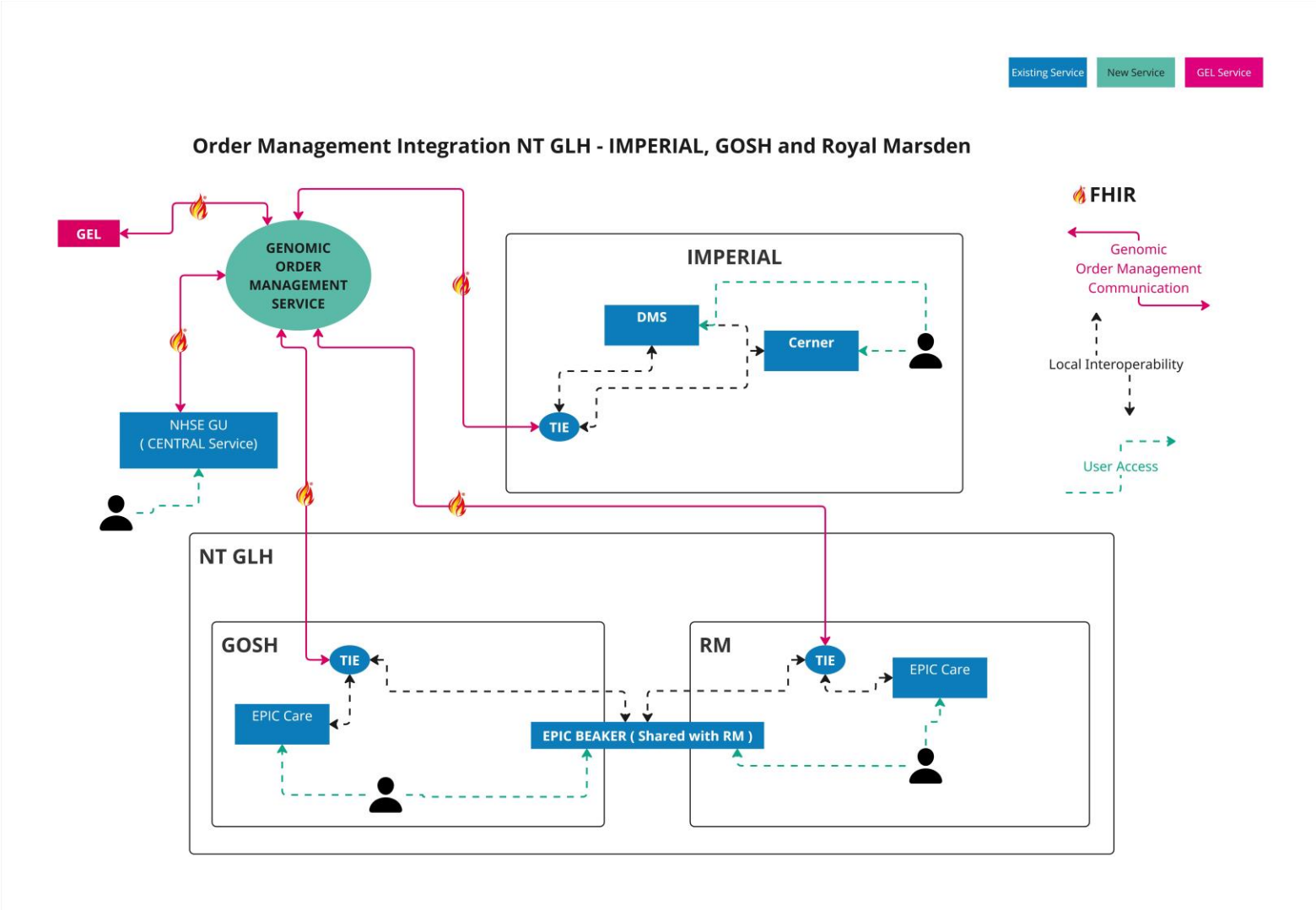
FHIR guidance

- Guidance on Task creation vs update – Task mapping to v2 and LIMS representation
- DocumentReference vs. DiagnosticReport for reports

North Thames GLH (NTGLH) Alpha integration



North Thames GLH - Current Implementation



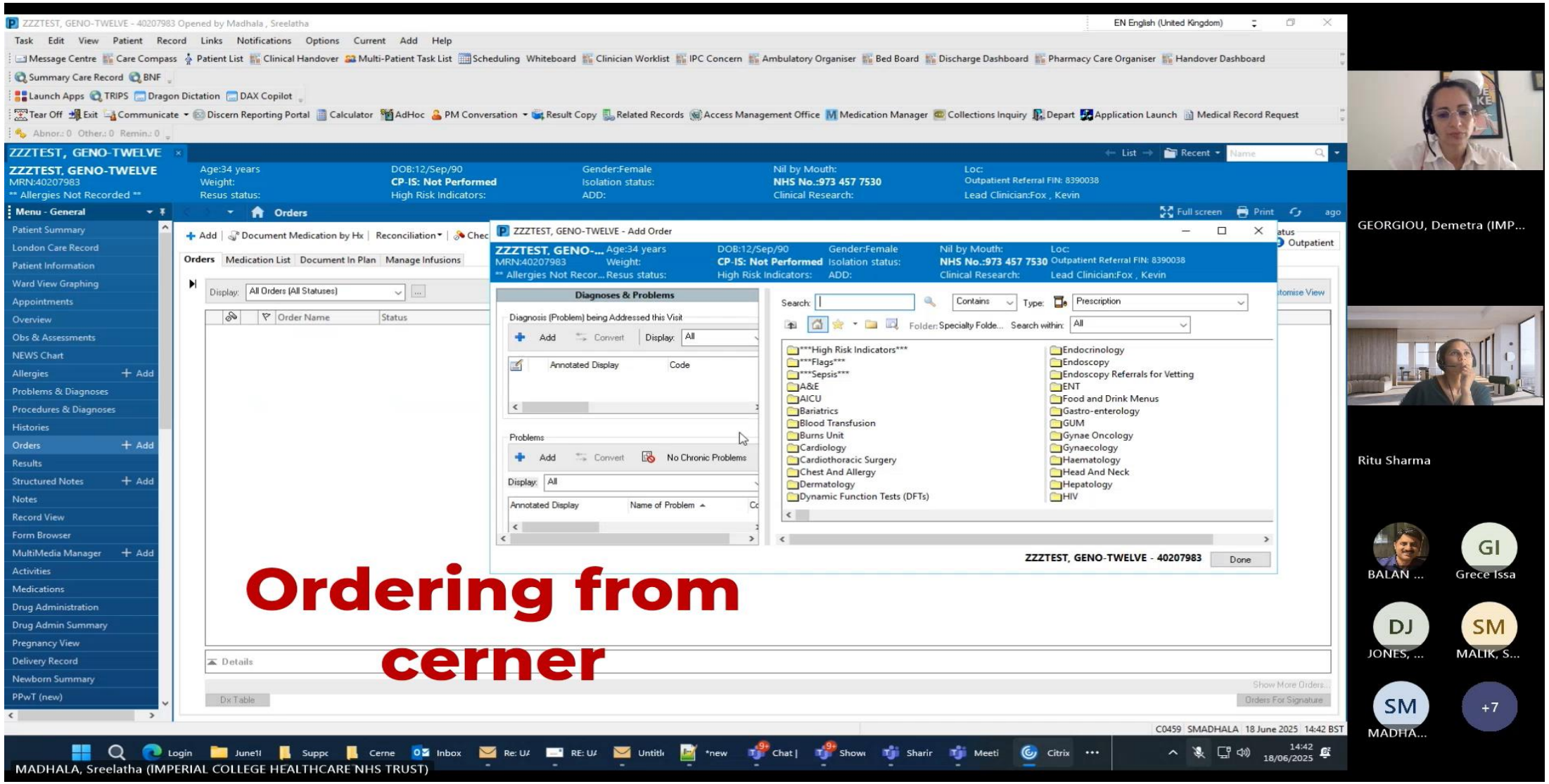
Alpha Connectathon Learnings

- Native FHIR adoption was limited
  - Mostly through translation
- API utilisation coverage is fair and needs more uptake
- Provider local systems that are home grown or developed internally challenge the interoperability
- IAM is challenging and would need accommodate local requirements
- Terminology use in terms of SNOMED, HPO had limited implementation have challenges in the oncoming phases for BETA/LIVE
- Broker implementation was stable and provided resilient service throughout the alpha phase.
- APIM use provided consistent gateway for API access
- Use of core services like PDS, ODS paving the way to other services adoption
  - MNS
  - PDM ...
- Notification using polling was limited
  - Would be addressed in BETA/LIVE using MNS

North Thames – Broker partner

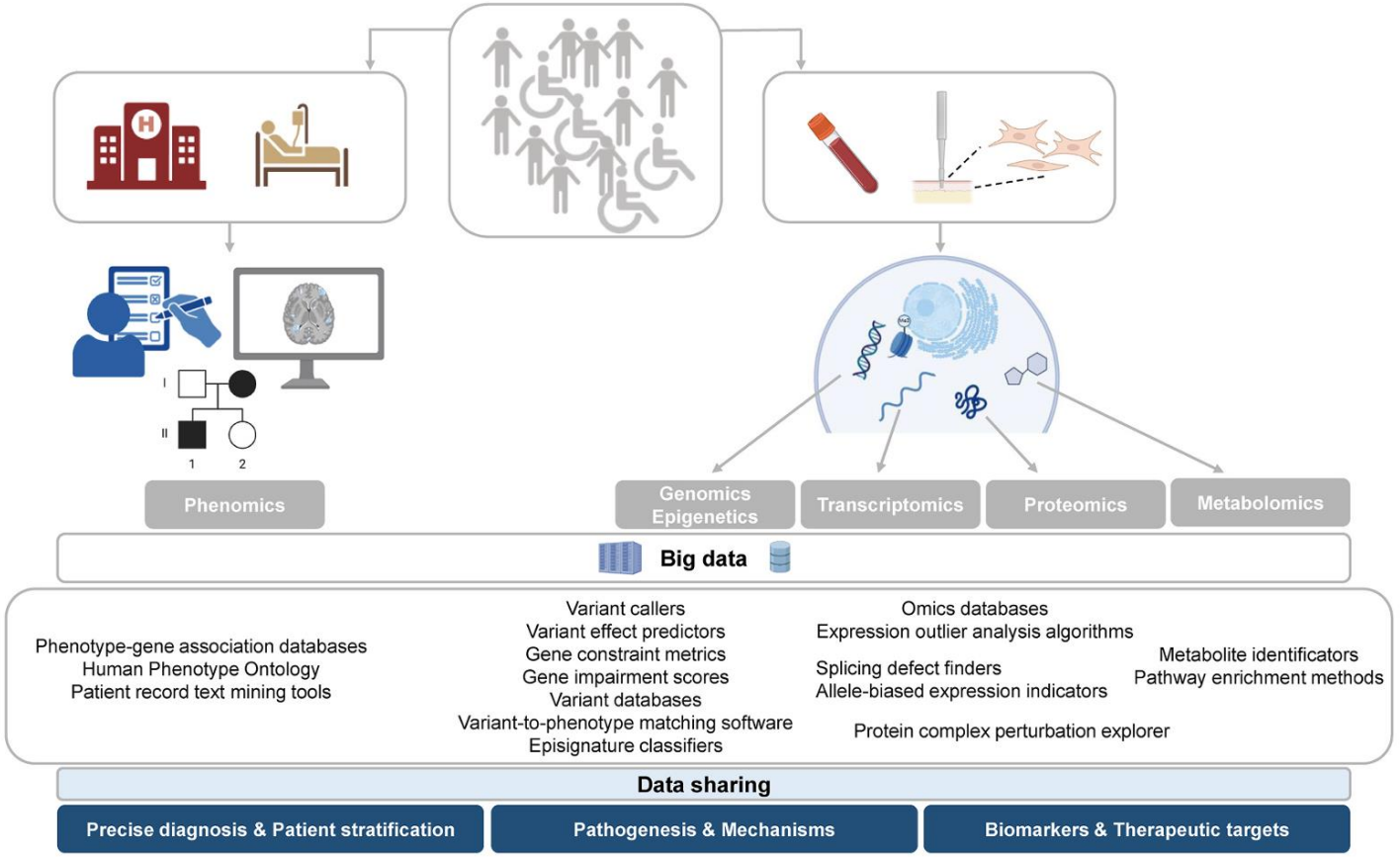
Demetra Georgiou - Imperial

North Thames GLH (NTGLH) Walkthrough

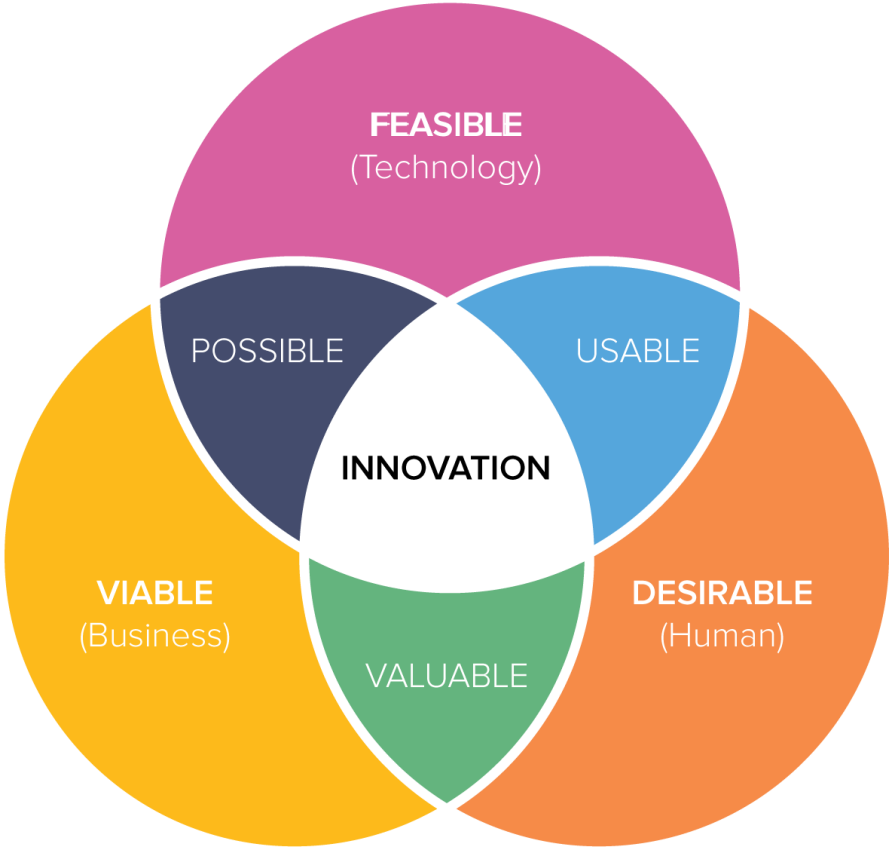


North Thames GLH (NTGLH)

- Why do we need all this?
  - Audit/governance/training/clinical care
  - Secondary uses (research, clinical trial)
- What if we do not go ahead?
  - Implications on patient care
  - Implications for research



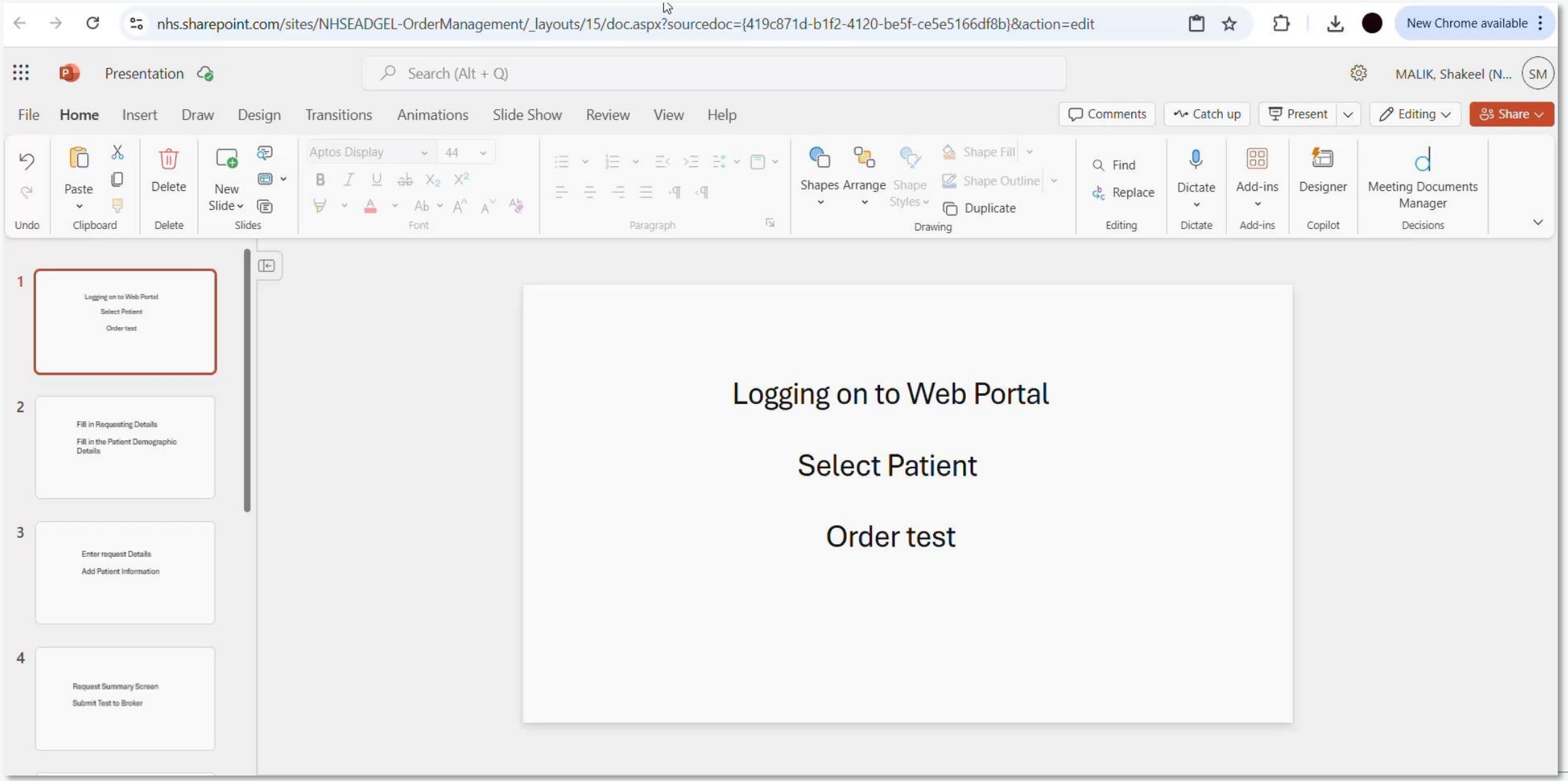
What do users need to see from GOM?



