



England

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...

The logo for techUK, featuring the word "tech" in a dark blue, lowercase sans-serif font, followed by "UK" in a light blue, uppercase sans-serif font. The logo is centered within a white rectangular box that has a subtle drop shadow.

techUK

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
NHS England



Ravi Natarajan
Technical Architect
NHS England



Beryl Cairns-Hockey
Project Manager
NHS England



Luisa Van Der End
Project Manager
NHS England



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



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**

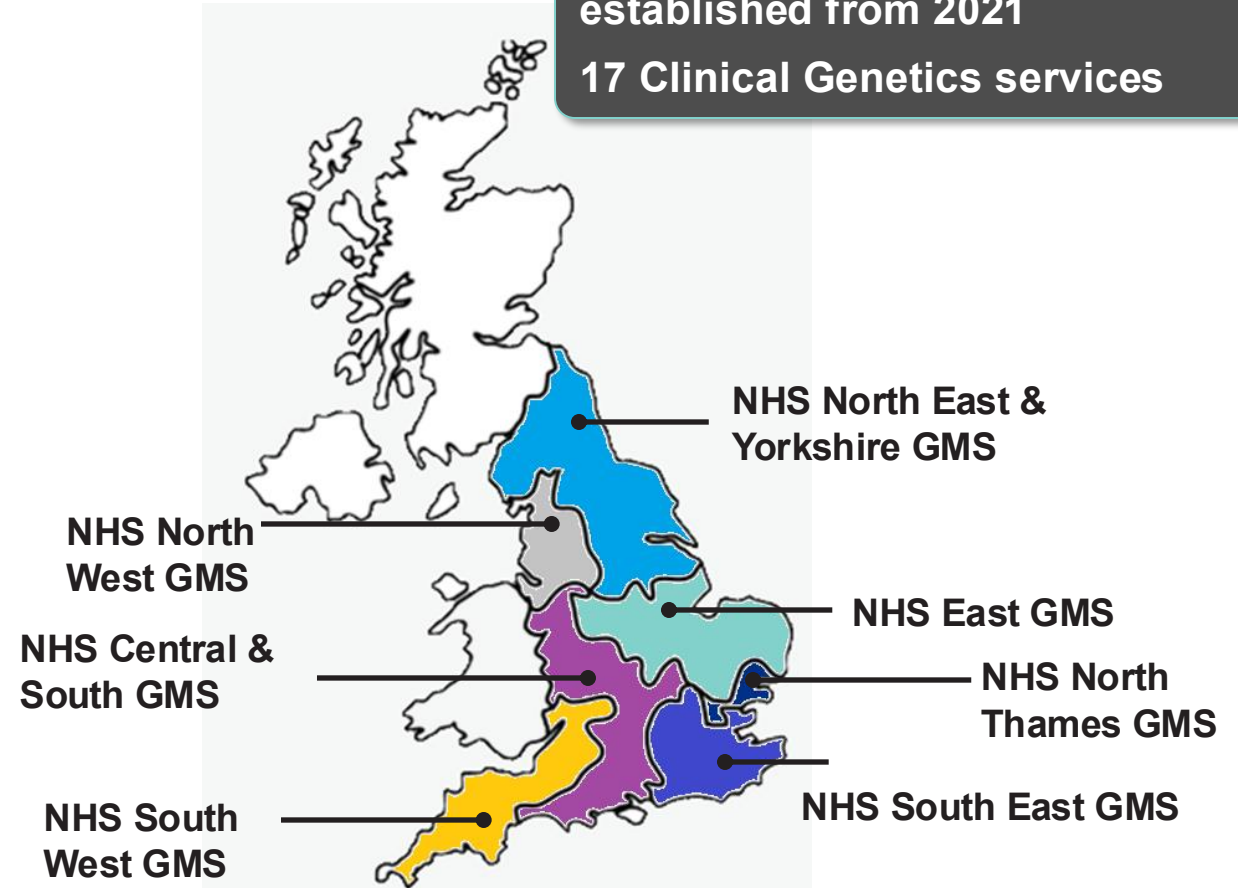
... to maximise diagnosis, increase access to precision medicine and efficiency.

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



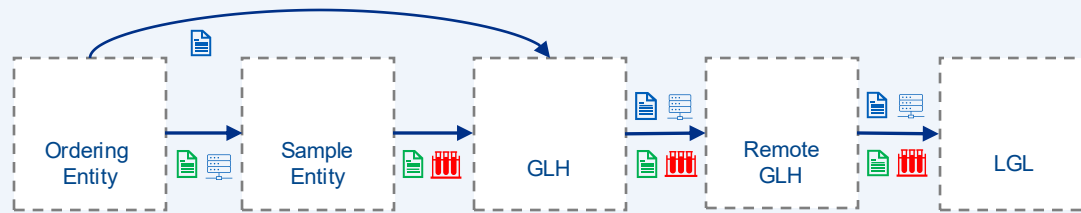
Digital Order Management

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

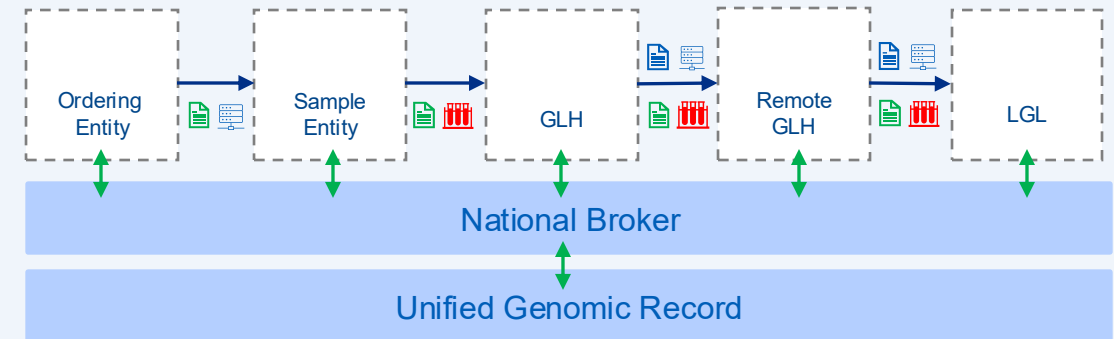
Challenges

- 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

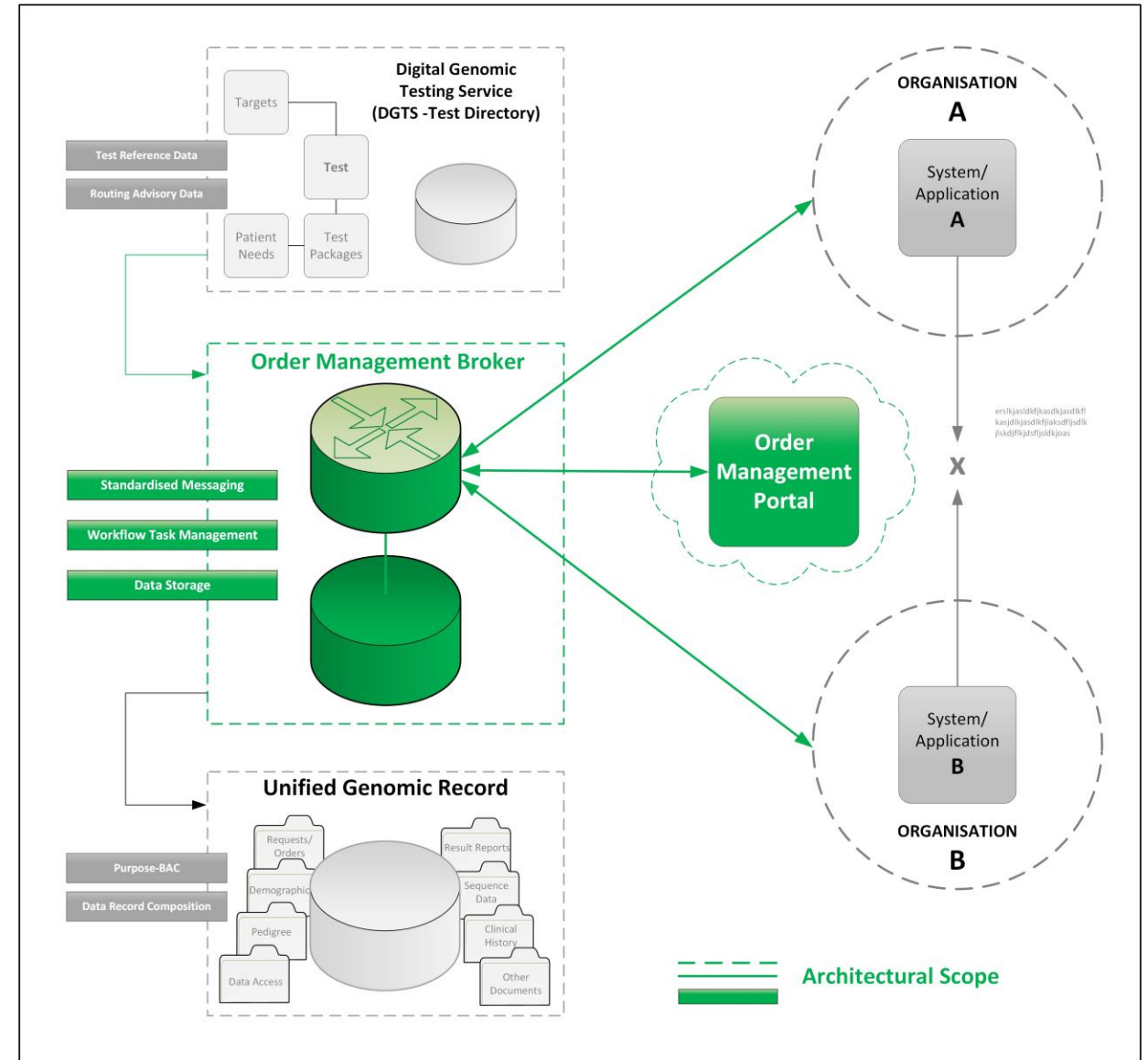
Architecture – Order Management Scope

OM broker infrastructure

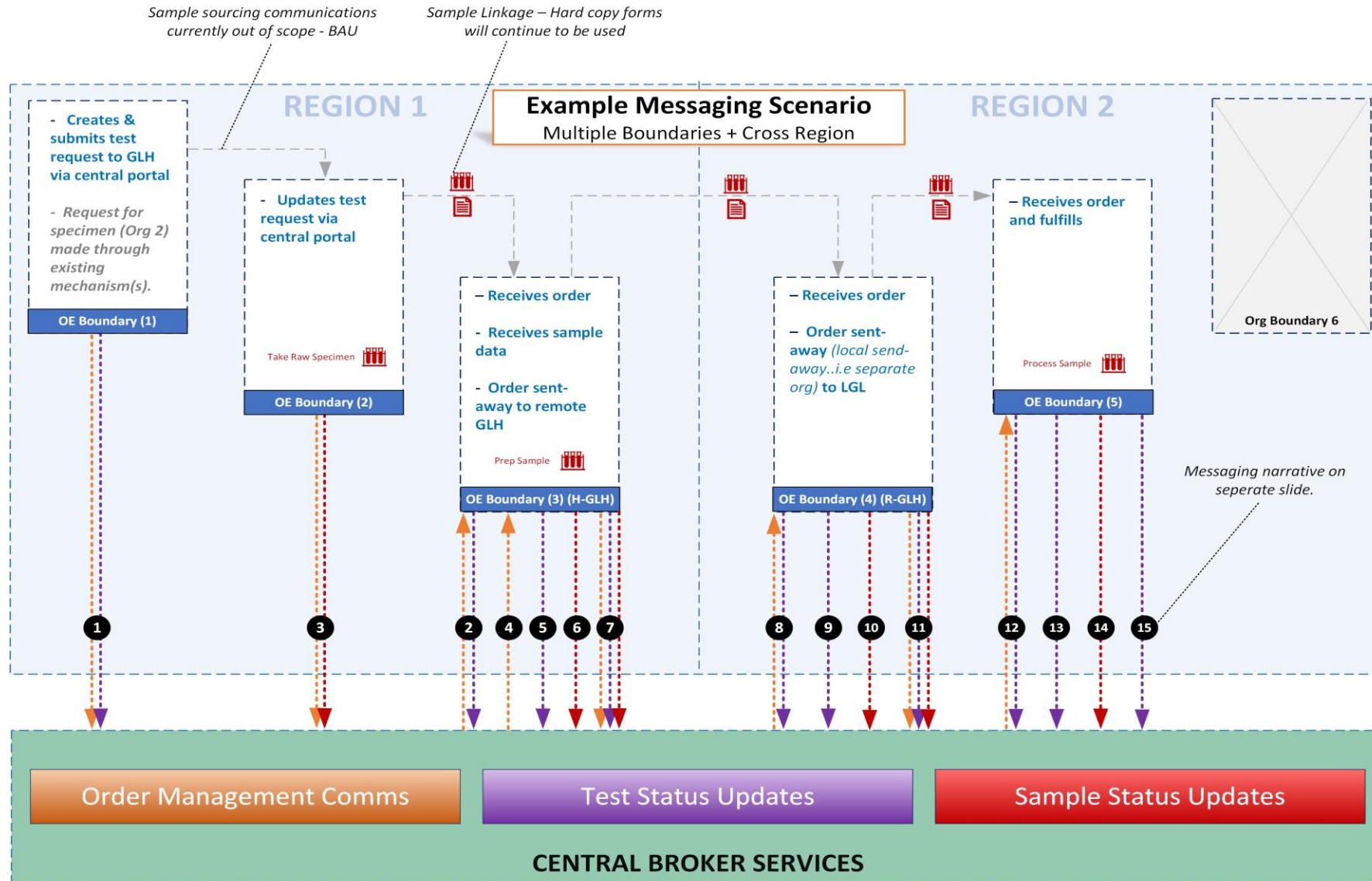
- Facilitate standardised messaging between organisational boundaries
- Provide a workflow task management capability
- Provide the data storage needed to support the movement of data between organisations throughout the workflow
- Consume the DGTS reference data
- Foundation component for UGR