Alpha Web Portal

- We have completed UAT of the Alpha National Genomic Web Portal
- This is a Proof-of-Concept (POC) to prove key patterns and standards between a frontend system and the broker
- It has:
    - Matured technical and requirement articulation
    - Demonstrated key parts of the end-to-end process
    - Used a proxy digital Test Directory
    - Demonstrated basic routing
    - Demonstrated the usage of statuses and ownership
    - Helped prove the FHIR standards

Alpha Web Portal - Demo



Order Management

Transition Planning - Requirements

- A ground-up BRD has been written and will be distributed for wider feedback once final key requirements have been incorporated.
- A collection of requirements that were validated for Alpha but deferred to Beta will be included.
- UAT feedback for the portal has effectively been taken as a feed of new requirements.
- Some of the niche categories are being elaborated and require further validation. e.g. MIE & use of external identifiers

PLAN: Stable drafts for BRD and requirements catalogues by mid July, with opportunity to continue feedback loops until Private Beta advertises circa end of July

Order Management

Transition Planning - Documentation

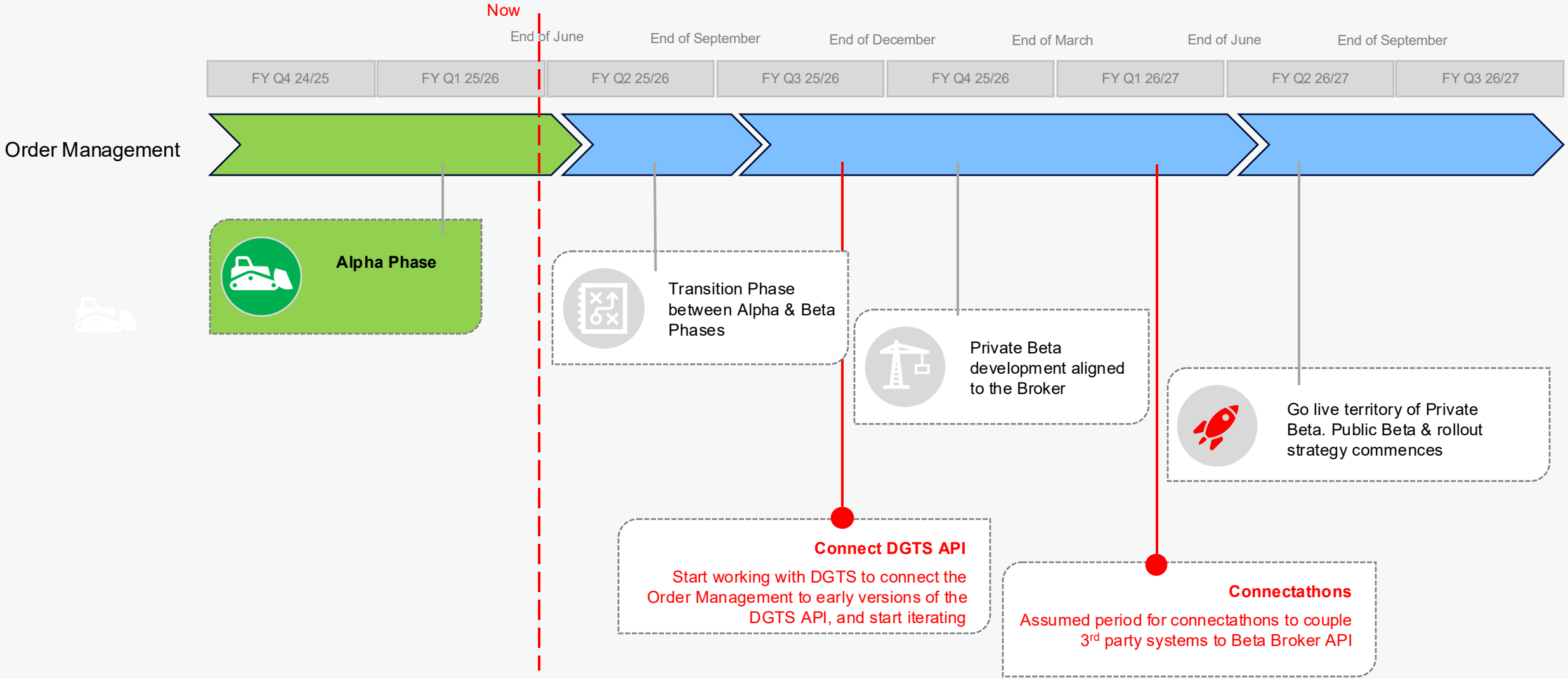
- High Level Architectural Design draft circulated for internal review
- Low Level Architectural Design in progress
- Alpha completion report in progress
- 1st draft of Alpha PID circulated internally
- Draft Private BETA BRD circulated for internal review

Order Management

Transition Planning - Other

- Lessons learned
- Clinical Safety case Documentation
- Governance
- Procurement process
- DWGS alignment
- MI requirements
- Rollout plans

Order Management – Plan on a Page



Call to action

Are your technical teams aware of the published Alpha APIs?

As the project continuous to evolve from Discovery to Alpha, and then into Beta practical feedback from organisations and suppliers will help ensure your voice is heard and thoughts considered in the design of features and capabilities.

Further connectathons?

Participants of the connecthathons in Q1 ‘25, and the subsequent demonstrations of the North Thames integration has shown how useful and successful connectathons are. Over the course of the next 18 months would suppliers like further connectathons?

How do we progress native interoperability in EPR & LIMS?

The Alpha has demonstrated the fundamental patterns and architecture. The work has demonstrated the ability to integrate in across the existing technology. But strategically, we want native FHIR interoperability out of the box with frontend systems.

How can you get involved?

Digital Genomic Test Service (DGTS)

Luisa Van Der End
Jim Phelps
Ravi Natarajan

DGTS Project Summary

Foundation

The Digital Genomics Test Service (DGTS) builds upon the current Genomic Test Directory, which is a critical foundation for genomic testing in England.

The Genomic Test Directory is currently maintained and published via Excel/PDF formats.

Supports a live national genomic testing service.

Why Digitise?

To enable a modern, scalable, and interoperable genomic testing service.

Digitisation supports better integration across EPRs, LIMS, and NHS systems.

Scope of Work

New data model: Transitioning from excel to a formal, scalable structure allows improved quality, standardisation, and enhancement.

Authoring environment: A digital platform to manage test additions and updates in a controlled, auditable way.

Data transformation: Mapping and aligning legacy directory content to the new model, ensuring clinical accuracy and validation readiness.

Progress to Date

MVP build underway:

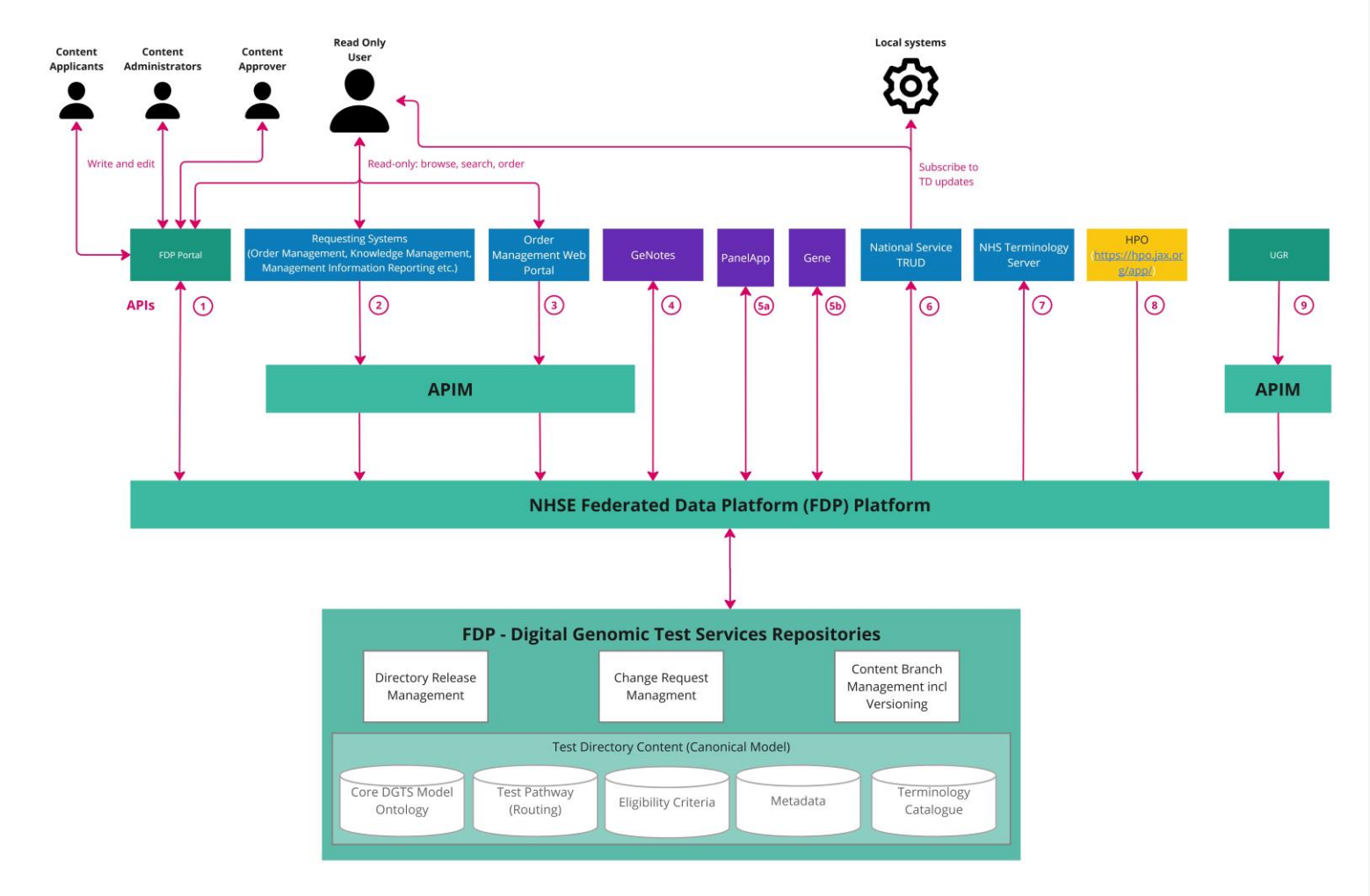
First class searching and browsing capabilities and frontends for viewing all directory data.

Integrations with external reference sources to enrich test information (e.g. PanelApp and HGNC Genenames).

Carefully managed transition to protect existing services and systems in use across the NHS.

DGTS Architecture

Federated Data Platform & Service Integration Overview



High-level architecture of the Digital Genomic Test Service (DGTS) integrated with NHSE's Federated Data Platform

FDP Capabilities

- Content management
- Validation
- Data Ingestion
- Data Pipelines
- Mapping
- Meta model
- Roles management (NHS login)
- Versioning & status handling
- Routing & PLCM
- NHS-compliant Web UI

APIM API Interactions

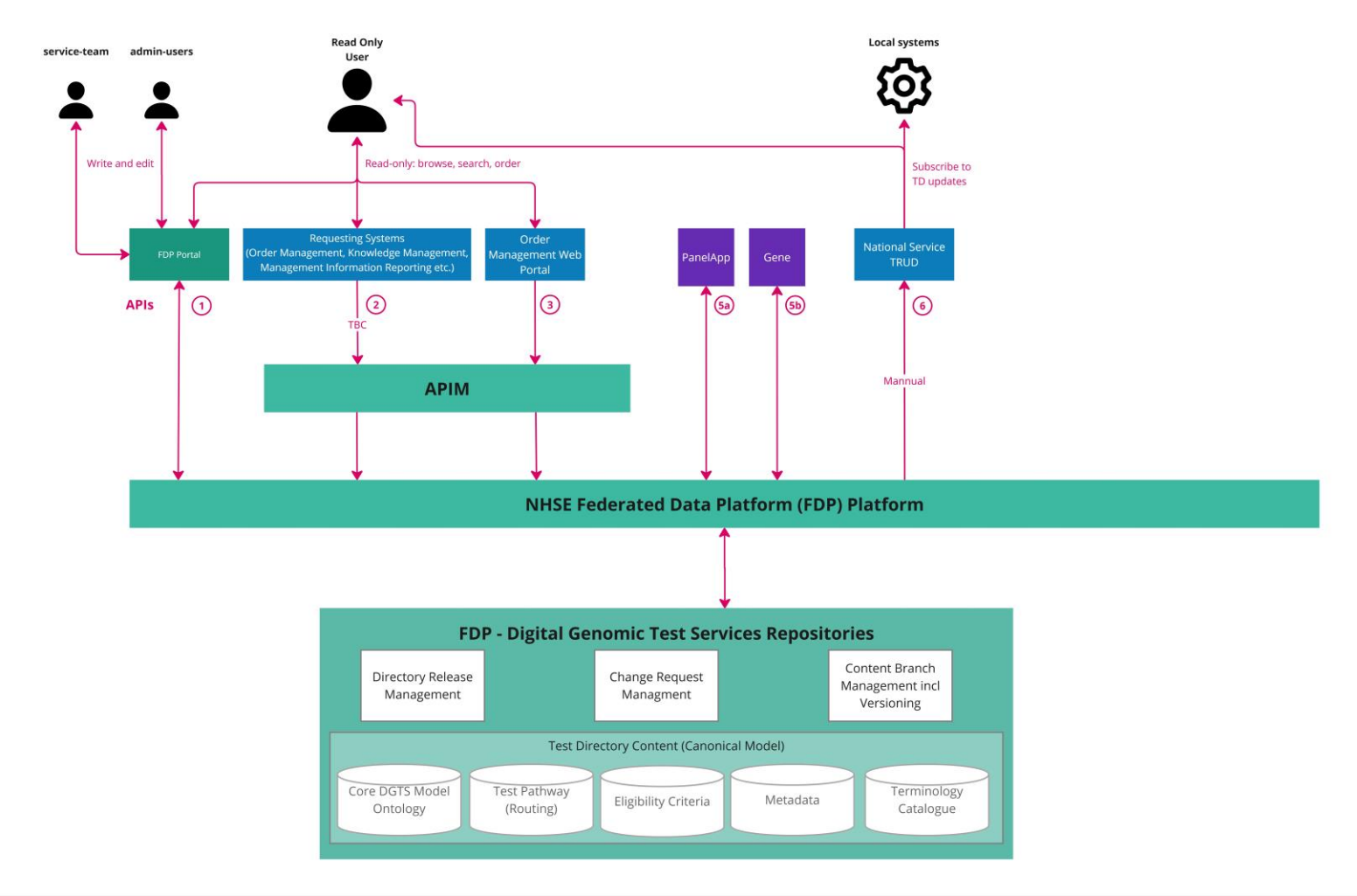
- Order Management
- Unified Genomic Record (UGR)
- Other integration endpoints

Release Process

- Streamlined via TRUD

DGTS Architecture - MVP

Focused MVP Delivery - Federated Data Platform & Service Integration Overview



Architecture

- Adhering to the final vision
- FDP Adoption for major features
- API interaction through API
- TRUD Release
- Draft DGTS standards for stakeholder feedback

High-level architecture of the Digital Genomic Test Service (DGTS) integrated with NHSE's Federated Data Platform

DGTS MVP Objectives

Primary Objective

The primary objective of the MVP delivery is to create a scalable, interoperable, central infrastructure that will provide access to all test centric reference data within the national GMS while meeting requirements and expectations around governance, audit and access.