OM portal

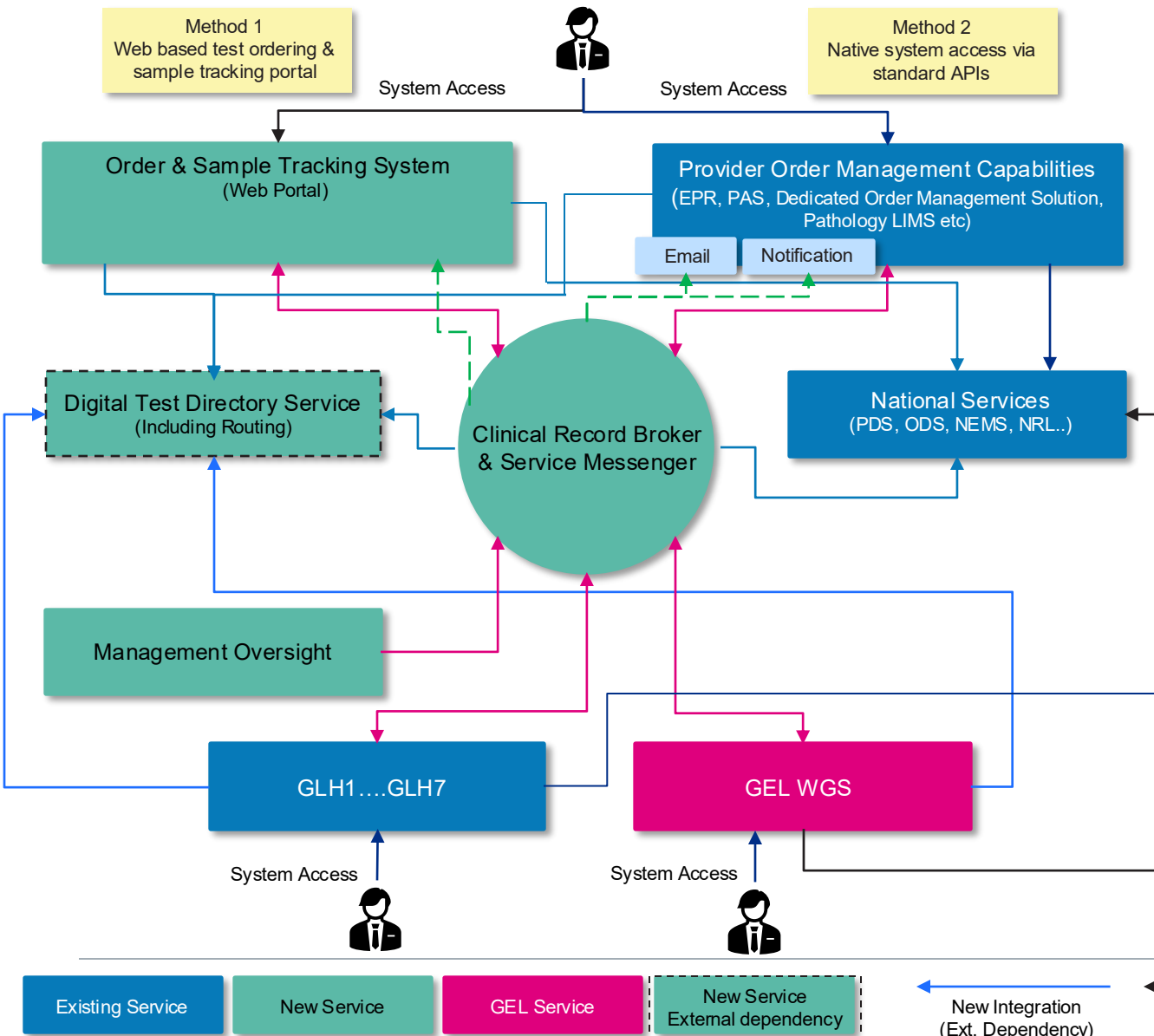
- Provides a user interface for ordering and managing genomic test requests, leveraging all functional capabilities of the broker



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



The ROYAL MARSDEN
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.



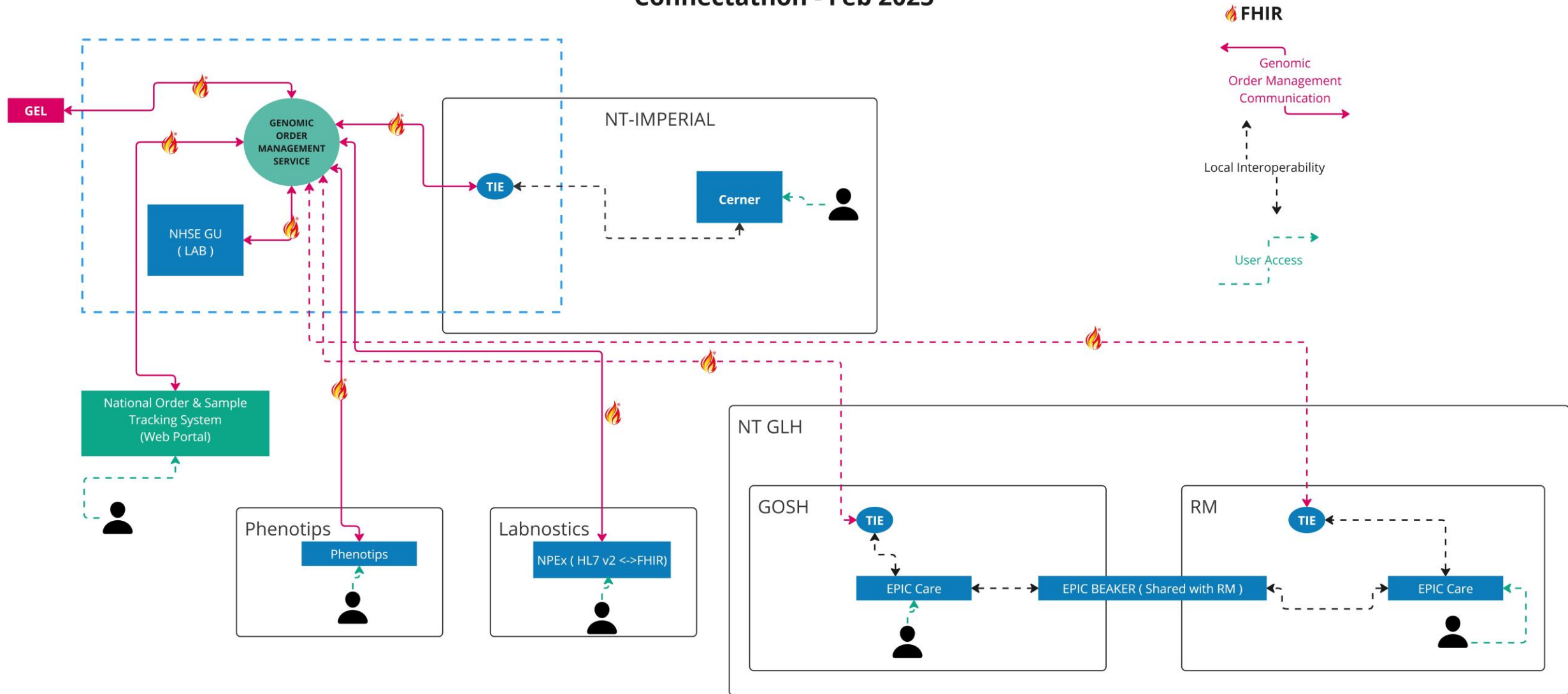
The Alpha integration will generate a blueprint for other trusts to adopt for their infrastructure.



Connectathon

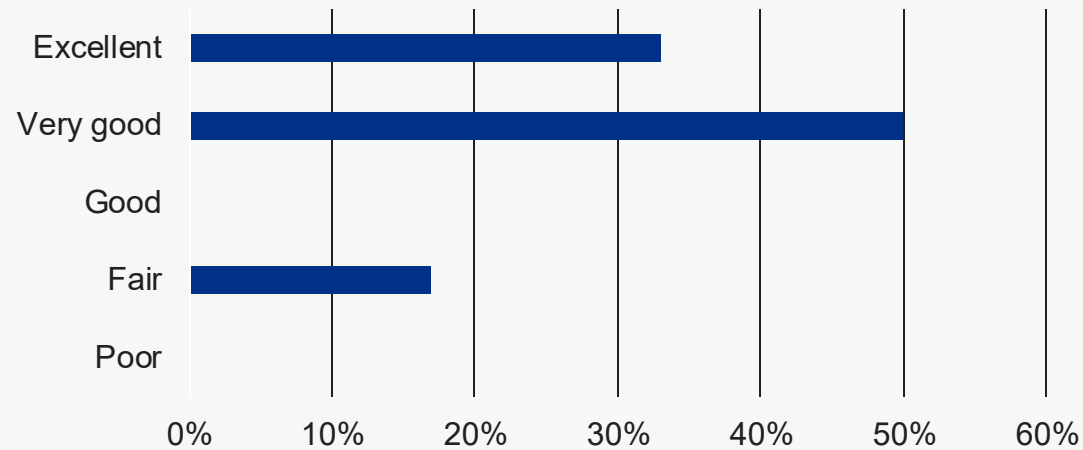
- 2 sessions held at TechUK in London – opportunity for suppliers to test workflows connected to the genomic order management service
 - 15th January
 - 21st February
- Approximately 60 people attended across the 2 dates, including Imperial, Royal Marsden, GOSH, GEL, broker developer Aire Logic, web 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

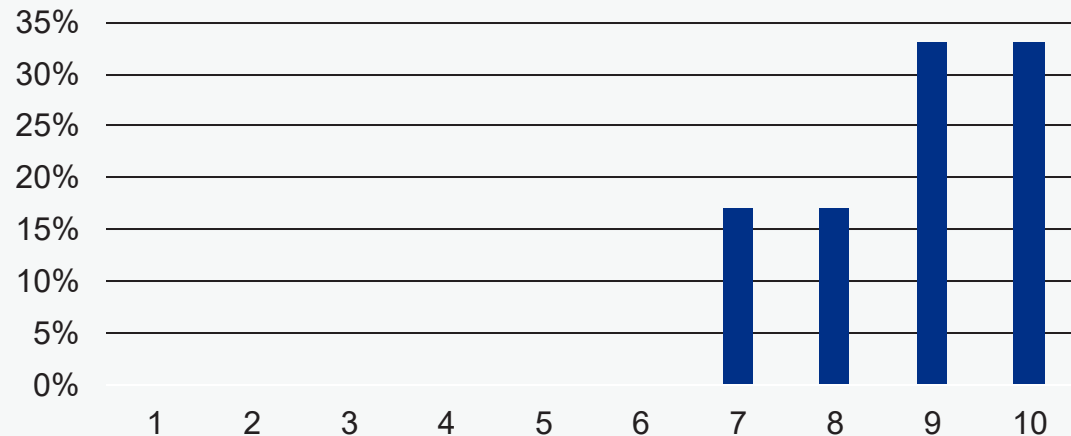


Survey responses

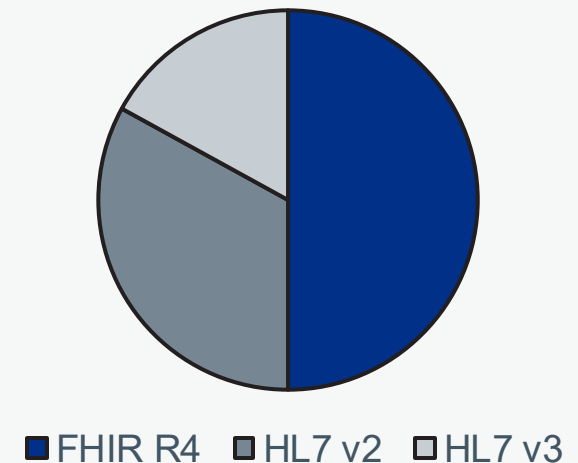
How would you rate the overall organisation and execution of the Connectathon



Out of 10, how would you rate the quality of the provided data on the FHIR server/POSTman collections?



If you use Interoperability standards, please can you advise which standard you are using?



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

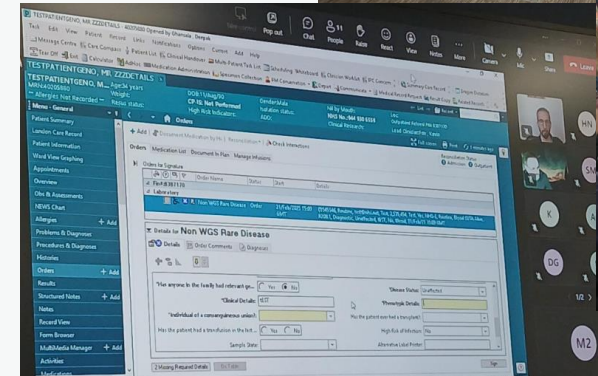
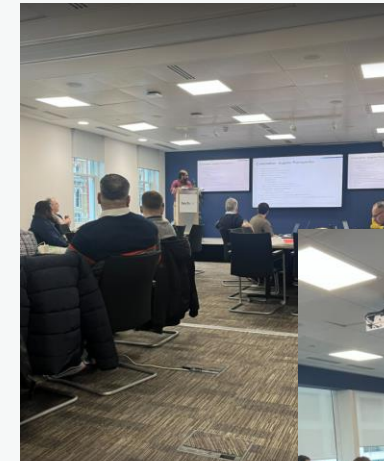
- Resolvable canonical URLs for FHIR assets

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

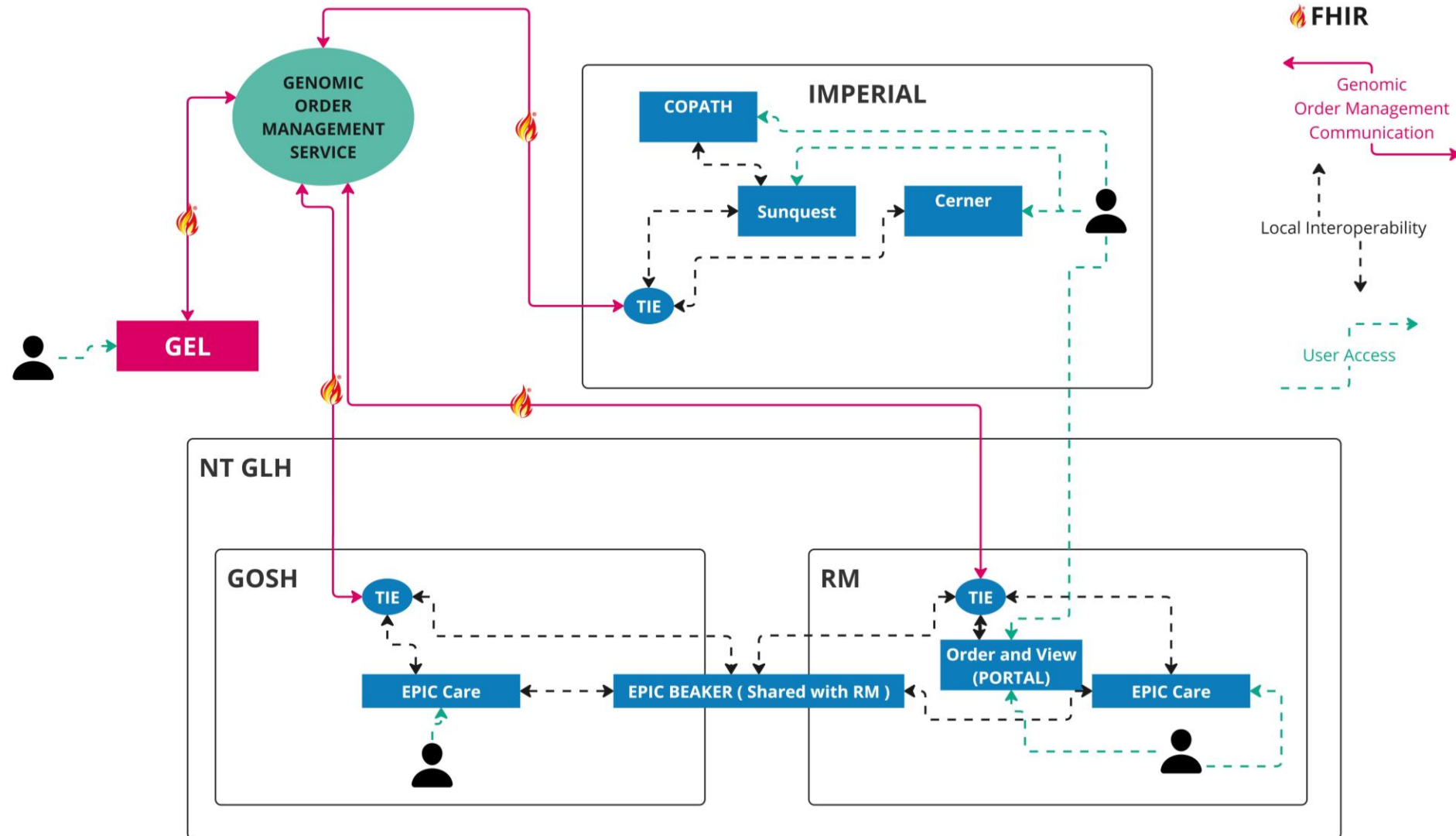
Broker capability

- 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

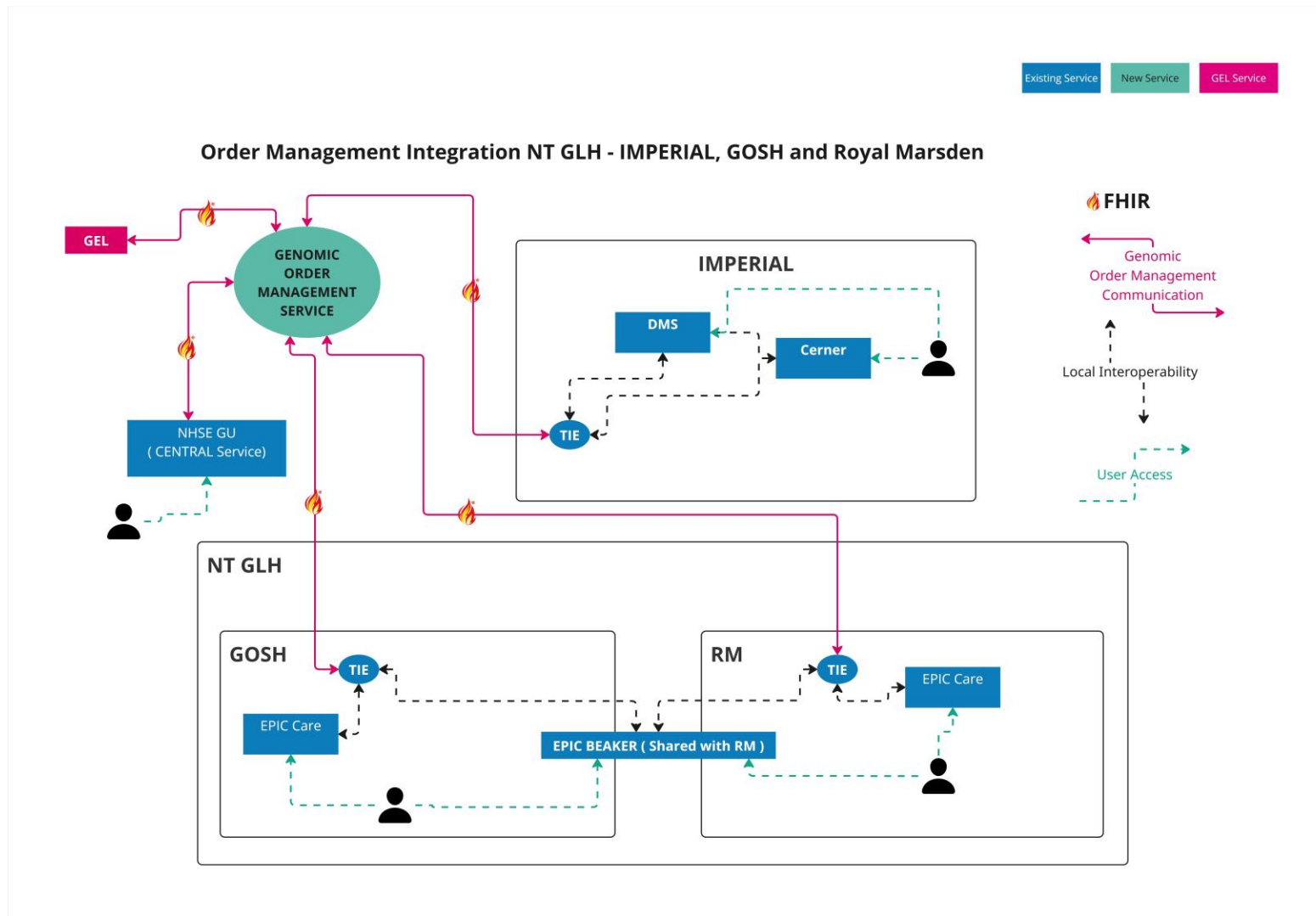
FHIR guidance

- 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

ZZZTEST, GENO-TWELVE - 40207983 Opened by Madhala, Sreelatha

EN English (United Kingdom)

Task Edit View Patient Record Links Notifications Options Current Add Help

Message Centre Care Compass Patient List Clinical Handover Multi-Patient Task List Scheduling Whiteboard Clinician Worklist IPC Concern Ambulatory Organiser Bed Board Discharge Dashboard Pharmacy Care Organiser Handover Dashboard

Summary Care Record BNF

Launch Apps TRIPS Dragon Dictation DAX Copilot

Tear Off Exit Communicate Discern Reporting Portal Calculator AdHoc PM Conversation Result Copy Related Records Access Management Office Medication Manager Collections Inquiry Depart Application Launch Medical Record Request

Abnor.: 0 Other.: 0 Remins.: 0

ZZZTEST, GENO-TWELVE

Age: 34 years DOB: 12/Sep/90 Gender: Female Nil by Mouth: NHS No.: 973 457 7530 Loc: Outpatient Referral FIN: 8390038

Weight: CP-IS: Not Performed Isolation status: ADD: Clinical Research: Lead Clinician: Fox, Kevin

Resus status: High Risk Indicators: ADD:

Menu - General

- Patient Summary
- London Care Record
- Patient Information
- Ward View Graphing
- Appointments
- Overview
- Obs & Assessments
- NEWS Chart
- Allergies + Add
- Problems & Diagnoses
- Procedures & Diagnoses
- Histories
- Orders + Add
- Results
- Structured Notes + Add
- Notes
- Record View
- Form Browser
- MultiMedia Manager + Add
- Activities
- Medications
- Drug Administration
- Drug Admin Summary
- Pregnancy View
- Delivery Record
- Newborn Summary
- PPwT (new)

Orders

Display: All Orders (All Statuses)

ZZZTEST, GENO-TWELVE - Add Order

Age: 34 years DOB: 12/Sep/90 Gender: Female Nil by Mouth: NHS No.: 973 457 7530 Loc: Outpatient Referral FIN: 8390038

Weight: CP-IS: Not Performed Isolation status: ADD: Clinical Research: Lead Clinician: Fox, Kevin

Resus status: High Risk Indicators: ADD:

Diagnoses & Problems

Diagnosis (Problem) being Addressed this Visit

+ Add - Convert Display: All

Annotated Display Code

Problems

+ Add - Convert No Chronic Problems

Display: All

Annotated Display Name of Problem

Search: Contains Type: Prescription

Folder: Specialty Folder Search within: All

- ***High Risk Indicators***
- ***Flags***
- ***Sepsis***
- A&E
- A&I
- Bariatrics
- Blood Transfusion
- Burns Unit
- Cardiology
- Cardiothoracic Surgery
- Chest And Allergy
- Dermatology
- Dynamic Function Tests (DFTs)
- Endocrinology
- Endoscopy
- Endoscopy Referrals for Vetting
- ENT
- Food and Drink Menus
- Gastro-enterology
- GUM
- Gynaecology
- Gynaecology
- Haematology
- Head And Neck
- Hepatology
- HIV

ZZZTEST, GENO-TWELVE - 40207983 Done

Details

Dx Table

Show More Orders... Orders For Signature

C0459 SMADHALA 18 June 2025 14:42 BST

MADHALA, Sreelatha (IMPERIAL COLLEGE HEALTHCARE NHS TRUST)

14:42 18/06/2025



GEORGIU, Demetra (IMP...)



Ritu Sharma



BALAN ...



Grece Issa



JONES, ...



MALIK, S...

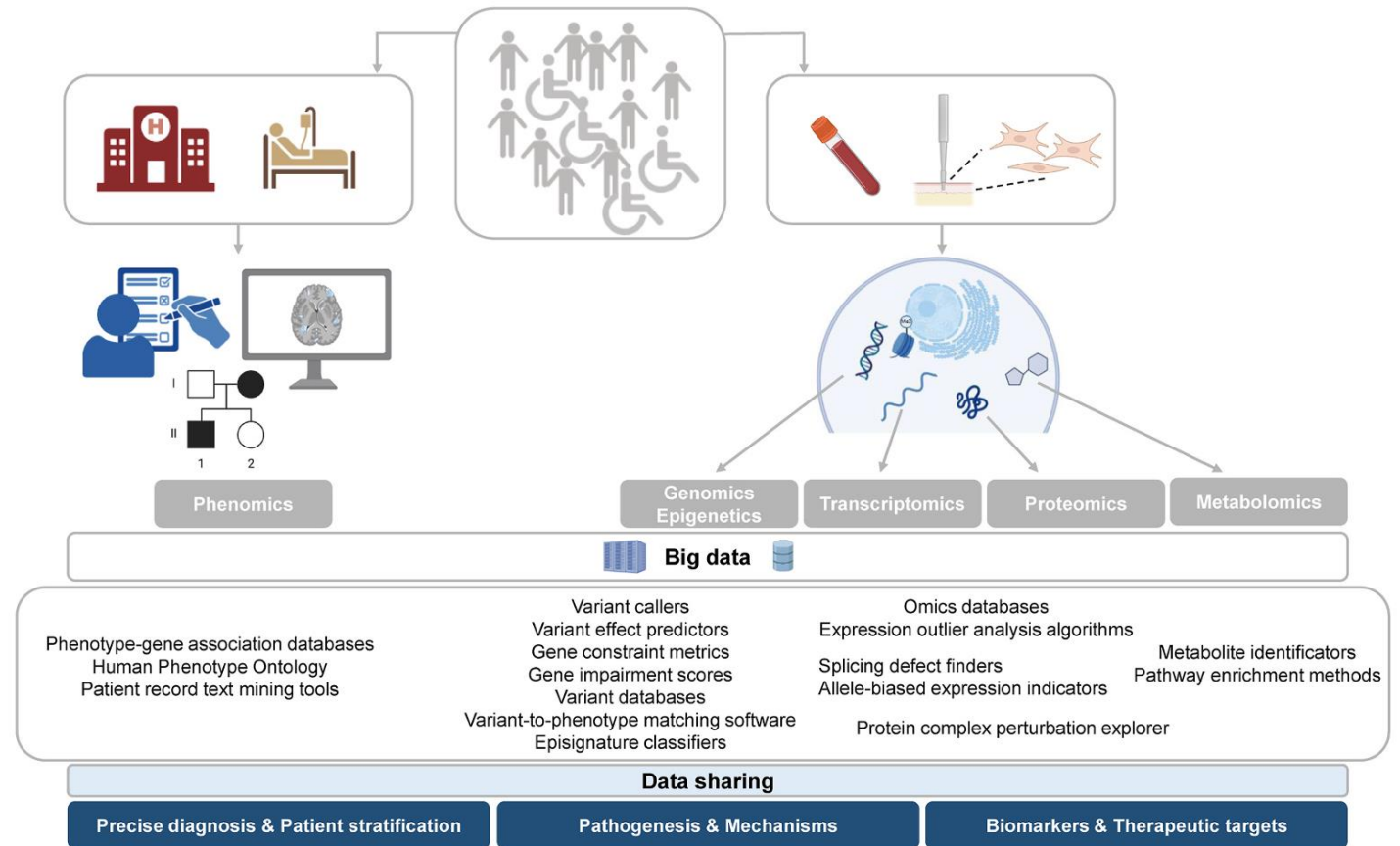


MADHA...