MVP Objective

Replace Excel and PDF as the means to maintain and manage genomic test data.

Create a dynamic DGTS data model that is structured, normalised and expandable to represent the core definition and scope of the NHS England portfolio of genomic tests and their associated metadata

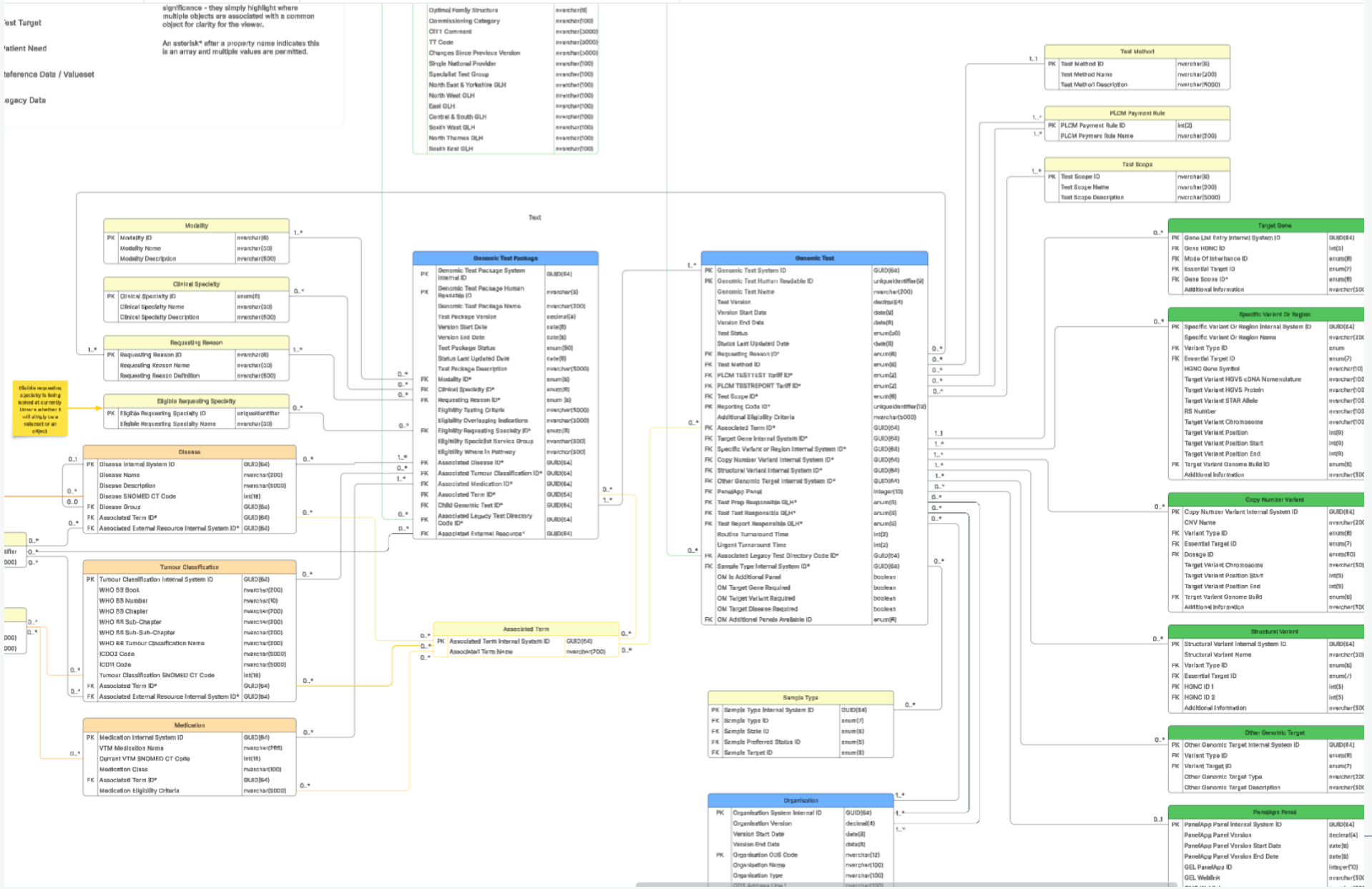
Fully transform and enrich legacy data in line with the new DGTS data model.

Update the portfolio of genomic tests to align with modern scientific and informatic practice.

Improve searchability and richness of data for all users including clinicians and patients.

Version control for individual genomic tests with fully audited changes.

Back-end: data ingestion and configuration



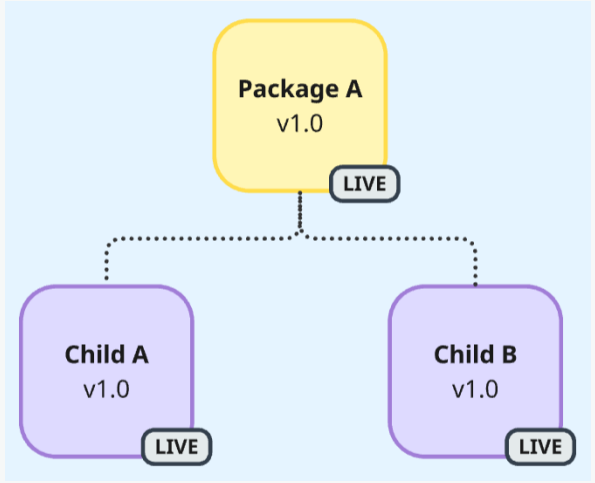
A **relational data model was created** with an agreed set of objects and associated properties that enables the DGTS to meet various business requirements.

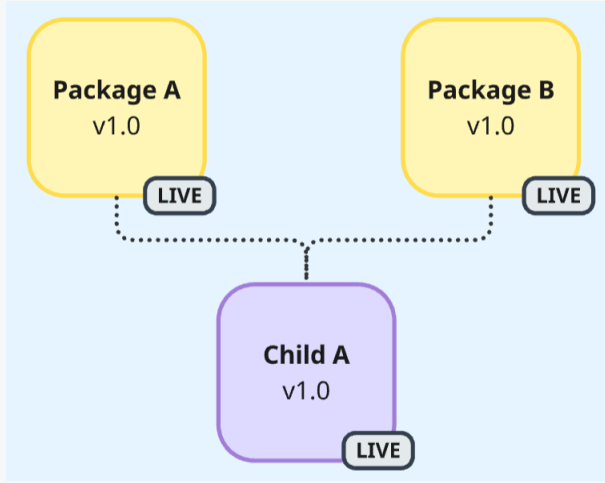
The model includes:

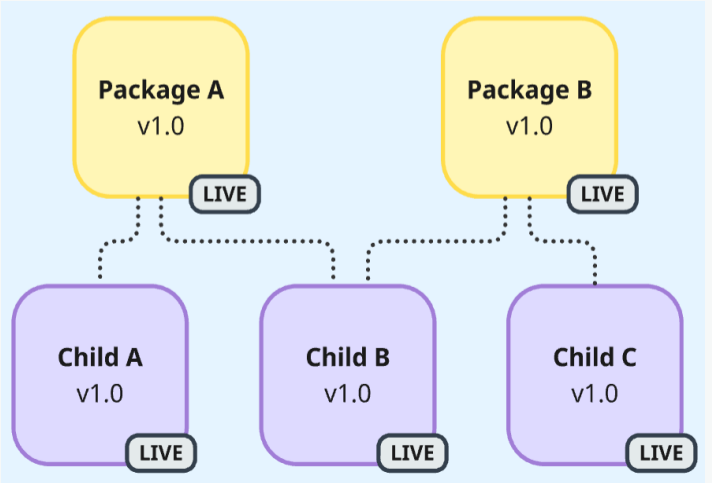
- **Core concepts** such as genomic tests and test packages.
- **Patient needs**: diseases, cancer type, medication.
- **Test targets**: genes, variants, panels.
- **Reference data**: HGNC genes, copy number variants, panels.
- **Valuesets**: consistent representation of key properties.
- Descriptions of 1..1, 1..* and *..* relationships.

Back-end: data ingestion and configuration

1. DGTS data model has been implemented into the Federated Data Platform (FDP) and all objects and properties created to form the DGTS Database. This includes the top-level architecture of test packages and child genomic tests.
2. A genomic test is a single investigation into one or more genomic targets to support delivery of patient care, delivered in a lab via a single test method and commissioned by NHS England.
3. A test package is a collection of one or more associated genomic tests.







4. This differs to the current approach where the same test can be duplicated numerous times in the Test Directory and each individual test must be kept up to date.

5. Every test package and test has a defined set of structured properties ensuring consistency across the portfolio.

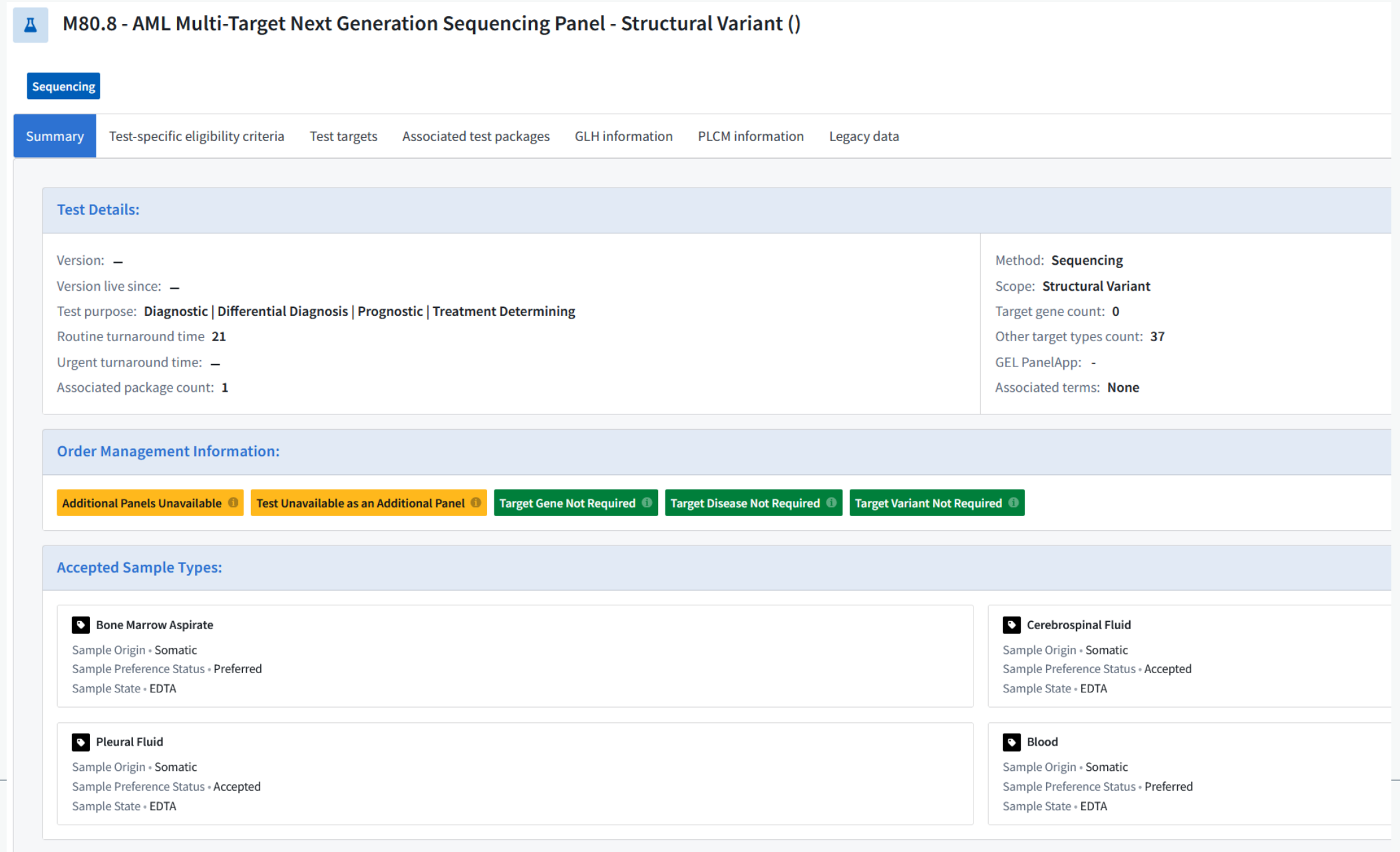
Back-end: data ingestion and configuration

1. The DGTS Database has been populated with all **legacy data -** all tests from the most recent release of the NGTD spreadsheets and PDF.
2. A **data transformation and validation app** has been created, allowing users to transform legacy tests into the new data model with relational aliasing and backward compatibility.
3. Ultimately, we are changing this:

Test Code	Test Name	Target Gene(s) [essential]	Test Scope	Technology
M80.8	Multi-target NGS panel - structural variant (To include detection of t(15;17)(q24;q21) PML-RARA , t(8;21)(q22;q22) RUNX1-RUNX1T1, inv(16)(p13.1q22) CBFB-MYH11, t(9;11)(p21;q23) MLLT3-KMT2A & other 11q23.3 (MLL i.e. KMT2A) rearrangements, t(6;9)(p22;q34) DEK-NUP214, inv(3)(q21q26) GATA2-MECOM, t(1;22)(p13;q13) RBM15-MRTFA, t(9;22)(q34;q11) BCR-ABL1, Complex karyotype, t(3;5)(q25;q34) NPM1-MLF1, t(5;11)(q35;p15.5) NUP98-NSD1, t(7;12)(q36;p13) MNX1-ETV6, inv(16)(p13.3q24.3) CBFA2T3-GLIS2), other NUP98 rearrangements,	To include detection of t(15;17)(q24;q21) PML-RARA , t(8;21)(q22;q22) RUNX1-RUNX1T1, inv(16)(p13.1q22) CBFB-MYH11, t(9;11)(p21;q23) MLLT3-KMT2A & other 11q23.3 (MLL i.e. KMT2A) rearrangements, t(6;9)(p22;q34) DEK-NUP214, inv(3)(q21q26) GATA2-MECOM, t(1;22)(p13;q13) RBM15-MRTFA, t(9;22)(q34;q11) BCR-ABL1, Complex karyotype, t(3;5)(q25;q34) NPM1-MLF1, t(5;11)(q35;p15.5) NUP98-NSD1, t(7;12)(q36;p13) MNX1-ETV6, inv(16)(p13.3q24.3) CBFA2T3-GLIS2, NUP98 rearrangements other than NUP98-NSD1	Structural variant detection	Panel

4. Into this...

Back-end: data ingestion & configuration



M80.8 - AML Multi-Target Next Generation Sequencing Panel - Structural Variant ()

Sequencing

Summary | Test-specific eligibility criteria | Test targets | Associated test packages | GLH information | PLCM information | Legacy data

Test Details:

Version: –
Version live since: –
Test purpose: **Diagnostic | Differential Diagnosis | Prognostic | Treatment Determining**
Routine turnaround time **21**
Urgent turnaround time: –
Associated package count: **1**

Method: **Sequencing**
Scope: **Structural Variant**
Target gene count: **0**
Other target types count: **37**
GEL PanelApp: -
Associated terms: **None**

Order Management Information:

Additional Panels Unavailable | Test Unavailable as an Additional Panel | Target Gene Not Required | Target Disease Not Required | Target Variant Not Required

Accepted Sample Types:

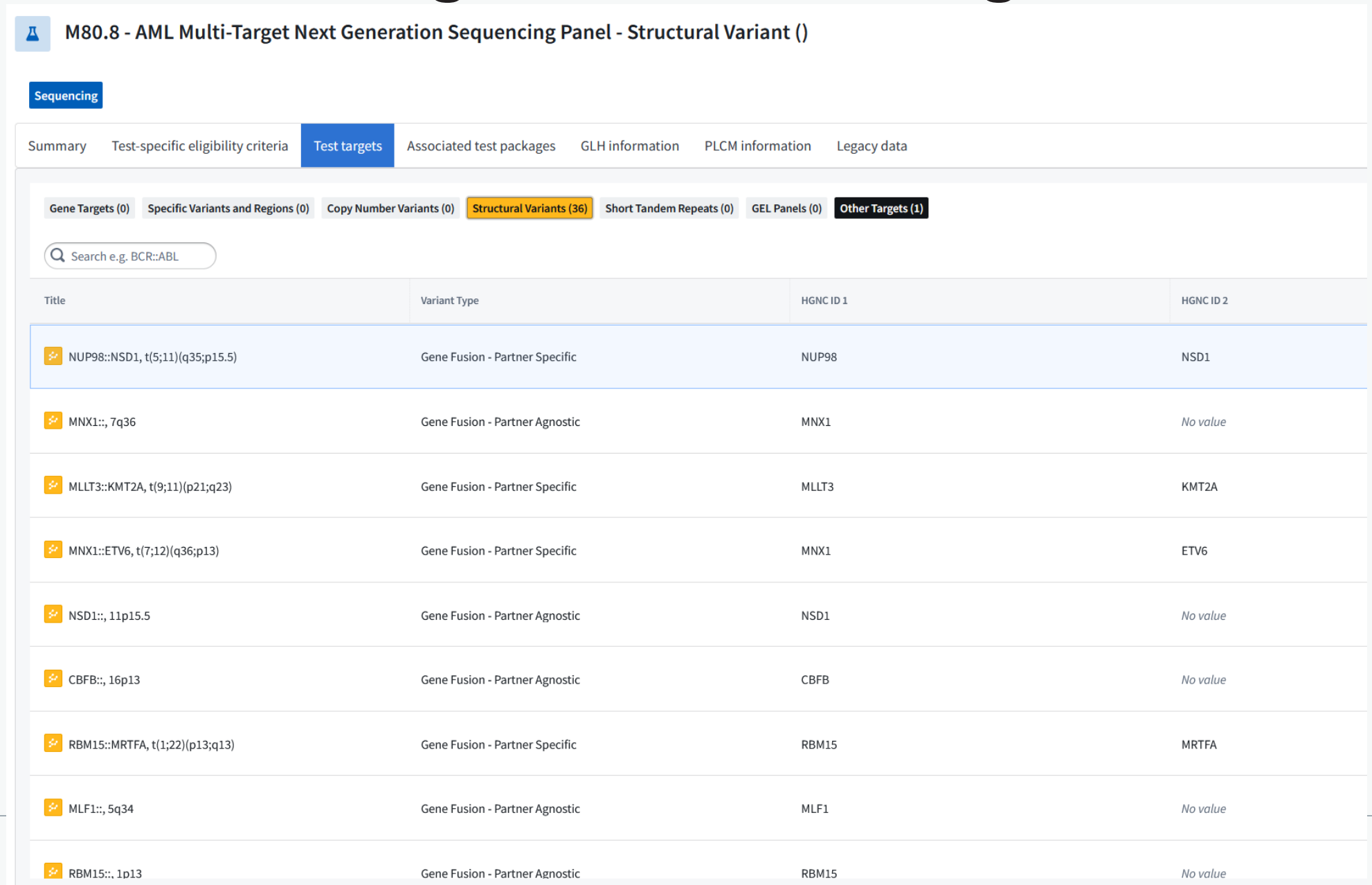
Bone Marrow Aspirate
Sample Origin • Somatic
Sample Preference Status • Preferred
Sample State • EDTA

Cerebrospinal Fluid
Sample Origin • Somatic
Sample Preference Status • Accepted
Sample State • EDTA

Pleural Fluid
Sample Origin • Somatic
Sample Preference Status • Accepted
Sample State • EDTA

Blood
Sample Origin • Somatic
Sample Preference Status • Preferred
Sample State • EDTA

Back-end: data ingestion and configuration

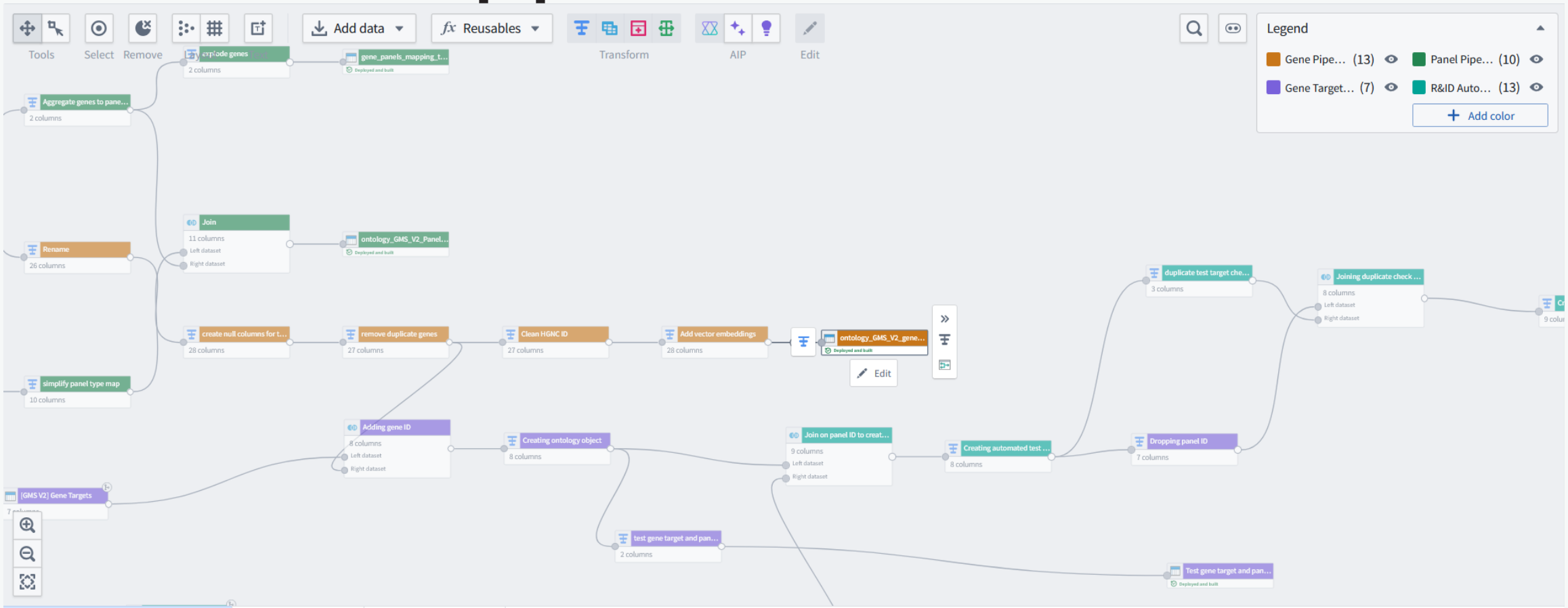


Back-end: data ingestion and configuration

We have also ingested the following data into the DGTS database:

1. **GEL PanelApp data**: API integration complete with all GMS signed-off panels being pulled in and checked on daily basis.
2. **HGNC gene reference data**: API integration complete, creating database of 44,000 genes with various metadata. Schedule tbc.
3. **ClinGen ISCA regions**: manual ingestion of the Dosage Sensitivity Curated Region List automatically from ClinGen's FTP resource. Schedule tbc.
4. **SNOMED CT**: subscribed to NHS Technology Reference Update Distribution (TRUD) to bring in full list of UK SNOMED CT terms. The best ingestion method and schedule tbc.

Back-end: data pipelines



Selection preview | Preview | Suggestions 1 | Pipeline warnings

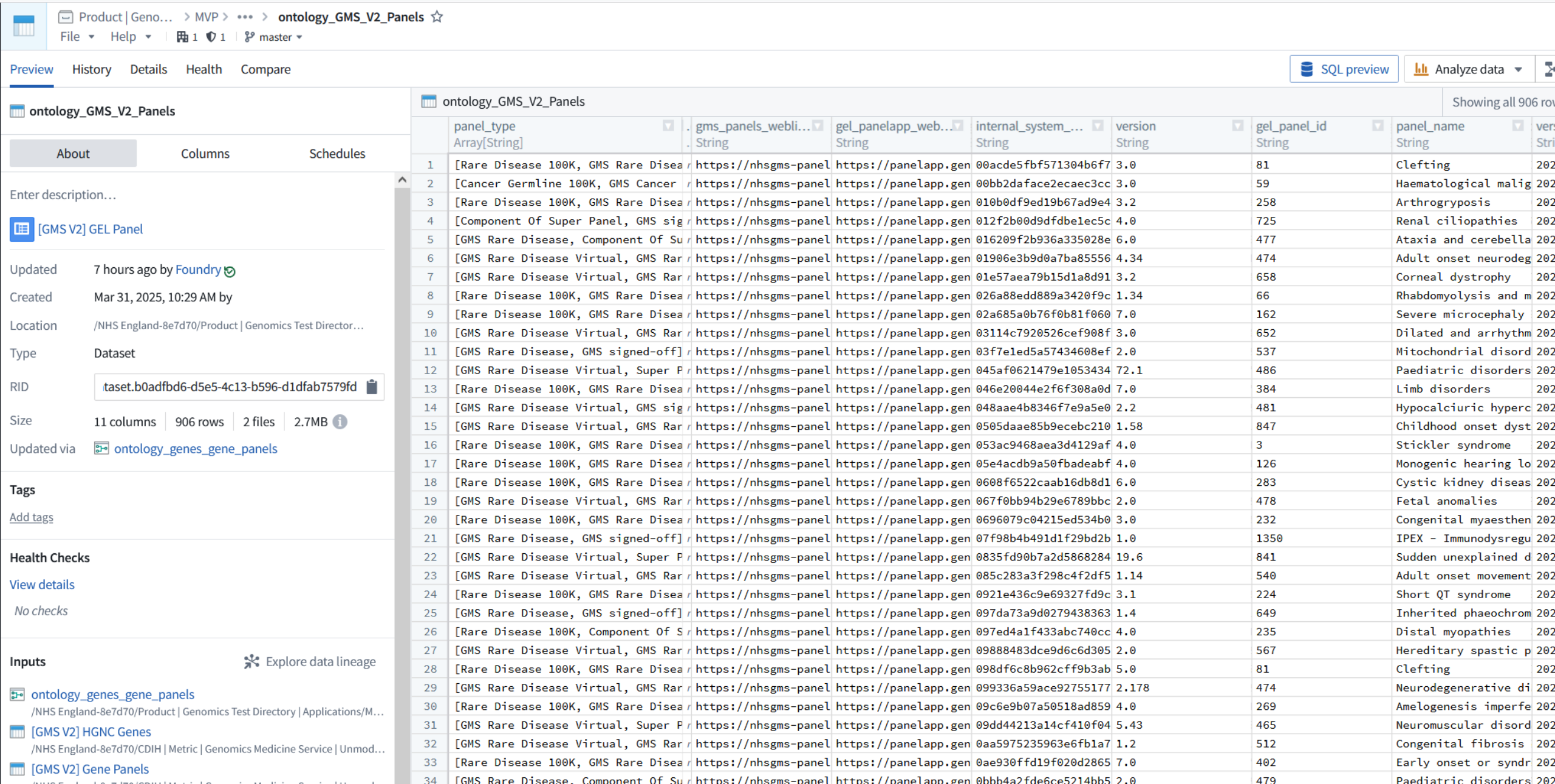
ontology_GMS_V2_genes — Previewing 10 rows — 28 columns — Search columns...

No inputs sampled

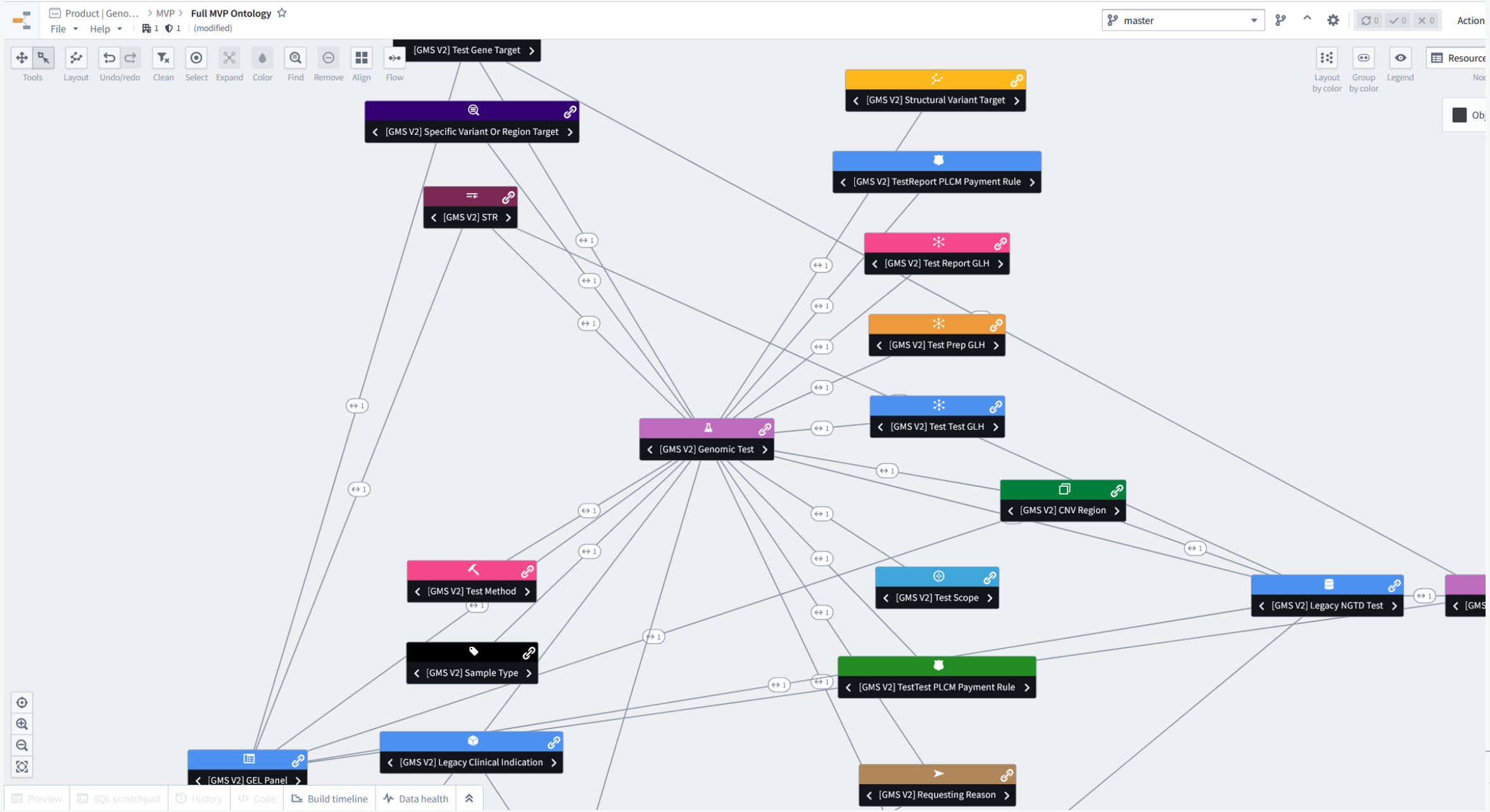
	vector Embedded vector	gene_id String	hgnc_release_dat... Date	gene_version String	variant_type String	hgnc_uuid String	hgnc_gene_aliase... Array[String]	hgnc_prev_symbol Array[String]	hgnc_mane_select Array[String]	internal_system_... String	hgnc_ge String
1	[-0.026402276, -0.02	10062	null	null	null	40cfface-d2f2-49a7-9	[FLJ20382, FLJ11189,	null	[ENST00000389120.8,	956ea3ab85a324143851	RNF20
2	[-0.016236302, -0.00	10084	null	null	null	76d2febe-a5f6-42e4-b	null	null	null	dd22eeacfef994f0496a	RNR3
3	[-0.025597235, -0.01	10099	null	null	null	0fb7a534-a77c-4dfb-a	[U102]	[RNU102]	null	f1ad2066c123ff3e296f	SNORD10
4	[0.016125077, 0.0076	1012	null	null	null	e5345cf0-f45b-4492-9	[BOP, CBT]	[BCP]	[ENST00000249389.3,	5bd1ab3a489b8b8d2de7	OPN1SW
5	[-0.007515428, -0.02	10123	null	null	null	efb324ea-ef78-4787-8	[U1C1, U1C21]	[RNU1C1, RNU1C2]	null	66a68c19e3c156e2b7a0	RNU1-2
6	[0.0024001542, -0.01	10140	null	null	null	895115f8-5c9e-406d-9	[U1P11]	[RNU1P8]	null	cd173a5	5
7	[-0.01591158, -0.016	10152	null	null	null	f64c187c-e548-48b3-b	[U2]	[RNU2B, RNU2-2P]	null	f3af1ed	