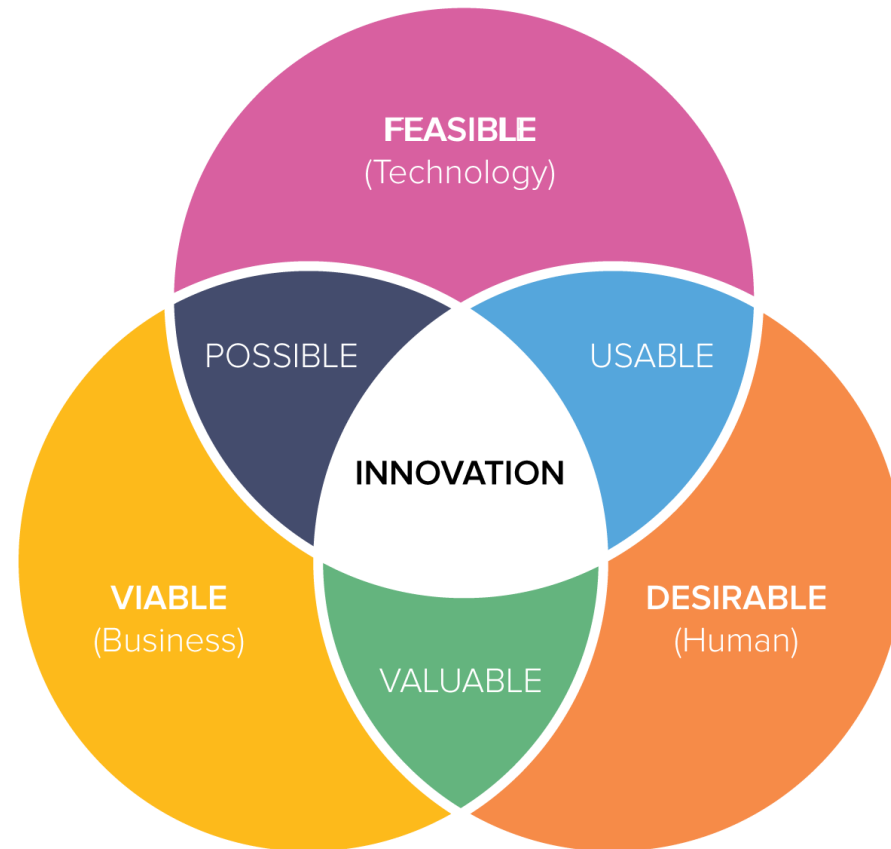


North Thames GLH (NTGLH)

- 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

The screenshot shows a Microsoft PowerPoint presentation titled "Alpha Web Portal - Demo" displayed in a web browser. The browser's address bar shows the URL: `nhs.sharepoint.com/sites/NHSEADGEL-OrderManagement/_layouts/15/doc.aspx?sourcedoc={419c871d-b1f2-4120-be5f-ce5e5166df8b}&action=edit`. The PowerPoint interface includes a ribbon with tabs: File, Home, Insert, Draw, Design, Transitions, Animations, Slide Show, Review, View, and Help. The "Home" tab is active, showing options for Undo, Paste, Delete, New Slide, and various font and paragraph formatting tools. The presentation consists of four slides, with the first slide selected and highlighted in the left-hand slide pane. The first slide, titled "Logging on to Web Portal", contains the following text: "Select Patient" and "Order test". The second slide, titled "Fill in Requesting Details", contains the text: "Fill in the Patient Demographic Details". The third slide, titled "Enter request Details", contains the text: "Add Patient Information". The fourth slide, titled "Request Summary Screen", contains the text: "Submit Test to Broker". The main content area of the presentation shows the first slide's content in a large, centered font.

1 Logging on to Web Portal
Select Patient
Order test

2 Fill in Requesting Details
Fill in the Patient Demographic Details

3 Enter request Details
Add Patient Information

4 Request Summary Screen
Submit Test to Broker

Logging on to Web Portal

Select Patient

Order test

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



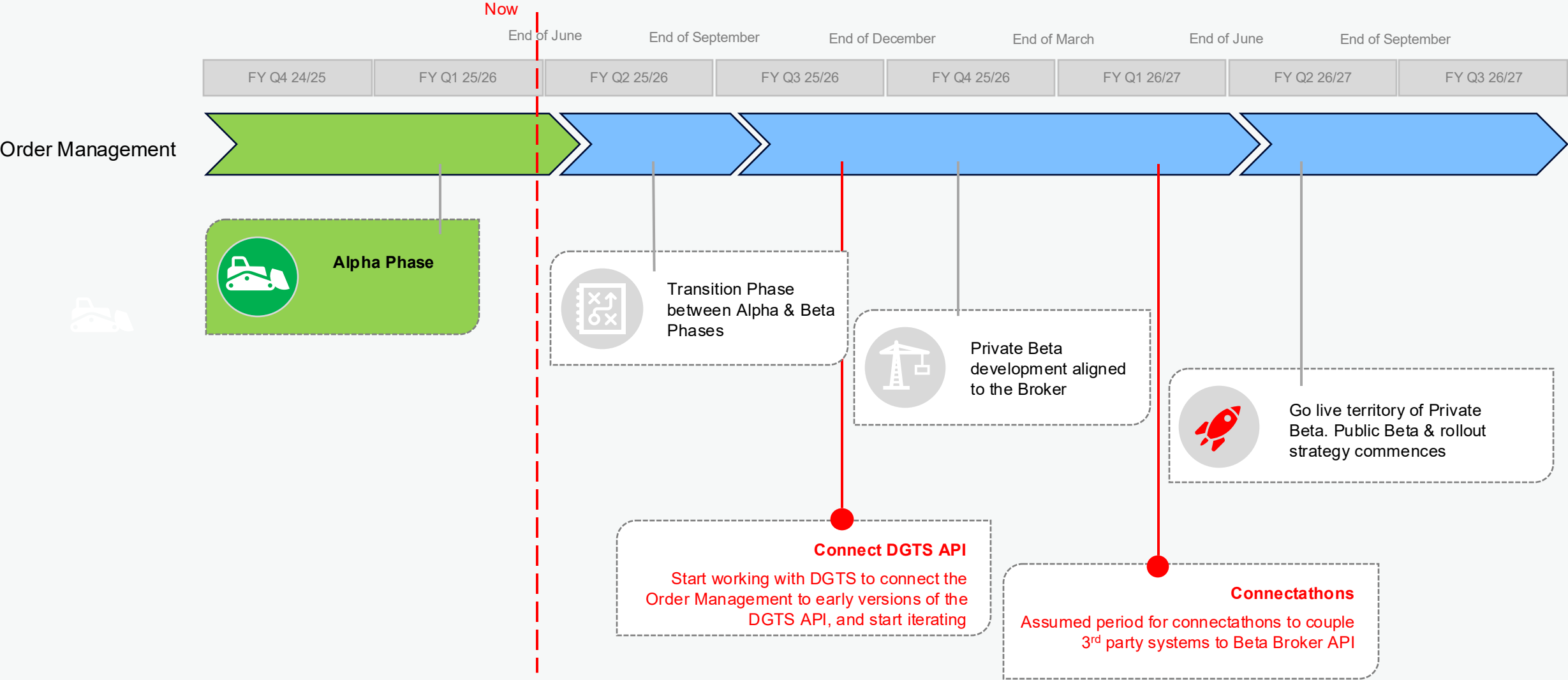
Order Management

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 connectathons 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:

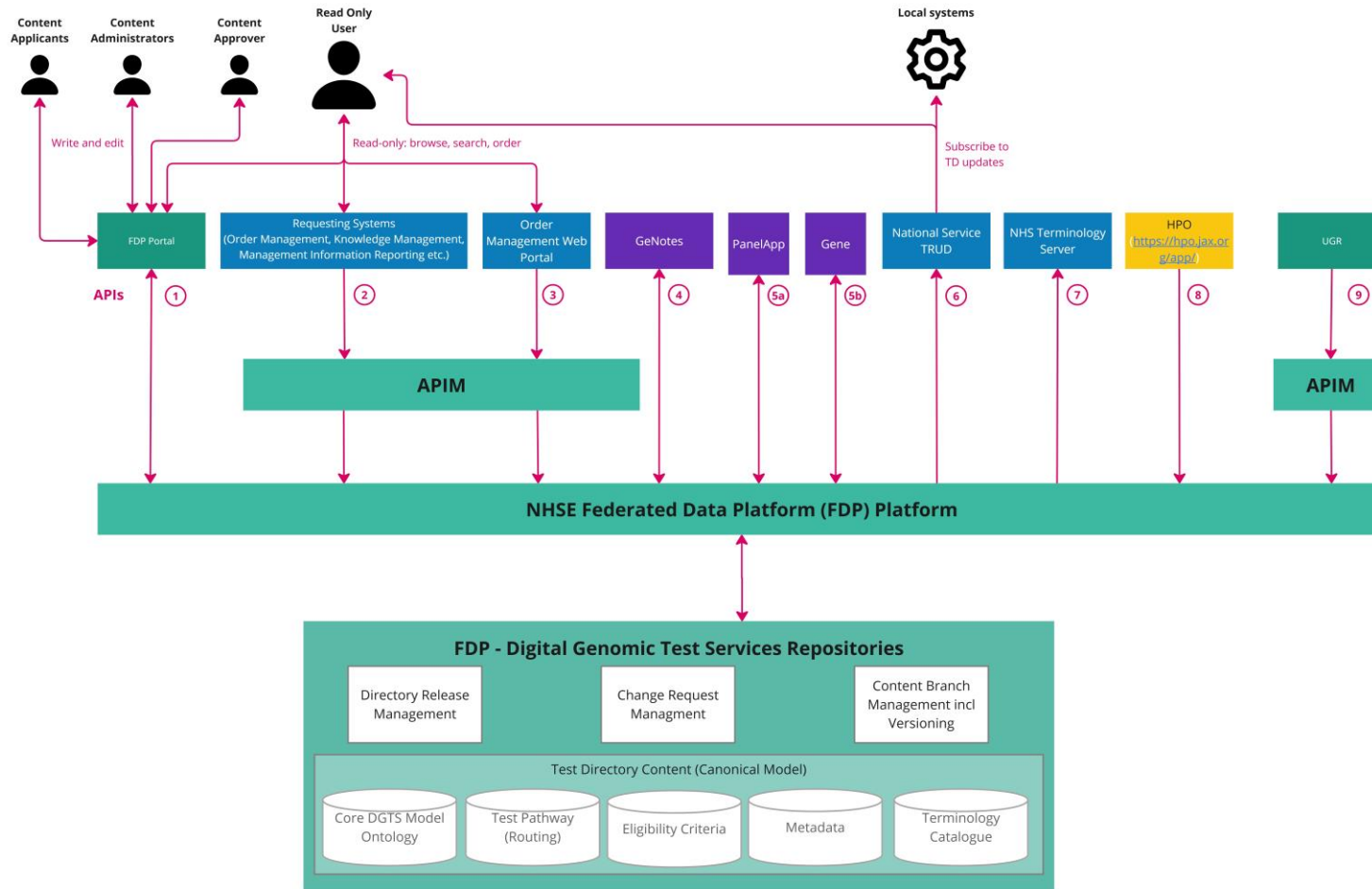
First class searching and browsing capabilities and frontends for viewing all directory data.

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

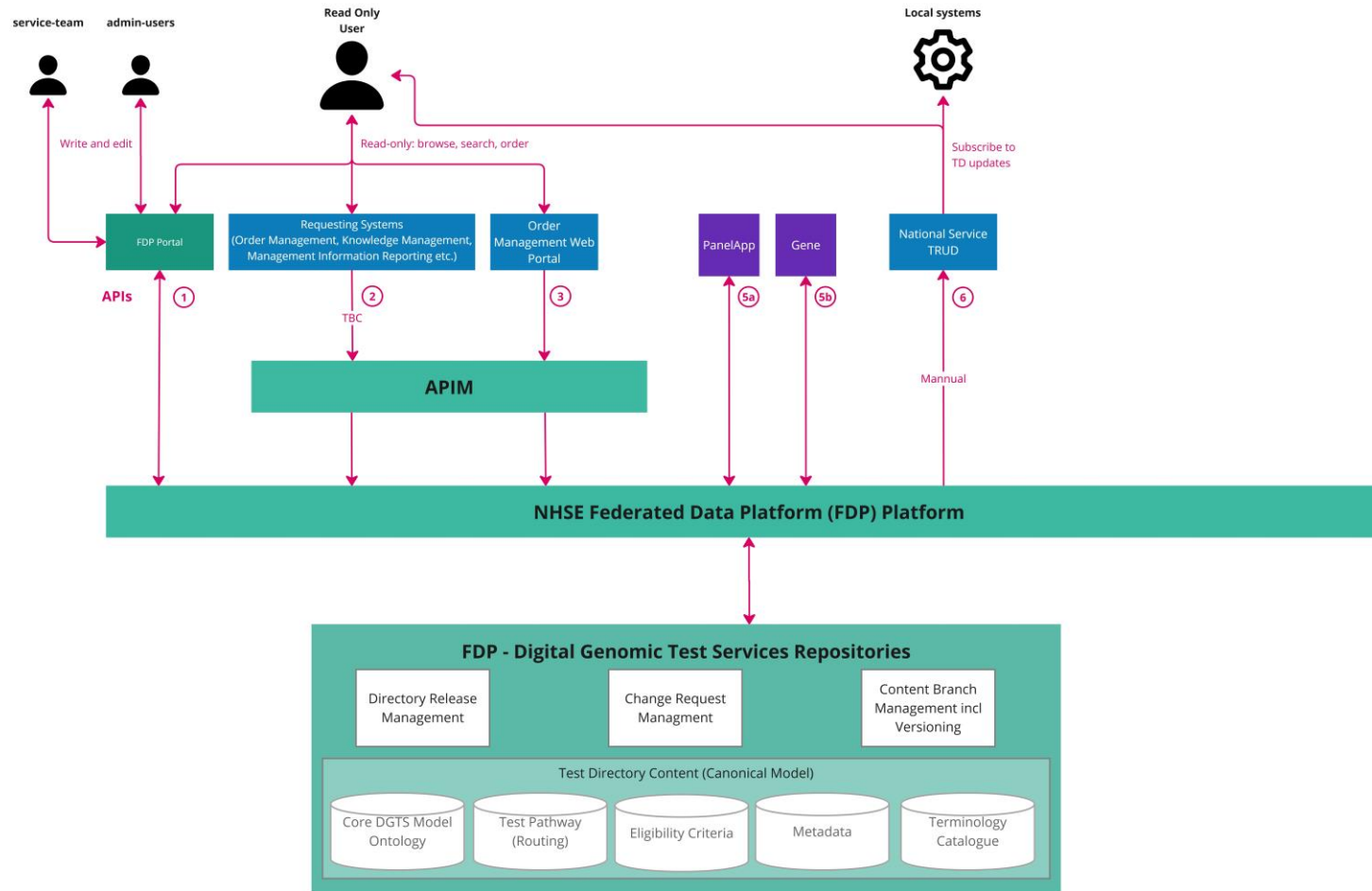
Release Process

- 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.

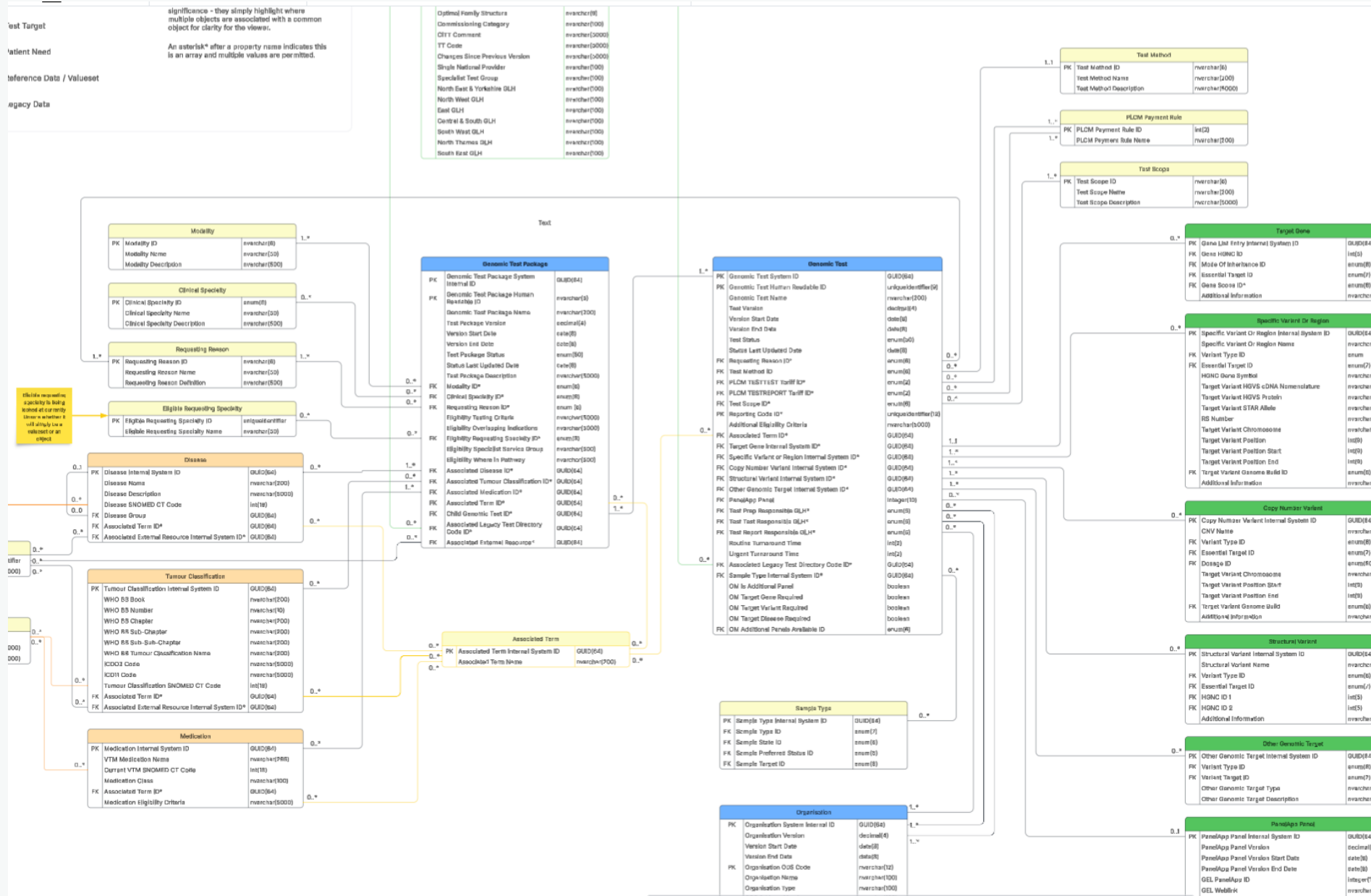
Version control for individual genomic tests with fully audited changes.

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.

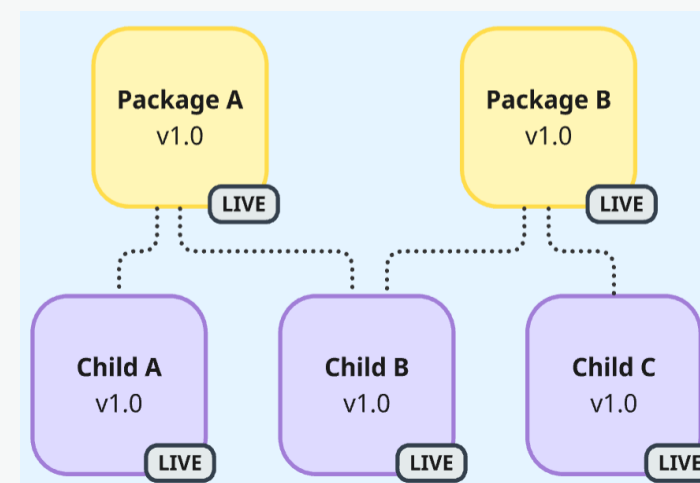
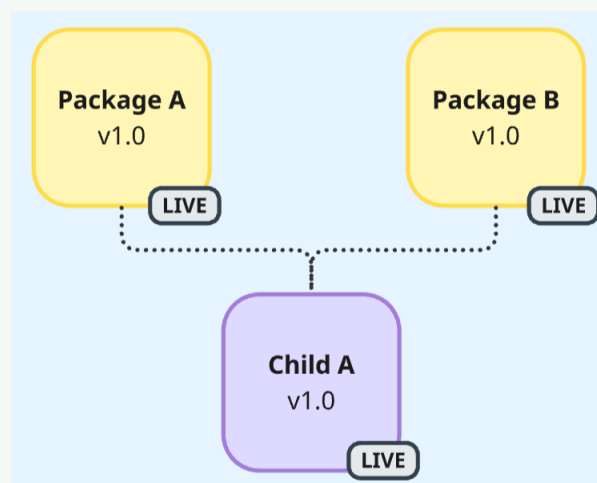
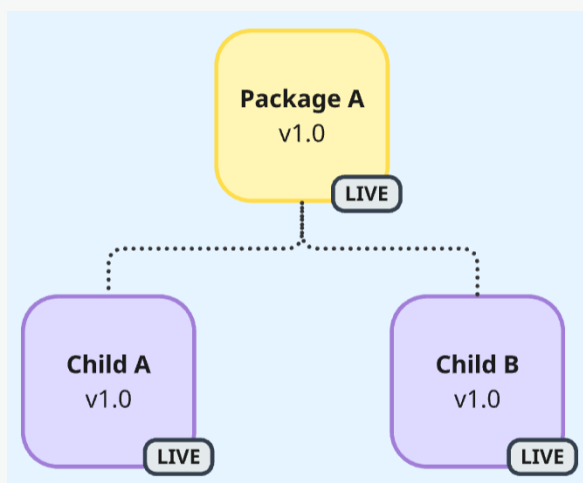
The model includes:

- **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) CBFβ-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) CBFβ-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

Pleural Fluid

Sample Origin • Somatic

Sample Preference Status • Accepted

Sample State • EDTA

Cerebrospinal Fluid

Sample Origin • Somatic

Sample Preference Status • Accepted

Sample State • EDTA

Blood

Sample Origin • Somatic

Sample Preference Status • Preferred

Sample State • EDTA

44

Back-end: data ingestion and 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

Gene Targets (0) Specific Variants and Regions (0) Copy Number Variants (0) **Structural Variants (36)** Short Tandem Repeats (0) GEL Panels (0) Other Targets (1)

Search e.g. BCR::ABL

Title	Variant Type	HGNC ID 1	HGNC ID 2
 NUP98::NSD1, t(5;11)(q35;p15.5)	Gene Fusion - Partner Specific	NUP98	NSD1
 MNX1::, 7q36	Gene Fusion - Partner Agnostic	MNX1	No value
 MLLT3::KMT2A, t(9;11)(p21;q23)	Gene Fusion - Partner Specific	MLLT3	KMT2A
 MNX1::ETV6, t(7;12)(q36;p13)	Gene Fusion - Partner Specific	MNX1	ETV6
 NSD1::, 11p15.5	Gene Fusion - Partner Agnostic	NSD1	No value
 CBFβ::, 16p13	Gene Fusion - Partner Agnostic	CBFB	No value
 RBM15::MRTFA, t(1;22)(p13;q13)	Gene Fusion - Partner Specific	RBM15	MRTFA
 MLF1::, 5q34	Gene Fusion - Partner Agnostic	MLF1	No value
 RBM15::, 1p13	Gene Fusion - Partner Agnostic	RBM15	No value

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

Tools

Select

Remove

exp/ode genes

gene_panels_mapping_t...

Transform

AIP

Edit

Aggregate genes to pane...

2 columns

Join

11 columns

Left dataset

Right dataset

ontology_GMS_V2_Panel...

create null columns for t...

28 columns

remove duplicate genes

27 columns

Clean HGNC ID

27 columns

Add vector embeddings

28 columns

ontology_GMS_V2_gene...

simplify panel type map

10 columns

Adding gene ID

8 columns

Left dataset

Right dataset

Creating ontology object

8 columns

Join on panel ID to creat...

9 columns

Left dataset

Right dataset

Creating automated test ...

8 columns

Dropping panel ID

7 columns

duplicate test target che...

3 columns

Joining duplicate check ...

8 columns

Left dataset

Right dataset

test gene target and pan...

2 columns

Test gene target and pan...

Legend

Gene Pipe... (13)

Panel Pipe... (10)

Gene Target... (7)

R&ID Auto... (13)

Add color

Selection preview

Preview

Suggestions 1

Pipeline warnings

ontology_GMS_V2_genes

Previewing 10 rows

28 columns

Search columns...

No inputs sampled

	vector	gene_id	hgnc_release_dat...	gene_version	variant_type	hgnc_uuid	hgnc_gene_aliase...	hgnc_prev_symbol	hgnc_mane_select	internal_system_...	hgnc_ge
	Embedded vector	String	Date	String	String	String	Array[String]	Array[String]	Array[String]	String	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	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	
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

Product | Geno... > MVP > ... > ontology_GMS_V2_Panels ☆

File ▾ Help ▾ | 1 1 1 master ▾

Preview

History

Details

Health

Compare

SQL preview

Analyze data ▾

ontology_GMS_V2_Panels

About

Columns

Schedules

Enter description...

[GMS V2] GEL Panel

Updated

7 hours ago by Foundry

Created

Mar 31, 2025, 10:29 AM by

Location

/NHS England-8e7d70/Product | Genomics Test Director...

Type

Dataset

RID

taset.b0adfbdb6-d5e5-4c13-b596-d1dfab7579fd

Size

11 columns | 906 rows | 2 files | 2.7MB

Updated via

ontology_genes_gene_panels

Tags

Add tags

Health Checks

View details

No checks

Inputs

Explore data lineage

ontology_genes_gene_panels

/NHS England-8e7d70/Product | Genomics Test Directory | Applications/M...

[GMS V2] HGNC Genes

/NHS England-8e7d70/CDIH | Metric | Genomics Medicine Service | Unmod...