Calculate row count

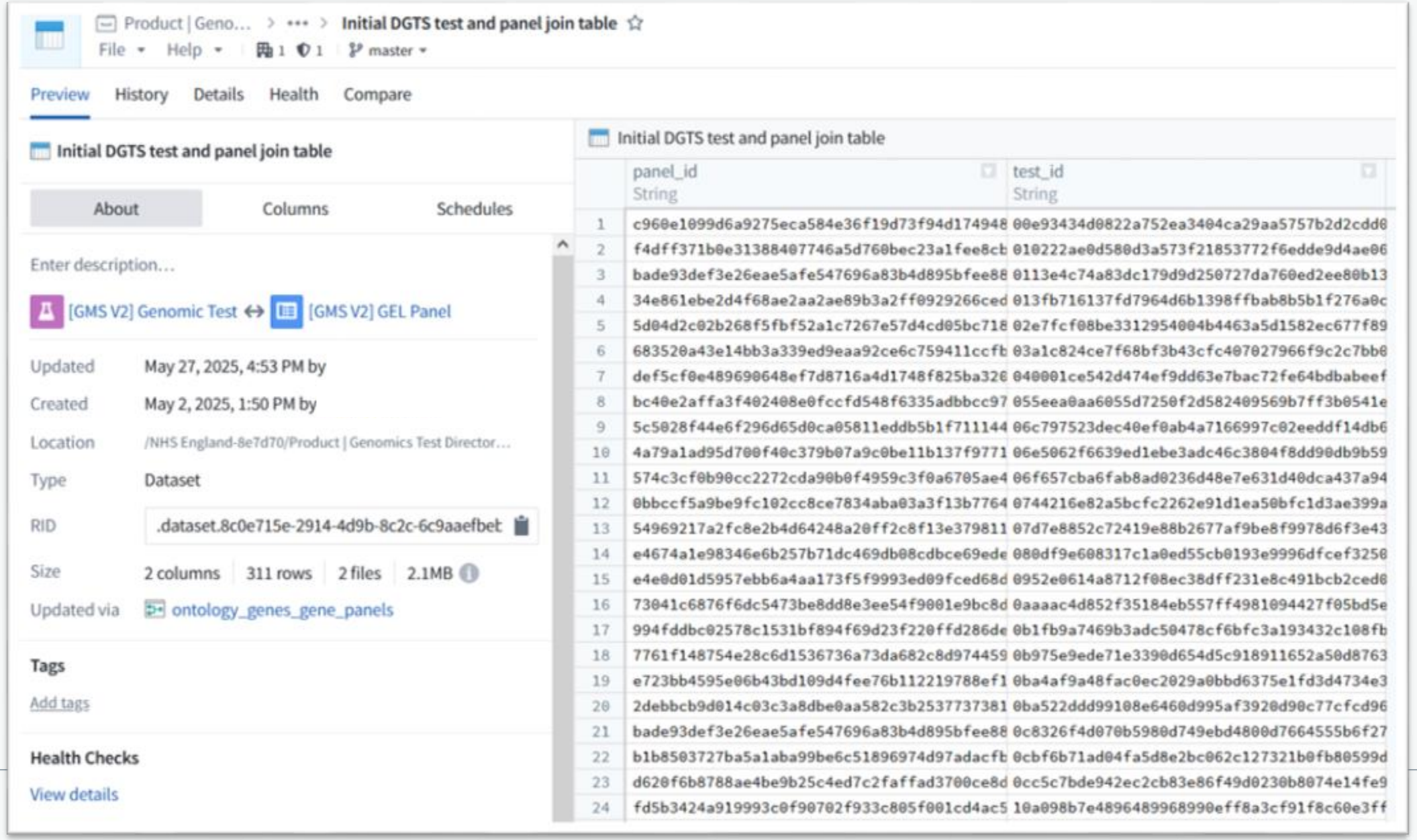
Back-end: object tables



Back-end: relational linkages



Back-end: join tables



Front-end

1. A front-end web application is being developed to allow users to search and interact with DGTS data. The vision is to create a solution akin to Wikipedia for genomic testing in England.
2. So far, we have built:
    a) Overall landing page for the service.
    b) A flexible 'search everything' capability which matches the user input string to a wide variety of properties in the data model with fuzzy matching using vectorisation.
    c) A gene browser where users can search for genes and view related tests.
    d) Object views to display genomic tests, test packages and genes.
    e) Basic navigation flows between objects.
3. We want to create:
    a) Refine existing object views and create views for disease, tumour classifications, medications etc.
    b) Refine and expand search results pages and navigation flows
    c) Create tabular and filterable views where users can browse lists and refine the list based on their chosen filters. For example: filtering a list of test packages by their associated clinical specialty.
    d) Overall improvements to functionality, usability and appearance.

Front-end

DEMO

How will the DGTS impact you?

1. Front-end web application
2. Test packages and child genomic tests and changes to portfolio
3. Test identifiers
4. New Excel and PDF files
5. Test-level versioning and status
6. DGTS API
7. Interactions with Order Management

How will the DGTS impact you?

1. **Front-end web application**
    - **Users move away from spreadsheets, PDFs and local apps and begin interacting with the DGTS online.**
    - **Users can still access spreadsheets and PDFs but will be directed to the web app.**
2. Test packages and child genomic tests and changes to portfolio
3. Test identifiers
4. New Excel and PDF files
5. Test-level versioning and status
6. DGTS API
7. Interactions with Order Management

How will the DGTS impact you?

1. Front-end web application
2. **Test packages and child genomic tests and changes to portfolio**
    - **Significant overlap to existing clinical indications and individual tests, however...**
    - **The process of data transformation and portfolio evaluation means the some tests are merged, some deactivated, some tests available in multiple test packages, some new test packages created.**
    - **Numerous changes to test targets.**
    - **Expectation that users will order at a test package level, with GLH staff selecting appropriate tests for the patient. Users can still order individual tests but we know the service will mainstream over time and user knowledge will decrease.**
3. Test identifiers
4. New Excel and PDF files
5. Test-level versioning and status
6. DGTS API
7. Interactions with Order Management

How will the DGTS impact you?

1. Front-end web application
2. Test packages and child genomic tests and changes to portfolio
3. **Test identifiers**
    - **Currently R and M codes are used to identify each test e.g. R100.1, M80.8**
    - **We are exploring whether these should change to a common identifier for all test packages and all tests.**
    - **With tests such as DPYD available in different test packages, which test ID should be kept? e.g. DPYD appears as M1.7, M3.7, and M6.5. Which should be kept?**
    - **Could these change to P1.T1, P3.T1 and P6.T1? (with P indicating package number and T1 indicating the DPYD test)**
4. New Excel and PDF files
5. Test-level versioning and status
6. DGTS API
7. Interactions with Order Management

How will the DGTS impact you?

1. Front-end web application
2. Test packages and child genomic tests and changes to portfolio
3. Test identifiers
4. **New Excel and PDF files**
    - **The existing Excel and PDF files will be generated by an export of the DGTS data rather than being maintained manually and made available via TRUD, where users and organisations can subscribe to and be notified of DGTS updates.**
    - **While the system will aim to match the existing structure, the export file will have changes. It is hoped most of these are quality of life improvements such as no tabs, no merged cells.**
    - **New information from the DGTS will be appended in additional columns to give more data to users.**
5. Test-level versioning and status
6. DGTS API
7. Interactions with Order Management

How will the DGTS impact you?

1. Front-end web application
2. Test packages and child genomic tests and changes to portfolio
3. Test identifiers
4. New Excel and PDF files
5. **Test-level versioning and status**
    - **It is difficult to properly track additions, changes and removals in the Test Directory spreadsheets and PDF.**
    - **Status and version control will be introduced at a test and test package level, with status indicating whether an object is available to order, has been retired or is due to be retired. Certain statuses may be restricted to certain user types e.g. GLHs can view all information about a genomic test or package before it is released.**
    - **Change to process of test retirement: tests are now decommissioned with a target date for retirement, beyond which no new orders are accepted. No date given to GLHs for when they can no longer accept an order for a retired test.**
    - **Ability to see how a test or test package appeared on a particular day in the past and how it has changed over time.**
6. DGTS API
7. Interactions with Order Management

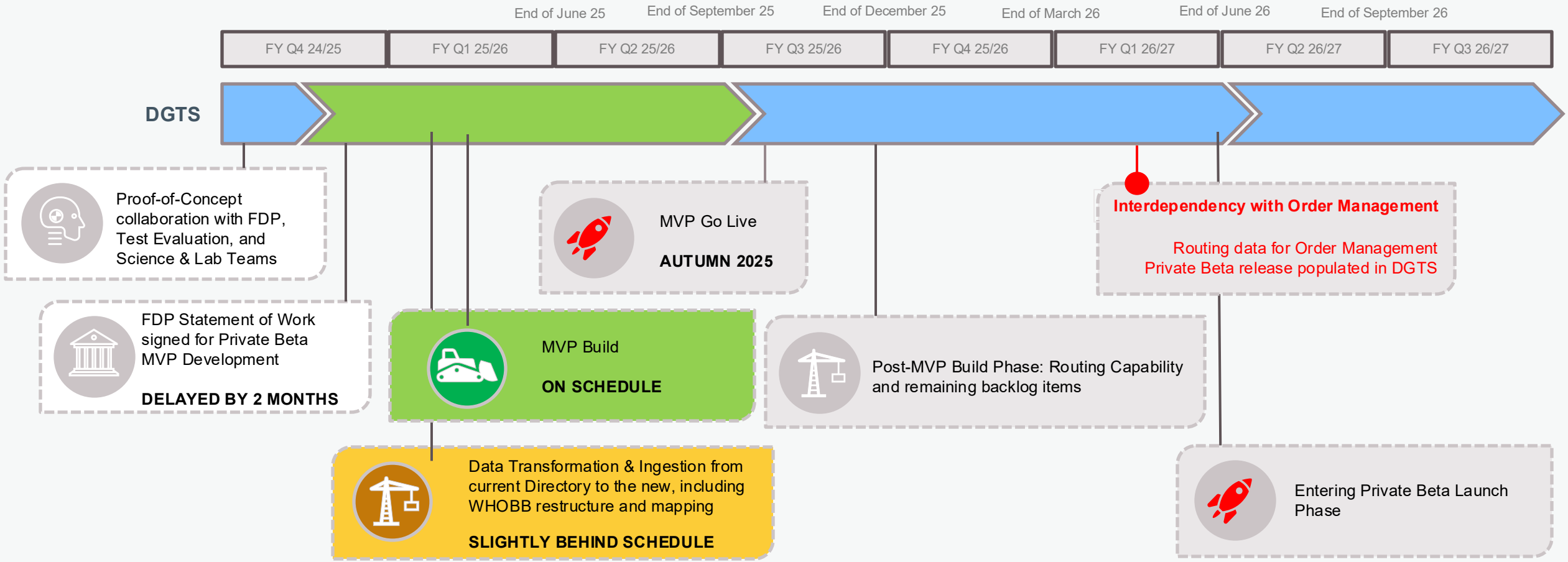
How will the DGTS impact you?

1. Front-end web application
2. Test packages and child genomic tests and changes to portfolio
3. Test identifiers
4. New Excel and PDF files
5. Test-level versioning and status
6. **DGTS API**
    - **DGTS reference data will be exposed via API.**
    - **Exact methods of access are to be agreed (e.g. is it open access or are keys required per app or per user? Is there any data that shouldn't be exposed?)**
    - **API documentation will be held within the DGTS and freely accessible.**
    - **What implementation guidance is helpful to vendors?**
7. Interactions with Order Management

How will the DGTS impact you?

1. Front-end web application
2. Test packages and child genomic tests and changes to portfolio
3. Test identifiers
4. New Excel and PDF files
5. Test-level versioning and status
6. DGTS API
7. **Interactions with Order Management**
    - **As OM delivery matures it needs to begin using DGTS data to check test package and test identifiers are valid and available to order.**
    - **There are other fields in DGTS that OM could consume to improve order quality such as when a Record of Discussion should be completed, when target variant needs to be provided etc.**
    - **Information about routing should be held in DGTS and consumed by OM.**

Digital Genomics Test Directory - Plan on a Page



Call to action

How do suppliers want to be involved?

By autumn 2025 the project will have transformed the data in the Test Directory from the existing binary file structure to the new data model. How do suppliers want to engage to understand how this affects them?

Future DGTS capabilities?

In 2026, DGTS is planned to be operational. Beyond publishing the binary files an API layer will be available to allow appropriate systems to interrogate the data in the DGTS. How will this benefit use cases in downstream supplier systems?

How can you get involved?

Unified Genomic Record (UGR)

Adam Laurent
Ravi Natarajan
Hamish Price

UGR Project Summary

Foundation

The UGR provides a single point of administration and access to genomic data for each patient.

It is a single point of entry to a concept consisting of various components.

Why Digitise?

Digitising the UGR realises the lifetime value of genomic reporting, enabling access for:

- Direct care
- Population health
- Research
- Operational insight

Scope of Work

The Discovery phase, focused on three priority use cases to prove out the value of the UGR.

- Pharmacogenomics
- Cancer Clinical Trials
- Inherited Cardiovascular Conditions

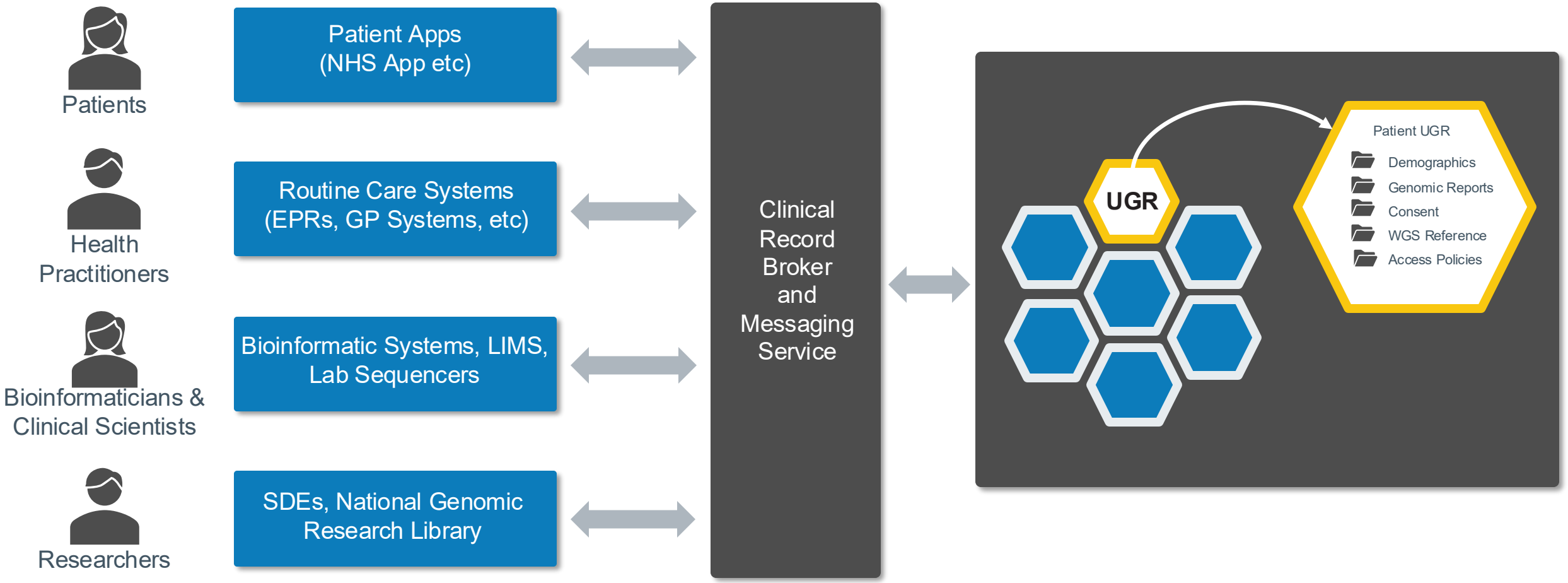
SPR and UGR

Discussions regarding the development of a Single Patient Record (SPR) are underway.

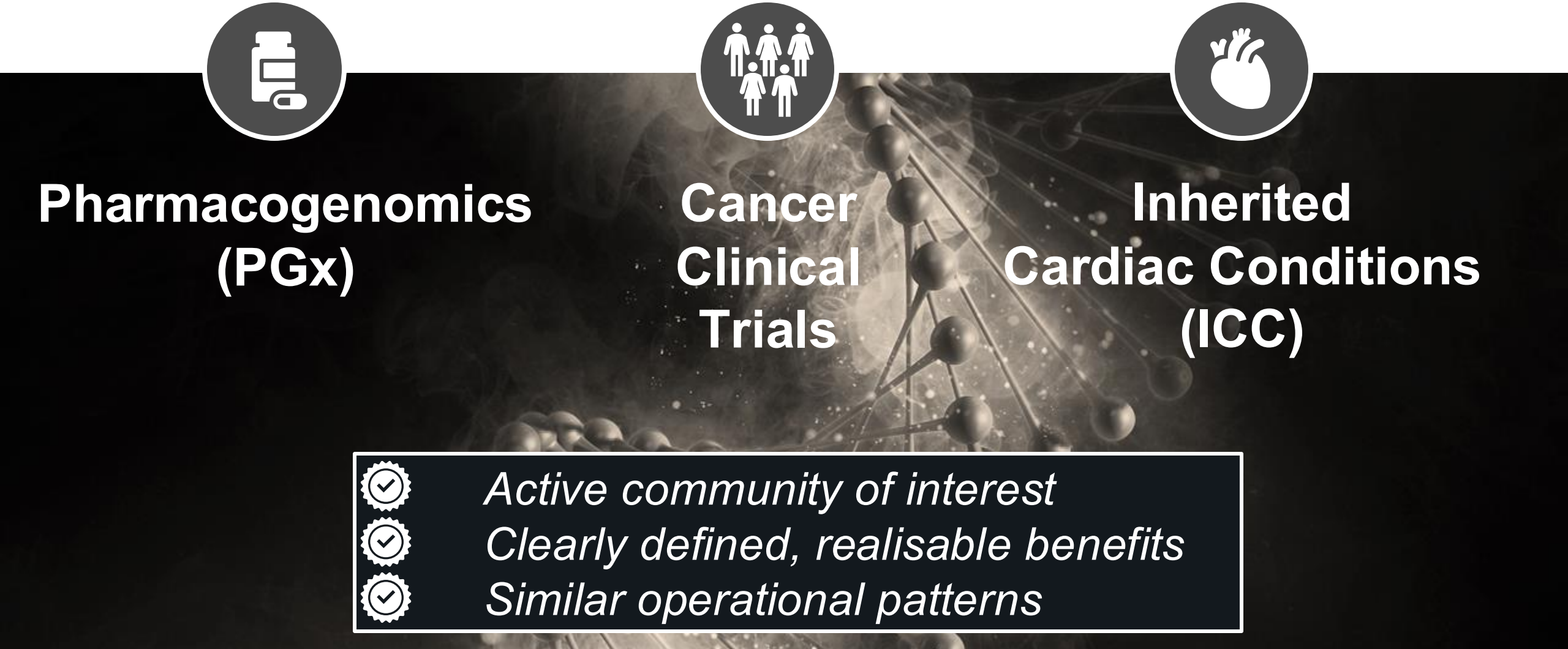
UGR is engaged in early conversations with SPR as a forerunner/pathfinder as many design principles and standards are common.