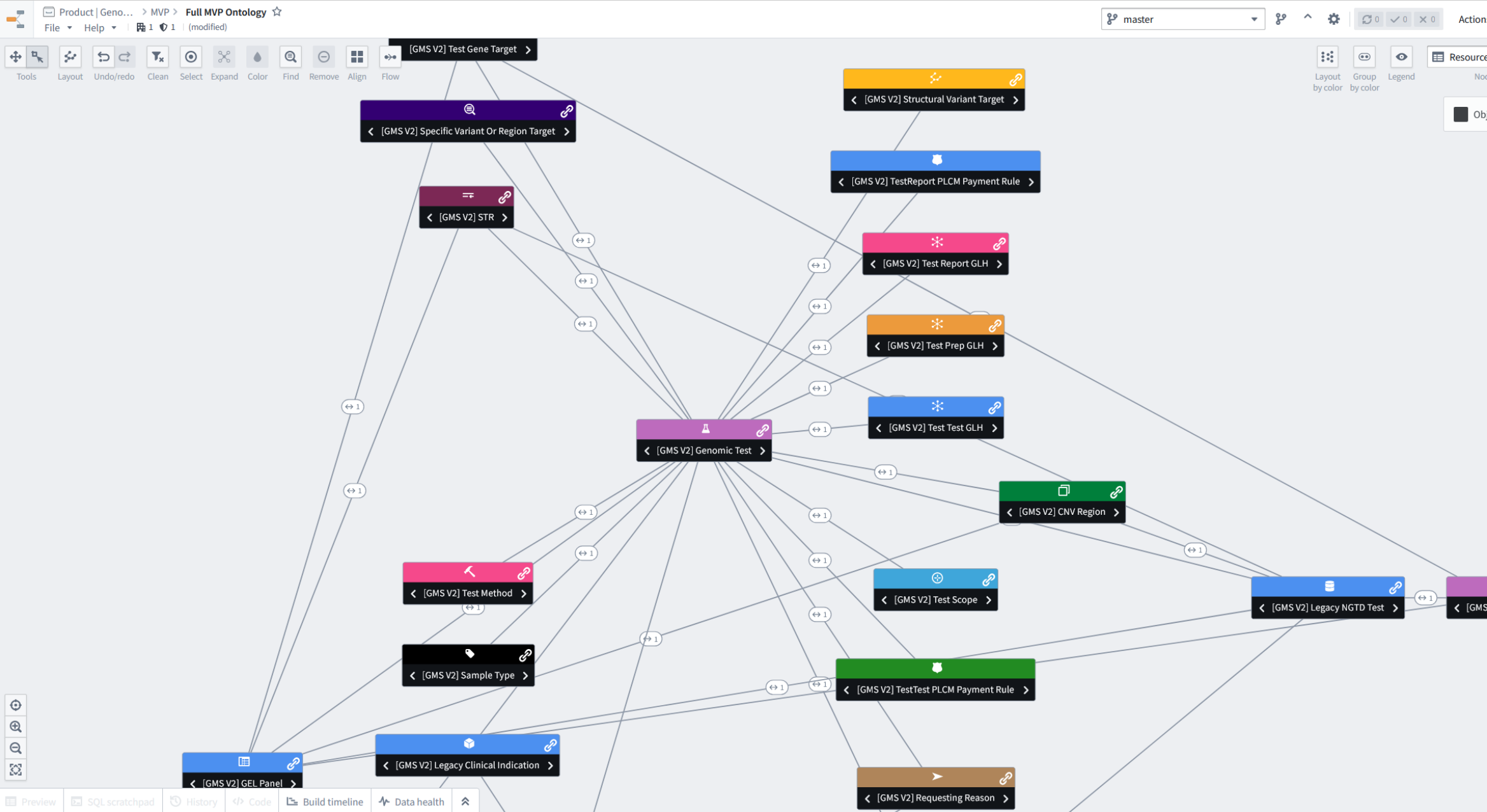
[GMS V2] Gene Panels

ontology_GMS_V2_Panels

	panel_type Array[String]	gms_panels_webli... String	gel_panelapp_web... String	internal_system_... String	version String	gel_panel_id String	panel_name String	ver... Stri
1	[Rare Disease 100K, GMS Rare Disea	https://nhsgms-panel	https://panelapp.gen	00acde5fbf571304b6f7	3.0	81	Clefting	202
2	[Cancer Germline 100K, GMS Cancer	https://nhsgms-panel	https://panelapp.gen	00bb2daface2ecaec3cc	3.0	59	Haematological malig	202
3	[Rare Disease 100K, GMS Rare Disea	https://nhsgms-panel	https://panelapp.gen	010b0df9ed19b67ad9e4	3.2	258	Arthrogryposis	202
4	[Component Of Super Panel, GMS sig	https://nhsgms-panel	https://panelapp.gen	012f2b00d9dfdbe1ec5c	4.0	725	Renal ciliopathies	202
5	[GMS Rare Disease, Component Of Su	https://nhsgms-panel	https://panelapp.gen	016209f2b936a335028e	6.0	477	Ataxia and cerebella	202
6	[GMS Rare Disease Virtual, GMS Rar	https://nhsgms-panel	https://panelapp.gen	01906e3b9d0a7ba85556	4.34	474	Adult onset neurodeg	202
7	[GMS Rare Disease Virtual, GMS Rar	https://nhsgms-panel	https://panelapp.gen	01e57aea79b15d1a8d91	3.2	658	Corneal dystrophy	202
8	[Rare Disease 100K, GMS Rare Disea	https://nhsgms-panel	https://panelapp.gen	026a88edd889a3420f9c	1.34	66	Rhabdomyolysis and m	202
9	[Rare Disease 100K, GMS Rare Disea	https://nhsgms-panel	https://panelapp.gen	02a685a0b76f0b81f060	7.0	162	Severe microcephaly	202
10	[GMS Rare Disease Virtual, GMS Rar	https://nhsgms-panel	https://panelapp.gen	03114c7920526cef908f	3.0	652	Dilated and arrhythm	202
11	[GMS Rare Disease, GMS signed-off]	https://nhsgms-panel	https://panelapp.gen	03f7e1ed5a57434608ef	2.0	537	Mitochondrial disord	202
12	[GMS Rare Disease Virtual, Super P	https://nhsgms-panel	https://panelapp.gen	045af0621479e1053434	72.1	486	Paediatric disorders	202
13	[Rare Disease 100K, GMS Rare Disea	https://nhsgms-panel	https://panelapp.gen	046e20044e2f6f308a0d	7.0	384	Limb disorders	202
14	[GMS Rare Disease Virtual, GMS sig	https://nhsgms-panel	https://panelapp.gen	048aae4b8346f7e9a5e0	2.2	481	Hypocalciuric hyperc	202
15	[GMS Rare Disease Virtual, GMS Rar	https://nhsgms-panel	https://panelapp.gen	0505daae85b9ecebc210	1.58	847	Childhood onset dyst	202
16	[Rare Disease 100K, GMS Rare Disea	https://nhsgms-panel	https://panelapp.gen	053ac9468aea3d4129af	4.0	3	Stickler syndrome	202
17	[Rare Disease 100K, GMS Rare Disea	https://nhsgms-panel	https://panelapp.gen	05e4acdb9a50fbadeabf	4.0	126	Monogenic hearing lo	202
18	[Rare Disease 100K, GMS Rare Disea	https://nhsgms-panel	https://panelapp.gen	0608f6522caab16db8d1	6.0	283	Cystic kidney diseas	202
19	[GMS Rare Disease Virtual, GMS Rar	https://nhsgms-panel	https://panelapp.gen	067f0bb94b29e6789bbc	2.0	478	Fetal anomalies	202
20	[Rare Disease 100K, GMS Rare Disea	https://nhsgms-panel	https://panelapp.gen	0696079c04215ed534b0	3.0	232	Congenital myaesthen	202
21	[GMS Rare Disease, GMS signed-off]	https://nhsgms-panel	https://panelapp.gen	07f98b4b491d1f29bd2b	1.0	1350	IPEX - Immunodysregu	202
22	[GMS Rare Disease Virtual, Super P	https://nhsgms-panel	https://panelapp.gen	0835fd90b7a2d5868284	19.6	841	Sudden unexplained d	202
23	[GMS Rare Disease Virtual, GMS Rar	https://nhsgms-panel	https://panelapp.gen	085c283a3f298c4f2df5	1.14	540	Adult onset movement	202
24	[Rare Disease 100K, GMS Rare Disea	https://nhsgms-panel	https://panelapp.gen	0921e436c9e69327fd9c	3.1	224	Short QT syndrome	202
25	[GMS Rare Disease, GMS signed-off]	https://nhsgms-panel	https://panelapp.gen	097da73a9d0279438363	1.4	649	Inherited phaeochrom	202
26	[Rare Disease 100K, Component Of S	https://nhsgms-panel	https://panelapp.gen	097ed4a1f433abc740cc	4.0	235	Distal myopathies	202
27	[GMS Rare Disease Virtual, GMS Rar	https://nhsgms-panel	https://panelapp.gen	09888483dce9d6c6d305	2.0	567	Hereditary spastic p	202
28	[Rare Disease 100K, GMS Rare Disea	https://nhsgms-panel	https://panelapp.gen	098df6c8b962cff9b3ab	5.0	81	Clefting	202
29	[GMS Rare Disease Virtual, GMS Rar	https://nhsgms-panel	https://panelapp.gen	099336a59ace92755177	2.178	474	Neurodegenerative di	202
30	[Rare Disease 100K, GMS Rare Disea	https://nhsgms-panel	https://panelapp.gen	09c6e9b07a50518ad859	4.0	269	Amelogenesis imperfe	202
31	[GMS Rare Disease Virtual, Super P	https://nhsgms-panel	https://panelapp.gen	09dd44213a14cf410f04	5.43	465	Neuromuscular disord	202
32	[GMS Rare Disease Virtual, GMS Rar	https://nhsgms-panel	https://panelapp.gen	0aa5975235963e6fb1a7	1.2	512	Congenital fibrosis	202
33	[Rare Disease 100K, GMS Rare Disea	https://nhsgms-panel	https://panelapp.gen	0ae930fffd19f020d2865	7.0	402	Early onset or syndr	202
34	[GMS Rare Disease, Component Of Su	https://nhsgms-panel	https://panelapp.gen	0bbb4a2fde6ce5214bb5	2.0	479	Paediatric disorders	202

Showing all 906 rows

Back-end: relational linkages



Back-end: join tables

Product | Geno... > *** > Initial DGTS test and panel join table ☆

File ▾ Help ▾ 1 1 master ▾

Preview History Details Health Compare

Initial DGTS test and panel join table

About Columns Schedules

Enter description...

[GMS V2] Genomic Test ↔ [GMS V2] GEL Panel

Updated May 27, 2025, 4:53 PM by

Created May 2, 2025, 1:50 PM by

Location /NHS England-8e7d70/Product | Genomics Test Director...

Type Dataset

RID .dataset.8c0e715e-2914-4d9b-8c2c-6c9aaefbet

Size 2 columns 311 rows 2 files 2.1MB

Updated via ontology_genes_gene_panels

Tags

Add tags

Health Checks

View details

Initial DGTS test and panel join table

	panel_id String	test_id String
1	c960e1099d6a9275eca584e36f19d73f94d174948	00e93434d0822a752ea3404ca29aa5757b2d2cdd0
2	f4dff371b0e31388407746a5d760bec23a1fee8cb	010222ae0d580d3a573f21853772f6edde9d4ae06
3	bade93def3e26eae5afe547696a83b4d895bfee88	0113e4c74a83dc179d9d250727da760ed2ee80b13
4	34e861ebe2d4f68ae2aa2ae89b3a2ff0929266ced	013fb716137fd7964d6b1398ffbab8b5b1f276a0c
5	5d04d2c02b268f5fbf52a1c7267e57d4cd05bc718	02e7fcf08be3312954004b4463a5d1582ec677f89
6	683520a43e14bb3a339ed9eaa92ce6c759411ccfb	03a1c824ce7f68bf3b43cfc407027966f9c2c7bb0
7	def5cf0e489690648ef7d8716a4d1748f825ba326	040001ce542d474ef9dd63e7bac72fe64bdbabeeff
8	bc40e2affa3f402408e0fccfd548f6335adbcc97	055eea0aa6055d7250f2d582409569b7ff3b0541e
9	5c5028f44e6f296d65d0ca05811eddb5b1f711144	06c797523dec40ef0ab4a7166997c02eaddf14db6
10	4a79a1ad95d700f40c379b07a9c0be11b137f9771	06e5062f6639ed1ebe3adc46c3804f8dd90db9b59
11	574c3cf0b90cc2272cda90b0f4959c3f0a6705ae4	06f657cba6fab8ad0236d48e7e631d40dca437a94
12	0bbccf5a9be9fc102cc8ce7834aba03a3f13b7764	0744216e82a5bcfc2262e91d1ea50bfc1d3ae399a
13	54969217a2fc8e2b4d64248a20ff2c8f13e379811	07d7e8852c72419e88b2677af9be8f9978d6f3e43
14	e4674a1e98346e6b257b71dc469db08cdbce69ede	080df9e608317c1a0ed55cb0193e9996dfcef3250
15	e4e0d01d5957ebb6a4aa173f5f9993ed09fced68d	0952e0614a8712f08ec38dff231e8c491bcb2ced0
16	73041c6876f6dc5473be8dd8e3ee54f9001e9bc8d	0aaaac4d852f35184eb557ff4981094427f05bd5e
17	994fddbc02578c1531bf894f69d23f220ffd286de	0b1fb9a7469b3adc50478cf6bfc3a193432c108fb
18	7761f148754e28c6d1536736a73da682c8d974459	0b975e9ede71e3390d654d5c918911652a50d8763
19	e723bb4595e06b43bd109d4fee76b112219788ef1	0ba4af9a48fac0ec2029a0bbd6375e1fd3d4734e3
20	2debbcb9d014c03c3a8dbe0aa582c3b2537737381	0ba522ddd99108e6460d995af3920d90c77cfcd96
21	bade93def3e26eae5afe547696a83b4d895bfee88	0c8326f4d070b5980d749ebd4800d7664555b6f27
22	b1b8503727ba5a1aba99be6c51896974d97adacfb	0cbf6b71ad04fa5d8e2bc062c127321b0fb80599d
23	d620f6b8788ae4be9b25c4ed7c2fa9fad3700ce8d	0cc5c7bde942ec2cb83e86f49d0230b8074e14fe9
24	fd5b3424a919993c0f90702f933c805f001cd4ac5	10a098b7e4896489968990eff8a3cf91f8c60e3ff

50

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.



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?

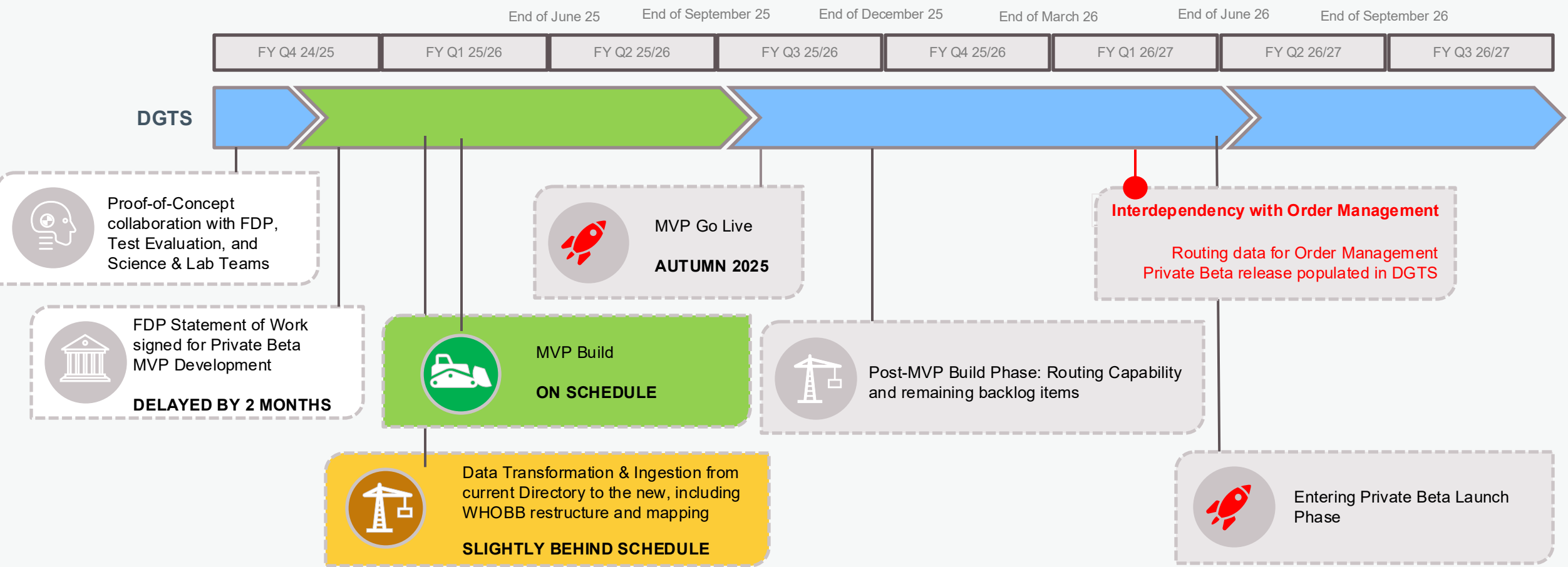
1. Front-end web application
2. Test packages and child genomic tests and changes to portfolio
3. Test identifiers
4. New Excel and PDF files
5. Test-level versioning and status
6. **DGTS API**
 - **DGTS reference data will be exposed via API.**
 - **Exact methods of access are to be agreed (e.g. is 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



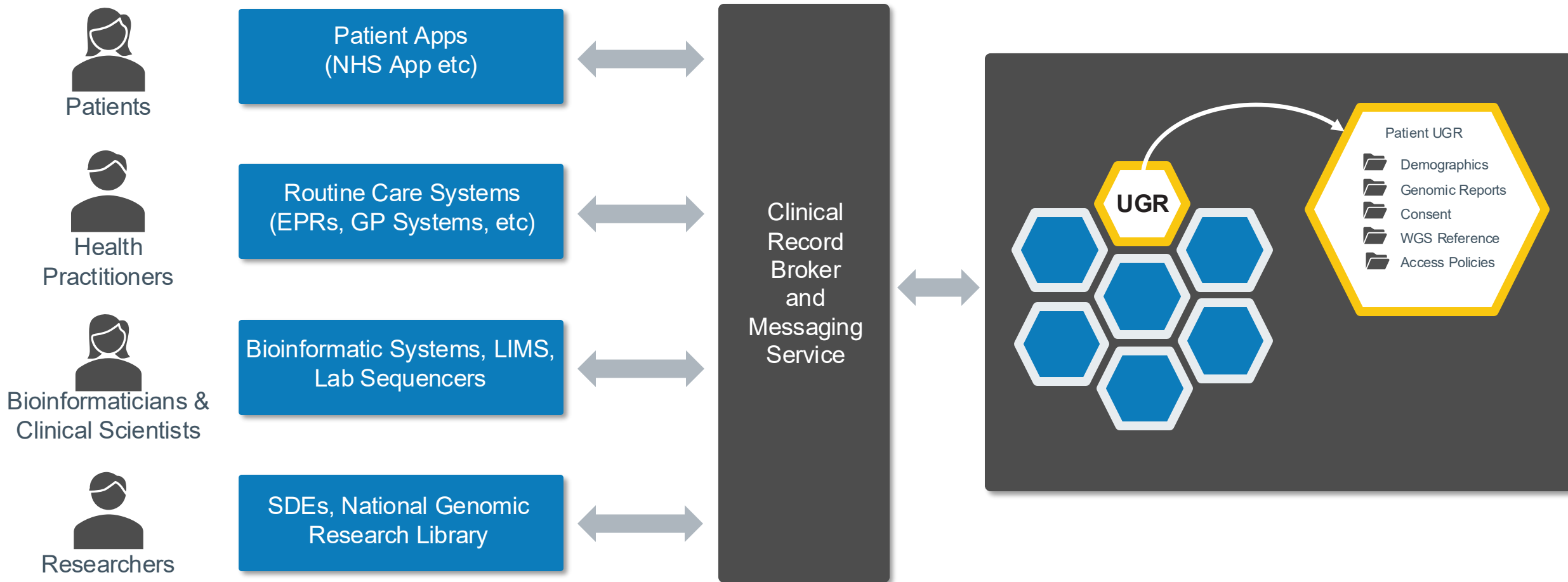
SPR and UGR

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



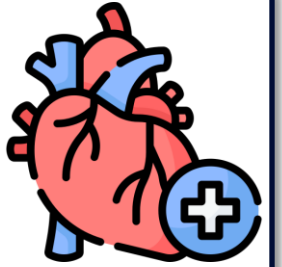
Joining with other datasets to support broader identification



Consent integration

What are Inherited Cardiovascular Conditions?

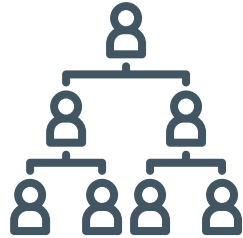
Inherited cardiovascular conditions are heart or blood vessel disorders passed down through families. They can affect how the heart functions and may increase the risk of serious events like heart attacks or sudden cardiac arrest.



How does the UGR help with ICCs



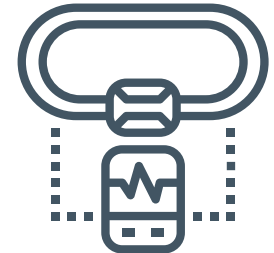
Removing database siloes to improve research potential



Identify family members potentially at risk

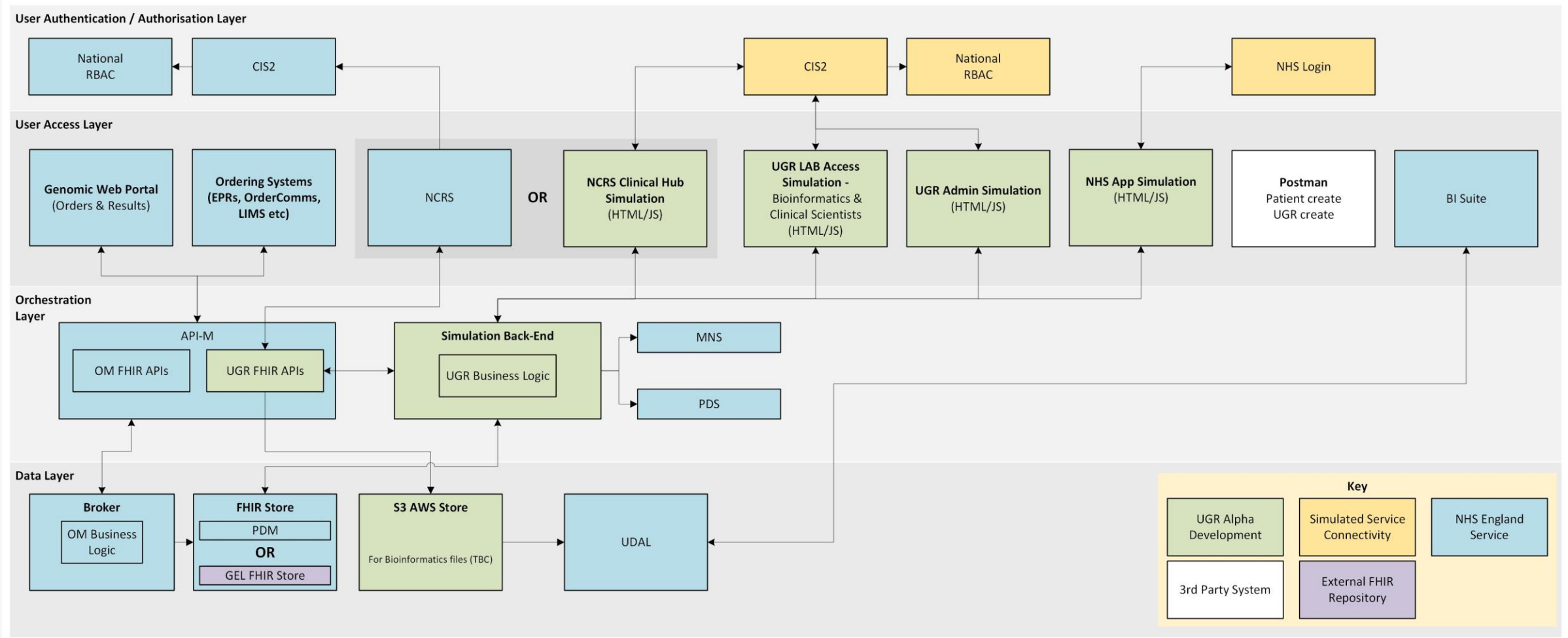


Reduce admin burden of notifying family members.



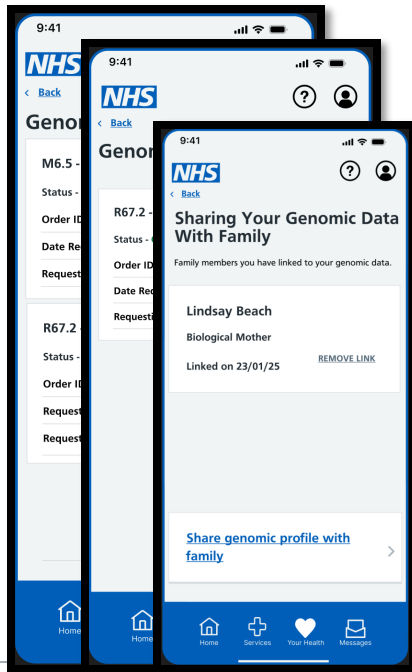
Routine care data will be mobilised for research

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



MS SANDY BEACH 45 years old, Female
Date of Birth: 01-Aug-1980 Address: The House, 79 Road Lane, LEEDS, LS23 5ER
NHS Number: 000 000 0000

Overview Patient Clinical Documents **Genomics**

Genomics

Key Genomic Information

- Genomic Overview
 - Genomic Reports
 - Linked Family Members
 - Genomic Orders
 - Raw Sequence Data
- ### Data Access
- Data Access Overview
 - Familial
 - Research / MI / Population Health

Genomic Overview

Notifications

Date Created	Detail	Status	Date Actioned	Link
10/02/24	Secondary findings - review required	Active		Report
02/02/24	Eligible for clinical trial	Active		Detail
01/11/24	Familial findings - review required	Active		Profile
11/01/24	Genomic report returned	Actioned	12/01/24	Report

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Consultant view - Notifications

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

MS SANDY BEACH 45 years old, Female
Date of Birth: 01-Aug-1980 Address: The House, 79 Road Lane, LEEDS, LS23 5ER
NHS Number: 000 000 0000

Overview Patient Clinical Documents **Genomics**

[Go back](#)

Genomics

Key Genomic Information

[Genomic Overview](#)
[Genomic Reports](#)
[Linked Family Members](#)
[Genomic Orders](#)
[Raw Sequence Data](#)

Data Access

[Data Access Overview](#)
[Familial](#)
[Research / MI / Population Health](#)

Genomic Orders

R67.2 Monogenic Hearing Loss

Status - **Complete**

Order ID	1102912
Date Requested	19/12/2024
Requesting Organisation	Oxford
Requester	Dr Jack Jones
Test Order Form	View
Reporting Laboratory	Liverpool
Date Report Issued	22/12/2024
Report	View

M6.5 DPYD Hotspot

Status - **Accepted**

Order ID	1102913
Date Requested	19/03/2024
Requesting Organisation	The Christie
Requester	Dr Lucy Batt
Test Order Form	View
Reporting Laboratory	N/A
Date Report Issued	N/A
Report	N/A

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Consultant view - Genomic Test/Report Details

- Test order information
- Report information

MS SANDY BEACH 45 years old, Female
 Date of Birth: 01-Aug-1997 Address: The House, 79 Road Lane, LEEDS, LS23 5ER
 NHS Number: 000 000 0000

Overview Patient Clinical Documents **Genomics**

< [Go back](#)

Genomics

Key Genomic Information

[Genomic Overview](#)

[Genomic Reports](#)

[Linked Family Members](#)

[Genomic Orders](#)

[Raw Sequence Data](#)

Data Access

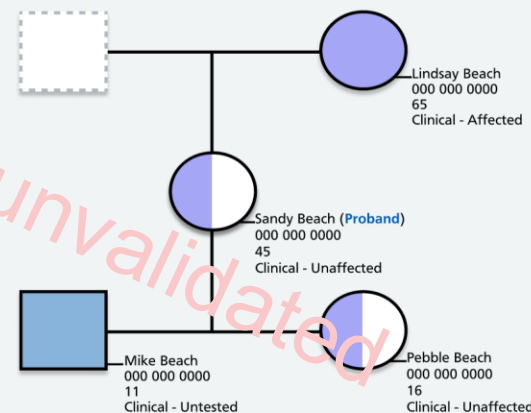
[Data Access Overview](#)

[Familial](#)

[Research / MI / Population Health](#)

Family Members linked to Sandy Beach

Disease: [Inherited Breast Cancer](#)



Other Diseases:

[Familial Hypercholesterolemia](#)

Consultant view - Linked family members

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

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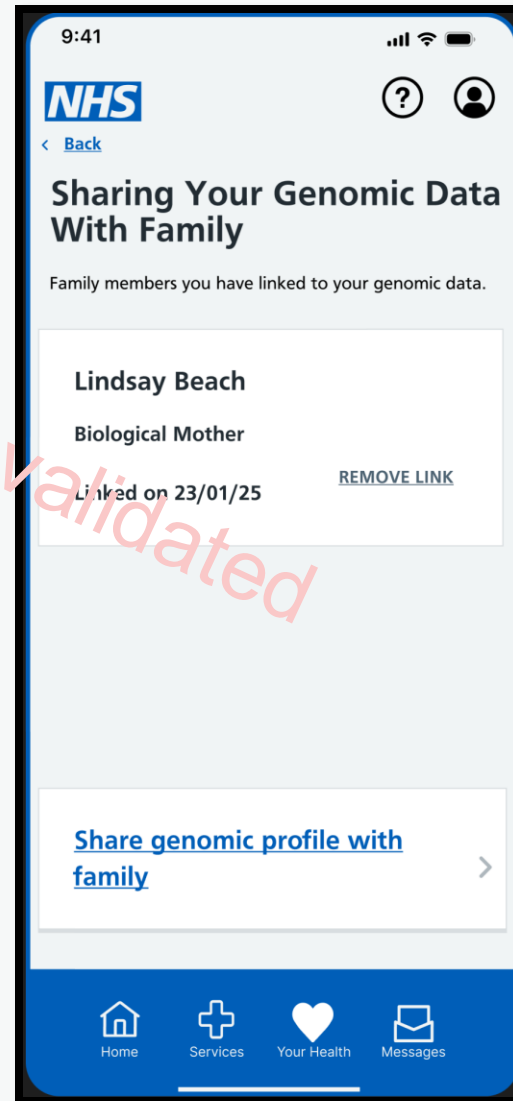
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Patient view - NHS App

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

Find patients for research

By NHS number

By Further Criteria

Demographics

Years of Birth

Sex

Ethnicities

Clinical History

Current conditions

Current medications

Previous medications

Genetics

Gene name

Variant type

Privacy Information

This application gives you access to patients' personal and confidential information. You should only access this information when it is necessary for your role and you must have a legitimate reason for doing so.