The Unified Genomic Record – The Vision

To provide a single point of administration and access to genomic data for each patient.



The Unified Genomic Record – Three Use Cases



Pharmacogenomics (PGx)

Cancer Clinical Trials

Inherited Cardiac Conditions (ICC)

- *Active community of interest*
- *Clearly defined, realisable benefits*
- *Similar operational patterns*

What is Pharmacogenomics?

Genomic data which supports the optimisation of medicine, reducing the risk of adverse events and improving patient outcomes

How does the UGR help PGx?

Enables PGx to scale at pace

Provide a dependable PGx profile per patient

1x∞

Reduce test duplication – test once, use many

Provides flexible architecture

What are Cancer Clinical Trials?

Research studies which test new ways to prevent, find, or treat cancer. They help discover better treatments and improve care using the latest scientific advances. Clinical trials often set genomic eligibility criteria for required candidates.

How does the UGR help Cancer Clinical Trials?

Identify eligible patients via one secure GMS wide search

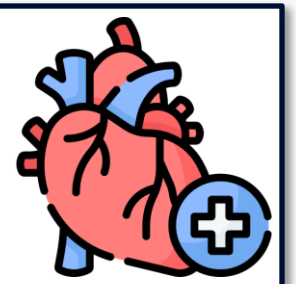
Potential to support research discovery tools

Joining with other datasets to support broader identification

Consent integration

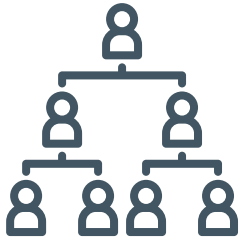
What are Inherited Cardiovascular Conditions?

Inherited cardiovascular conditions are heart or blood vessel disorders passed down through families. They can affect how the heart functions and may increase the risk of serious events like heart attacks or sudden cardiac arrest.



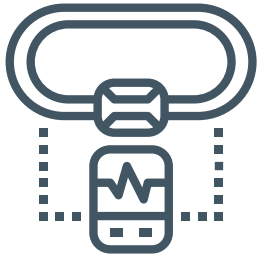
How does the UGR help with ICCs

Removing database siloes to improve research potential



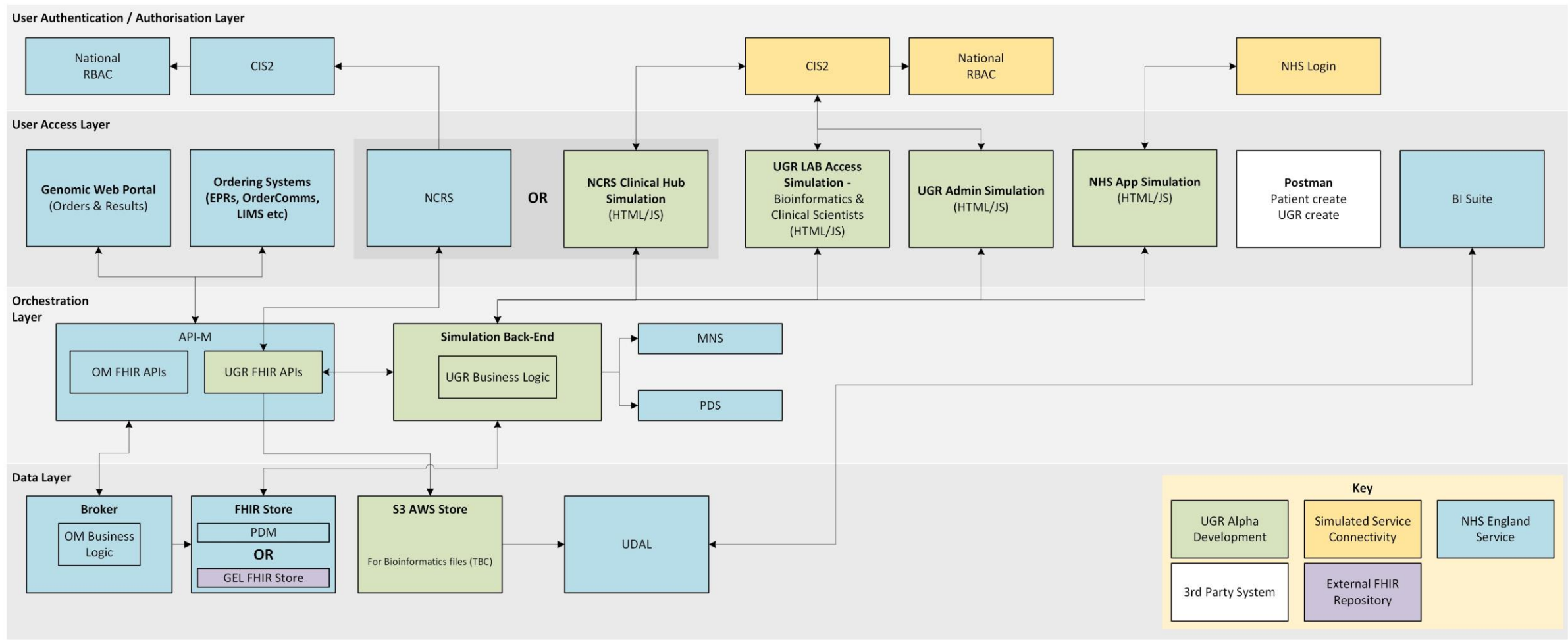
Identify family members potentially at risk

Reduce admin burden of notifying family members.



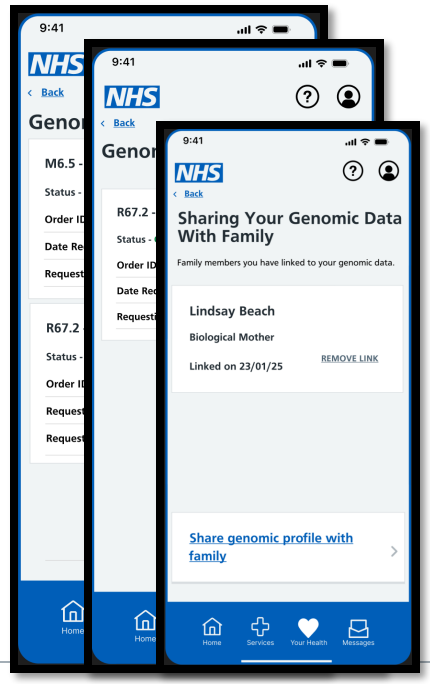
Routine care data will be mobilised for research

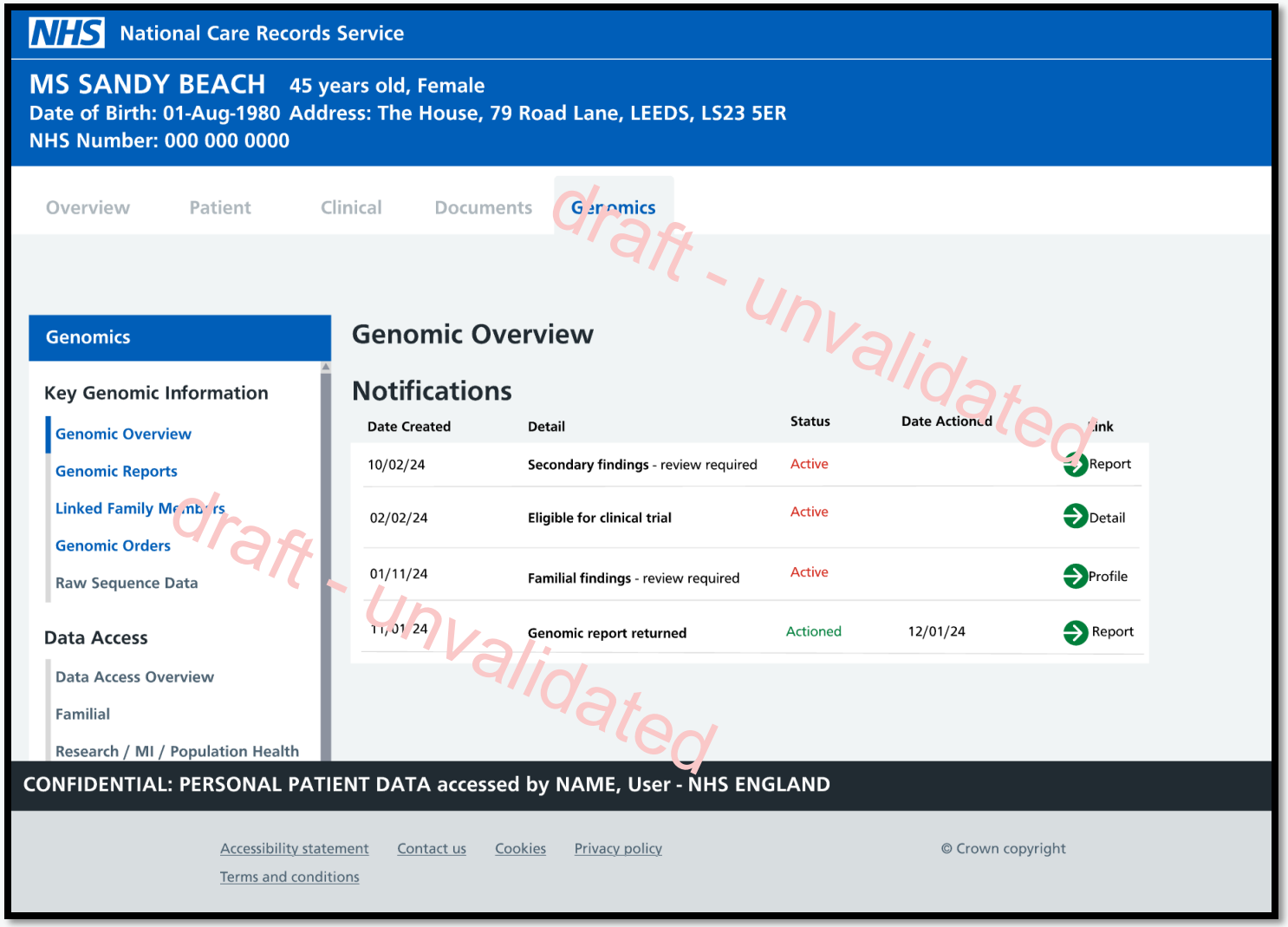
UGR Alpha Architecture



UGR – Alpha Wireframes

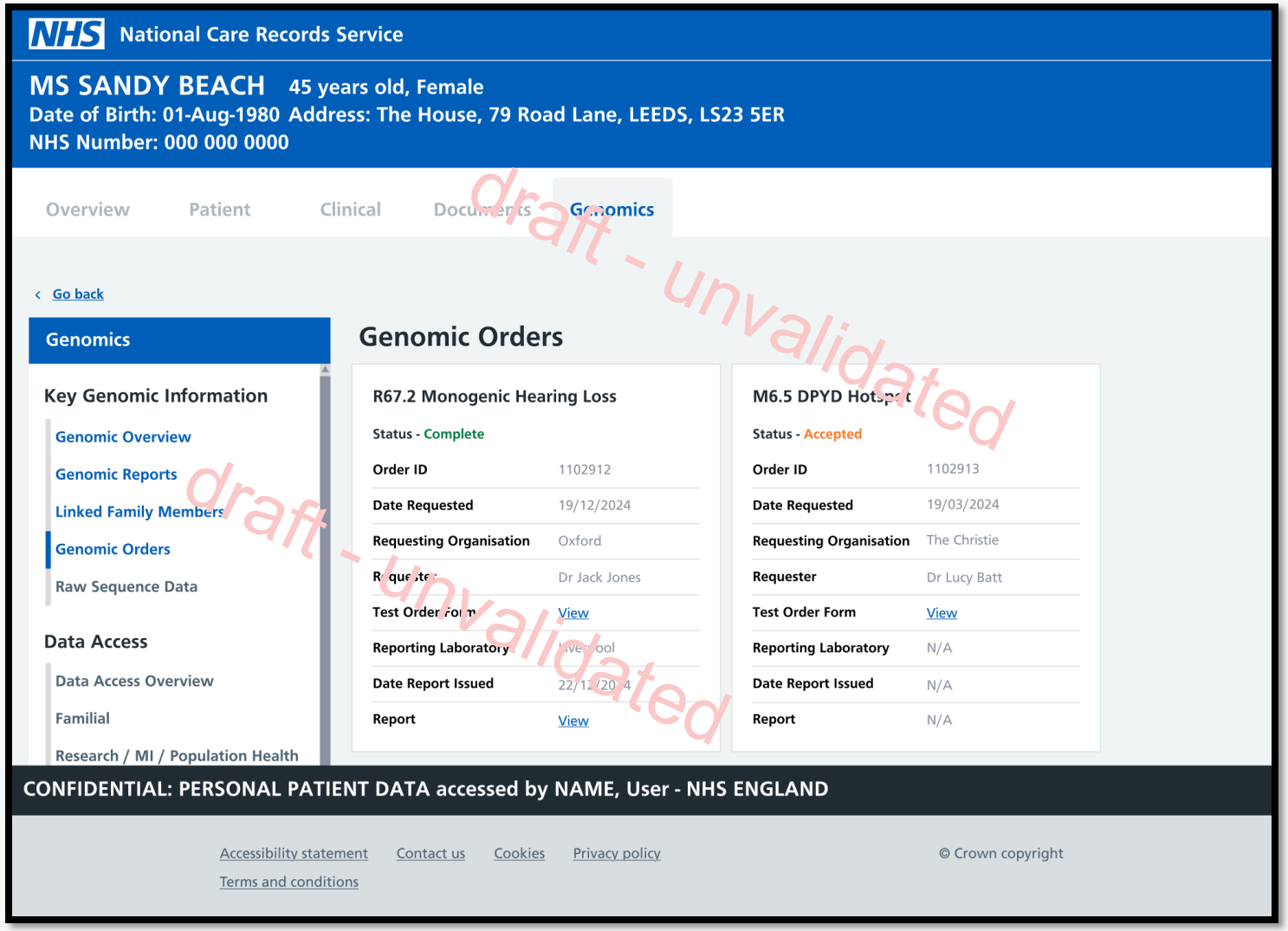
- As part of the Alpha Proof-of-Concept wireframes are being developed to help tell the ‘art of the possible’ story
- This includes showing how UGR could serve data to:
  - The National Care Record Service (NCRS)
  - Research Platforms
  - NHS app





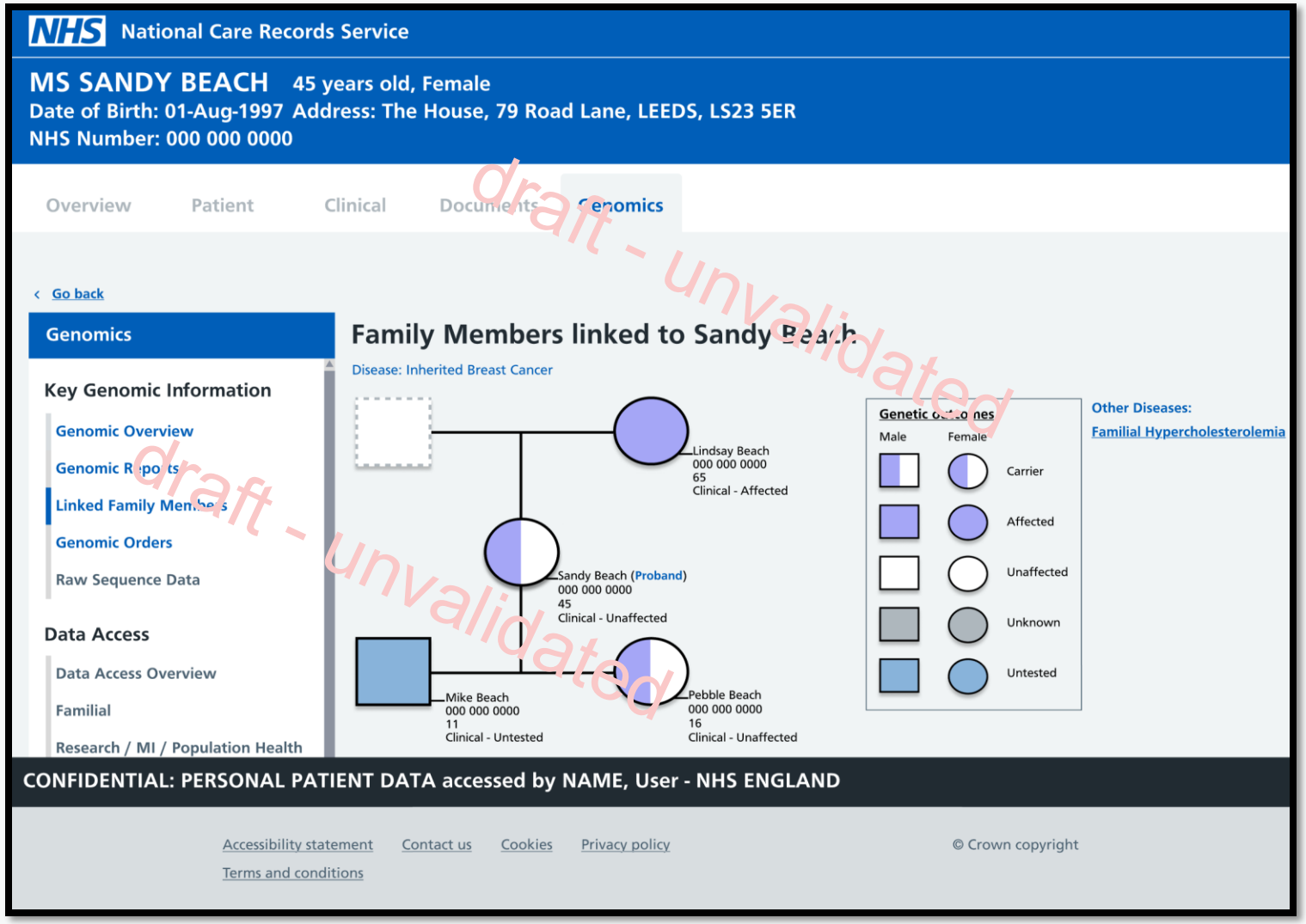
Consultant view - Notifications

- Secondary findings from genomic testing
- Clinical trial matching
- Findings from familial testing
- Test request status updates



Consultant view - Genomic Test/Report Details

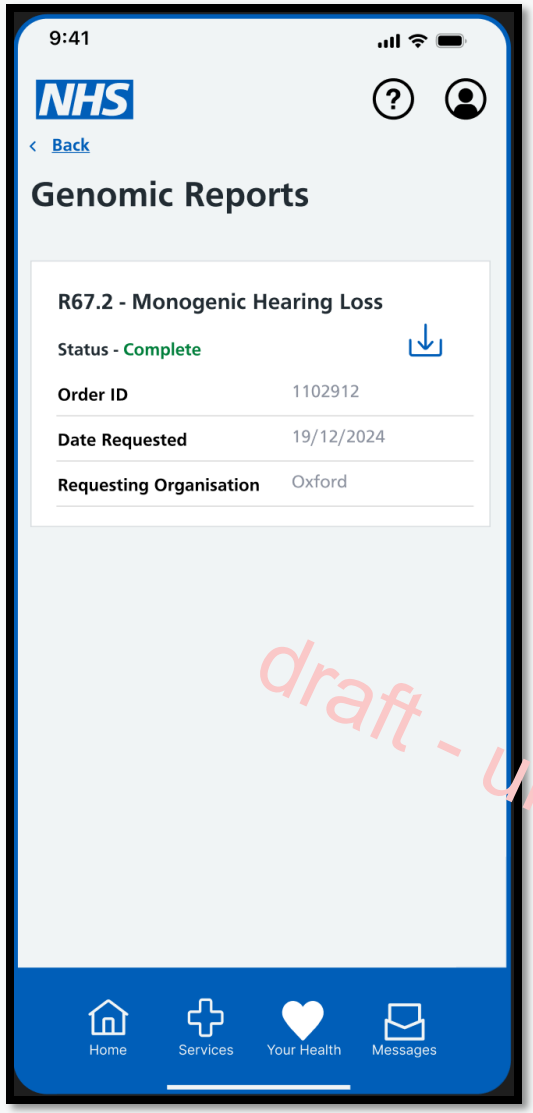
- Test order information
- Report information

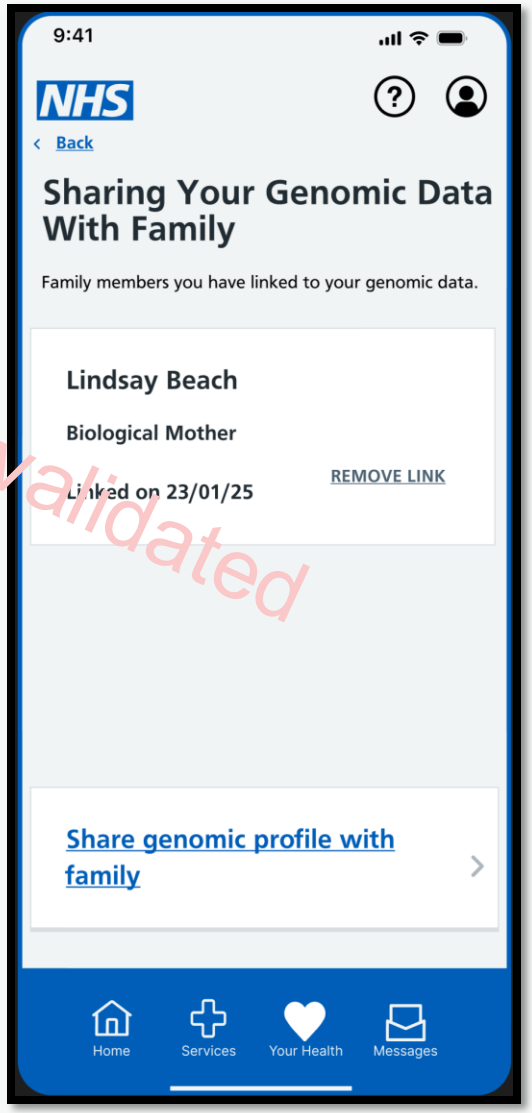


Consultant view -

Linked family members

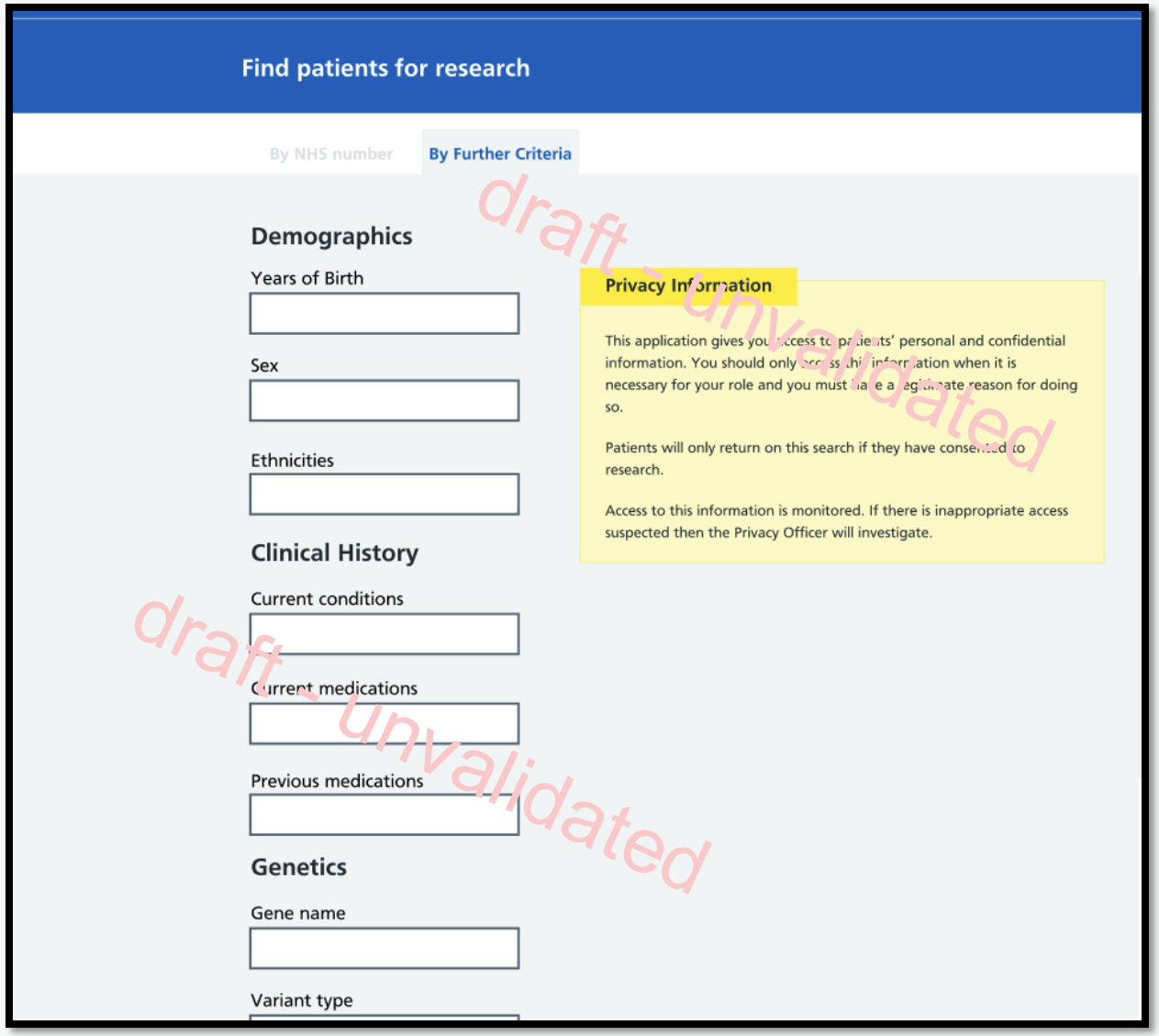
- Approved Family member links
- High level clinical information
- High level genomic information





Patient view -
NHS App

- Approved family member links
- Genomic ordering summary
- Genomic report access

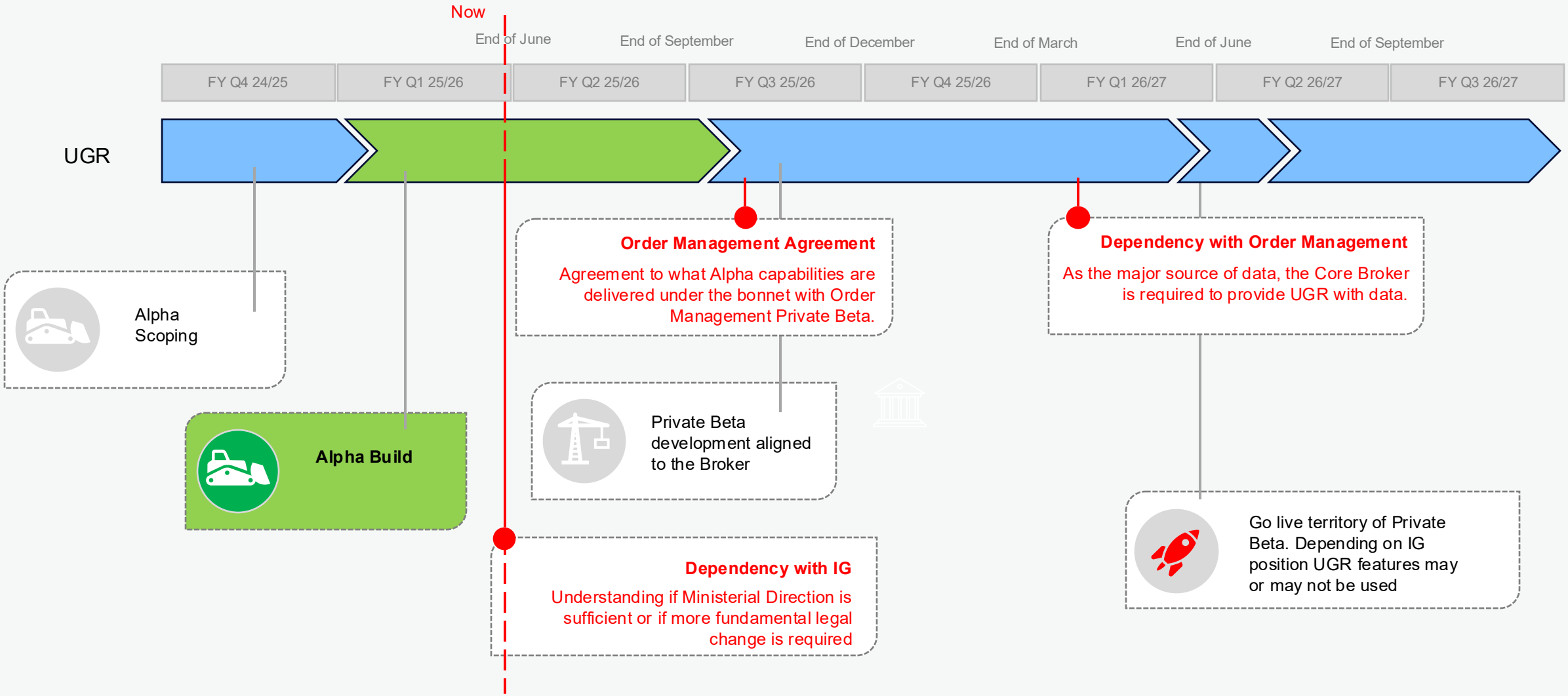


Researcher view -

Searching for eligible patients

- Search by demographic data
- Search by clinical history
- Search by genetic data
- Combine multiple criteria

UGR – Plan on a Page – The Wider Context



Call to action

How do suppliers want to be involved?

By autumn 2025 the project will be completing its Alpha phase. As UGR will have the potential to support many future use cases over the lifetime of a patient, and familial linkage, how would you like to be involved in design concepts?

How will UGR impact/ benefit you?

UGR will take time to deliver its full potential. It requires a volume of data from digital order management, and it also needs the current unstructured binary reports to move to structured reports. In the coming years, how will a fully capable UGR affect you?

Structured reports

The pre-discovery work has highlighted numerous early projects looking at the topic across NHSE and certain known standards like ISO20428. What insight does the room have on work to deliver genomic structured reports and standards?

How can you get involved?

Genomics Standards

Omar Khan
Ravi Natarajan

Standards

- To promote healthcare interoperability within NHS standards, play a key role.
- Standards drive:
  - Uniformity
  - Consistency
  - Reliability
  - and Conformity
- Are Patient Centric
- Allow the healthcare ecosystem and market to grow and bring innovation
- Improve Patient Safety and Empowerment
- Help accelerate accreditation
- Generate Cost improvements

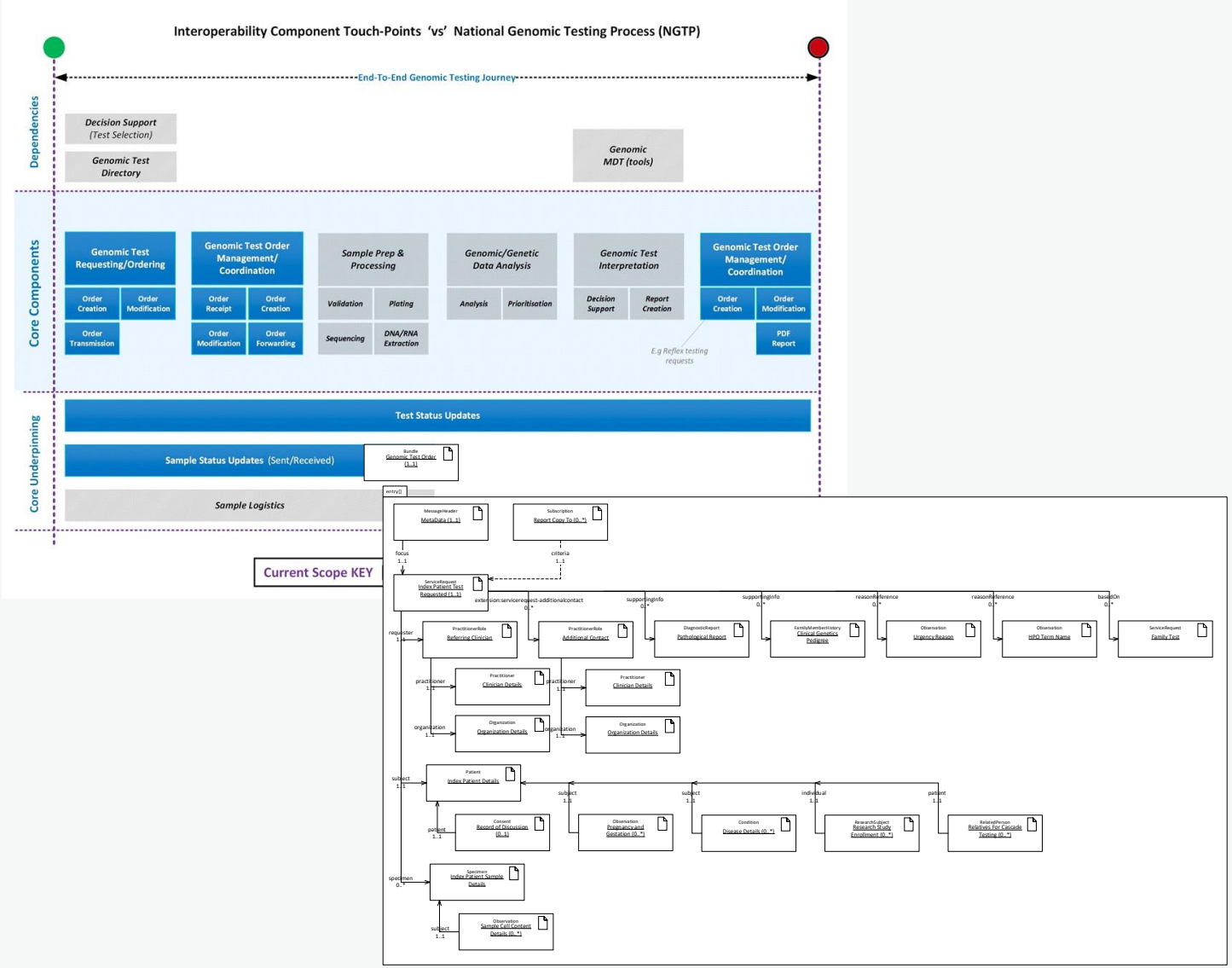
Initial investigation

Inputs:

- Use cases/user scenarios – National Genomic Testing Process
- Dataset and terminology needs – MDS
- Requirements

Areas of investigation:

- Test order data
- Order/Workflow Management
- Structured Reporting
- Genomic Data File Management
- Family linkage
- Unified Genomic Records
- Test Directories



Discovery

HL7 FHIR

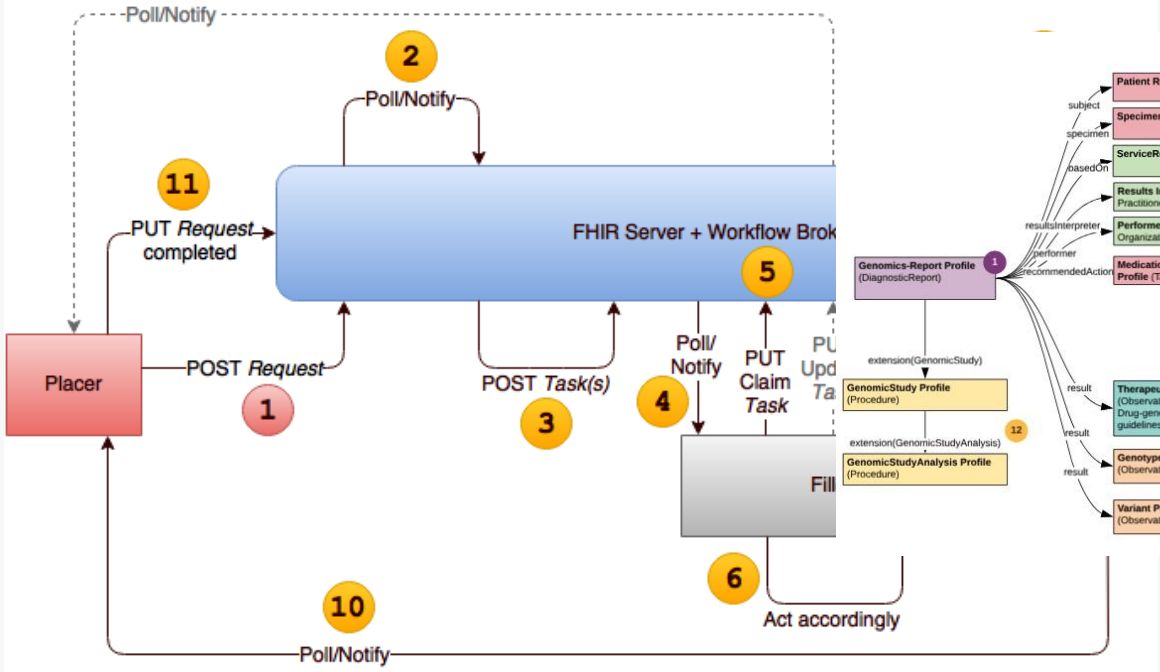
HL7 Europe

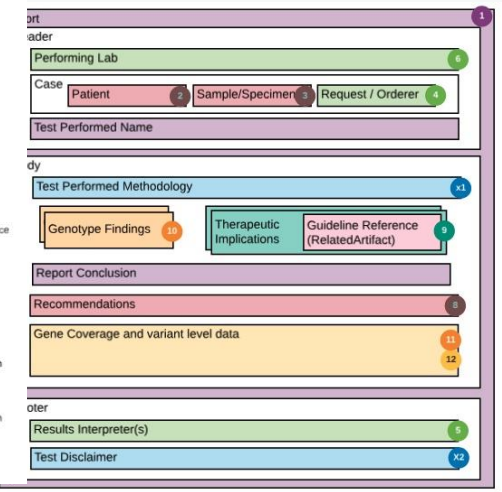
CEN ISO

openEHR

HL7 UK

IHE





NHS Data Model and Dictionary

PRSB Professional Record Standards Body

ACGS Association for Clinical Genomic Science

CDS HOOKS™

NHS Digital

OHDSI Observational Health Data Sciences and Informatics

Global Alliance for Genomics & Health

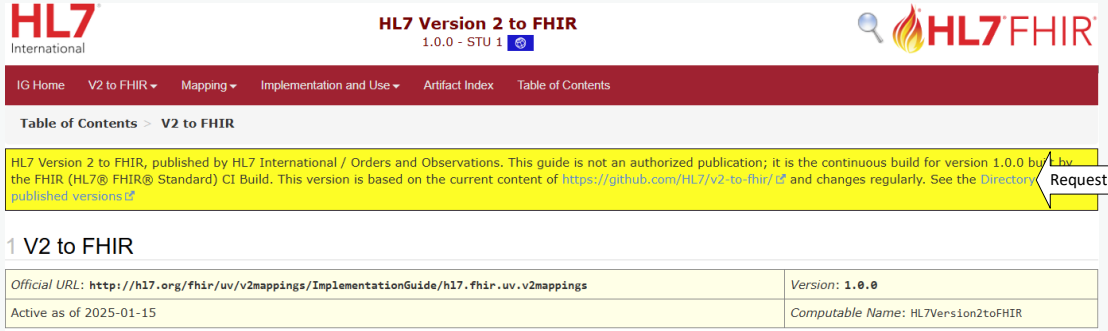
Discovery

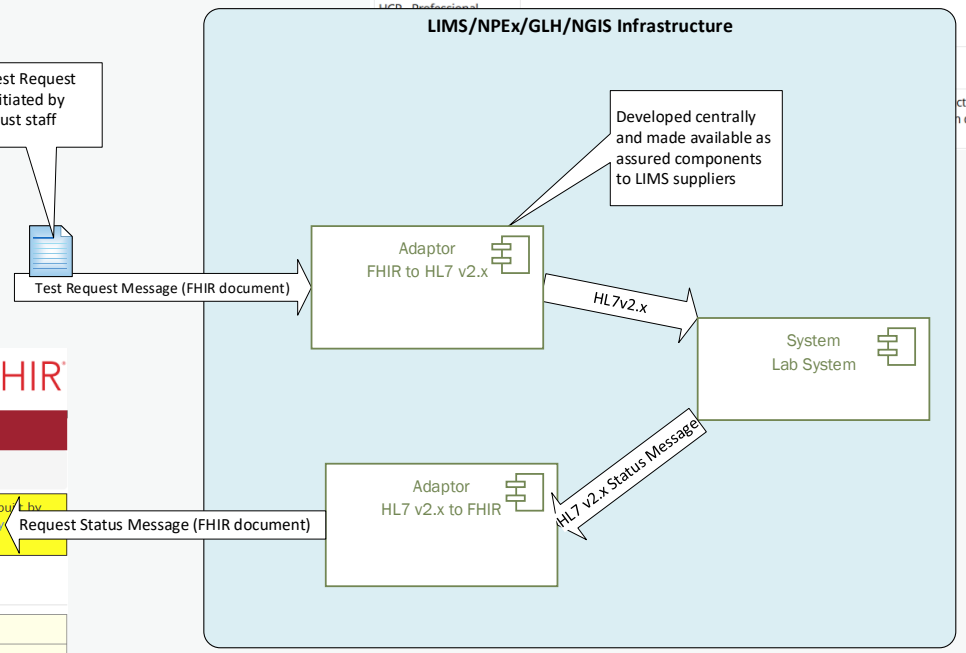
- **FHIR Workflow:** Basis for broker architecture
  - Multiple Tasks created based on NGTP/test type
- **IHE PaLM**: Basis for supported interactions
  - Source for HL7v2 Map (OML_O21 -> Genomics Test Order, ORU_R01 -> Genomics Test Result)
- **HL7 UK Core**: Profiles used within Genomics
  - Not profiled specifically for Genomics but promoted unmet requirements to Core
  - Additional guidance on populating elements provided in text
- **NHS England Pathology IG**: For inclusion of pathology reports as supporting-info
  - Aligned with to ensure consistent representation of report in NHS England (Messaging used vs. REST)
- **NHS Data Dictionary:** Used to align with NHS England concepts and ValueSets
- **PRSB**: Guidance for how to alert users to PGx contraindications
- **NHS England Digital Services:** Reuse of NHS England core services
  - PDS master patient index (NHS patients only), ODS master organisation index
- **HL7 EU Lab**: Aligned with for cross border sharing
  - Issues with LOINC codes (SNOMED used exclusively in UK), Patient birth date (fetal testing)
- **OpenEHR PGx Result/Variant Detail Archetypes**: Map to FHIR
  - Ambiguity in coding schemes for particular elements
- **HL7 CG WG Genomics Reporting IG**: Basis for structured report model
  - Similar issues regarding LOINC
- **ACGS**: Report structure
  - Working on alignment to FHIR models
  - Part of larger discovery around genomic report data models (including ISO standards)
- **GA4GH DRS/htsget**: Genomic Data discovery and access
  - Working on supporting both GA4GH and FHIR Genomics-operations
  - Also working on alignment to PhenoPackets and Pedigree guidance
- **OHDSI OMOP**: Map to FHIR
  - Utilising CDMH IG

FHIR vs. HL7v2

- HL7v2 more widespread in order comms and intra Trust messaging
- Messaging vs. RESTful interactions
- Point-to-point vs. central request orchestration and distributed fulfilment
- Data elements outside traditional OML_O21 message
- HL7v2 to FHIR maps provided to support uplift

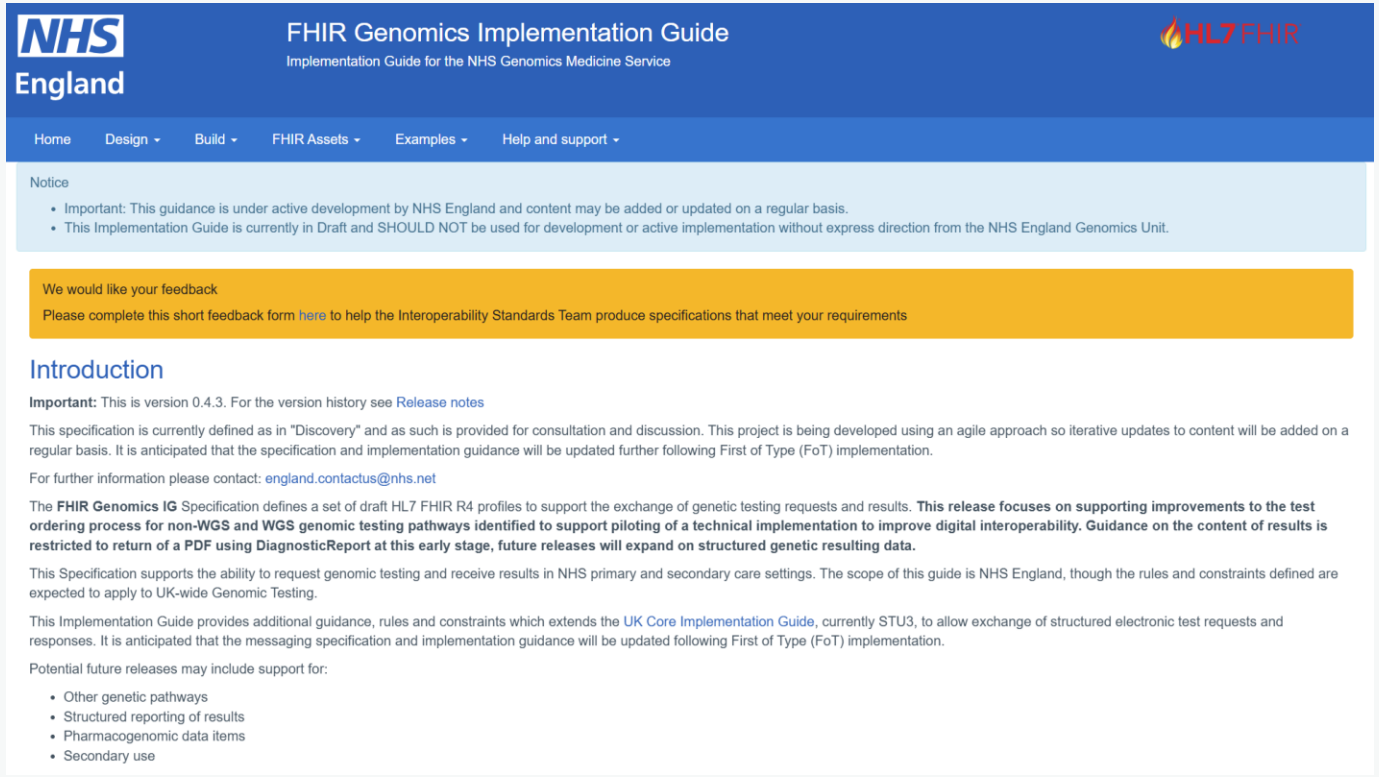
Source Data item	Target FHIR Element	HL7v2.5.1 Mapping	Description
HCP - Genomic test order role	Determined through where the PractitionerRole is referenced from e.g. for the requester: ServiceRequest.requestor, Additional contact: ServiceRequest.extension:additionalContact, Sample collection: Specimen.collection.collector etc.	multiple possible segments e.g. ORC-12 for requester	HCP's function within the genomic test ordering process.
HCP - Full name	PractitionerRole.practitioner.display (full delimited name can be retrieved via identifier e.g. from SDS)	ORC-12	HCP's full name.
HCP - Job Title	PractitionerRole.code (display can be used to capture human readable job role code)	CTD-1	HCP's job title.
HCP - Current Specialty	PractitionerRole.specialty	Additional CTD-1 segments	HCP's current specialty.
HCP - Phone	PractitionerRole.telecom:system=phone	ORC-14	HCP's phone number.
HCP - Email address	PractitionerRole.telecom:system=email	CTD-5.4	HCP's email address.
HCP - Organization name.	PractitionerRole.organization.display (full delimited name can be retrieved via identifier e.g. from ODS)	ORC-21	HCP's organization name.
HCP - Organization address.	PractitionerRole.organization (retrieved via ODS)	ORC-22	HCP's organization address.
HCP - Organization ODS code.	PractitionerRole.organization.identifier	ORC-21.10	HCP's organization ODS code.
HCP - Department name	PractitionerRole.healthcareService.identifier, could alternatively use PractitionerRole.healthcareService.display to record human readable version or PractitionerRole.specialty if mapped to clinical specialty **TBC**	Additional CTD-1 segments	HCP's department name.
HCP - Professional registration number	PractitionerRole.practitioner.identifier	ORC-12.1	HCP's professional registration number such as their GMC number.
		ORC-12.9	HCP's professional registration number type such GMC.
		CTD-6	HCP report preferred delivery method.
	ctpoint-comment h depending on	Additional CTD segment (CTD-5.4)	Central email address provided by the HCP.





Work still to do

- Align to existing data sets/data standards to reduce burden on Lab staff (and allow reuse of data) e.g. for NDRS
- Undertake Terminology review for strategic direction on use of Genomic specific nomenclatures/ontologies
- Expand Implementation guide to cover Structured Reporting and UGR/DGTS work
    - Including purpose-based access control and test metadata
- Proactive subscriptions/notifications
- Request/reporting frameworks/patterns for other domains e.g. Pathology/Radiology/NIR (TSAS)



Questions?

Wrap up

Thank You

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