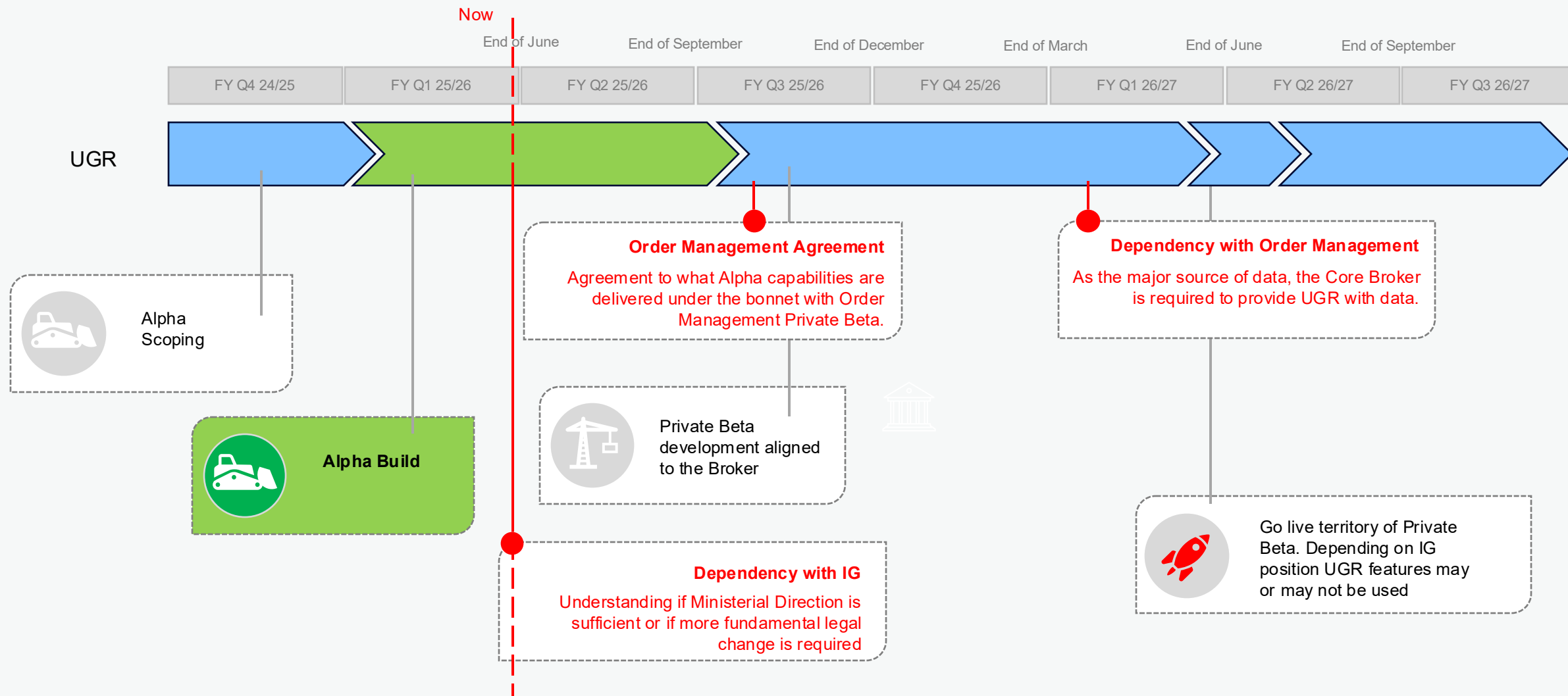
Patients will only return on this search if they have consented to research.

Access to this information is monitored. If there is inappropriate access suspected then the Privacy Officer will investigate.

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

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

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



Discovery

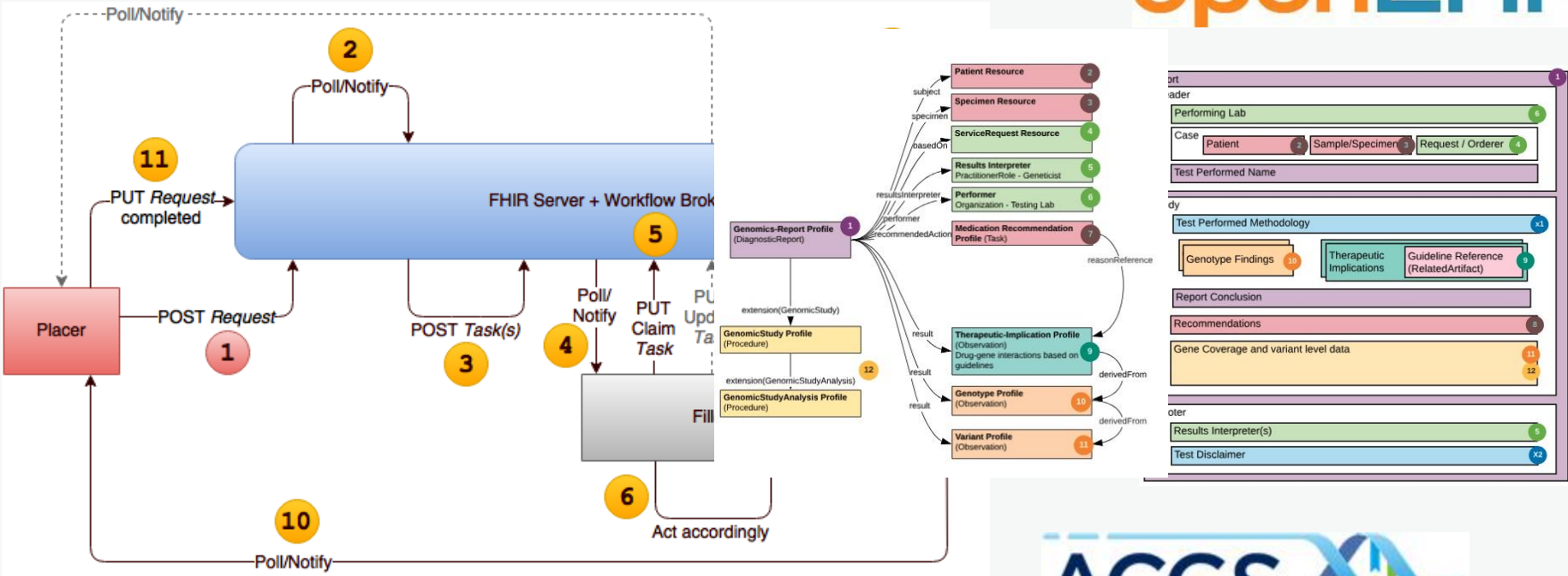


NHSDigital/Pathology-FHIR-Implementation-...

A repo to hold FHIR examples for the Pathology Implementation Guide

3 Contributors 0 Issues 0 Stars 0 Forks

NHS Data Model and Dictionary



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



HL7 Version 2 to FHIR
 1.0.0 - STU 1



[IG Home](#)
[V2 to FHIR](#)
[Mapping](#)
[Implementation and Use](#)
[Artifact Index](#)
[Table of Contents](#)

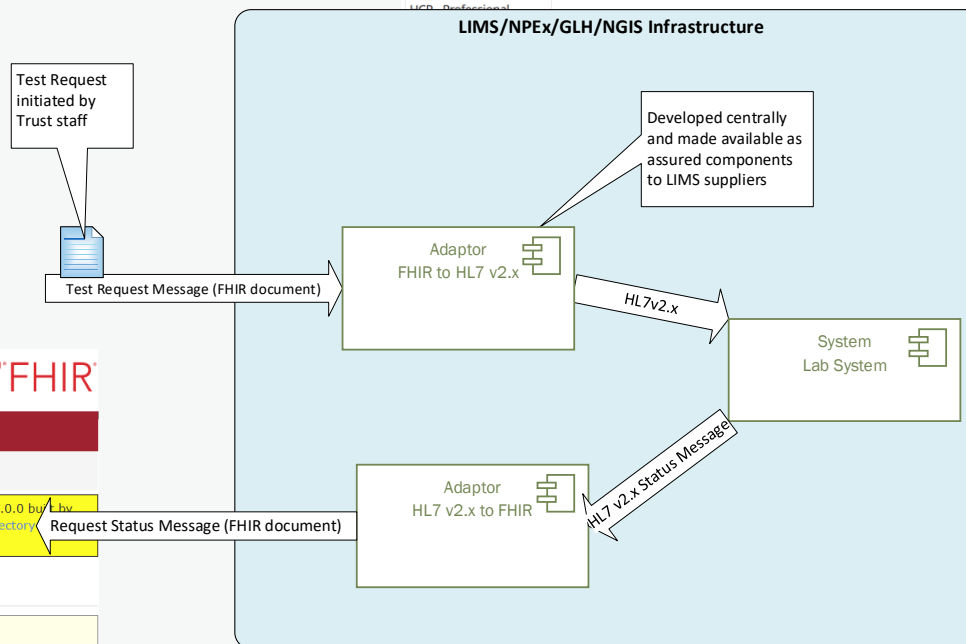
[Table of Contents](#) > [V2 to FHIR](#)

HL7 Version 2 to FHIR, published by HL7 International / Orders and Observations. This guide is not an authorized publication; it is the continuous build for version 1.0.0 by the FHIR (HL7® FHIR® Standard) CI Build. This version is based on the current content of <https://github.com/HL7/v2-to-fhir/> and changes regularly. See the [Directory](#) published versions.

1 V2 to FHIR

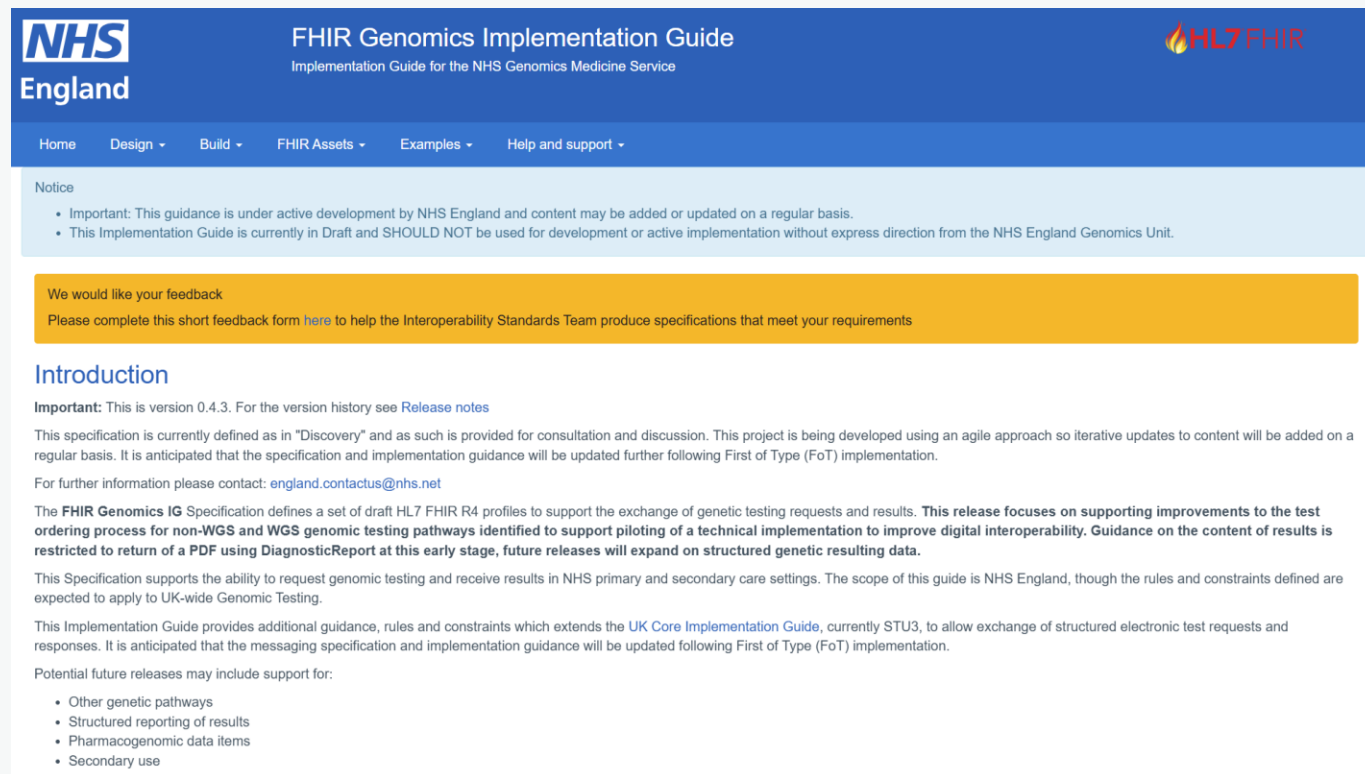
Official URL: http://hl7.org/fhir/uv/v2mappings/ImplementationGuide/hl7.fhir.uv.v2mappings	Version: 1.0.0
Active as of 2025-01-15	Computable Name: HL7Version2toFHIR

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.
HCP - Professional registration number type		ORC-12.9	HCP's professional registration number type such as GMC.
HCP - Report preferred delivery method		CTD-6	HCP report preferred delivery method.
Central email address provided by the HCP.		Additional CTD segment (CTD-5.4)	



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)



The screenshot shows the NHS England FHIR Genomics Implementation Guide website. The header includes the NHS England logo, the title 'FHIR Genomics Implementation Guide', and the subtitle 'Implementation Guide for the NHS Genomics Medicine Service'. A navigation bar contains links for Home, Design, Build, FHIR Assets, Examples, and Help and support. A notice section states that the guidance is under active development and should not be used for development or active implementation without express direction from the NHS England Genomics Unit. A feedback section invites users to complete a short feedback form. The introduction section provides details about the version (0.4.3), the purpose of the specification (supporting improvements to the test ordering process for non-WGS and WGS genomic testing pathways), and the scope of the guide (NHS England, though rules and constraints are expected to apply to UK-wide Genomic Testing). It also mentions that the guide extends the UK Core Implementation Guide (STU3) to allow exchange of structured electronic test requests and responses.

NHS England FHIR Genomics Implementation Guide
Implementation Guide for the NHS Genomics Medicine Service

Home Design Build FHIR Assets Examples Help and support

Notice

- Important: This guidance is under active development by NHS England and content may be added or updated on a regular basis.
- This Implementation Guide is currently in Draft and SHOULD NOT be used for development or active implementation without express direction from the NHS England Genomics Unit.

We would like your feedback
Please complete this short feedback form [here](#) to help the Interoperability Standards Team produce specifications that meet your requirements

Introduction

Important: This is version 0.4.3. For the version history see [Release notes](#)

This specification is currently defined as in "Discovery" and as such is provided for consultation and discussion. This project is being developed using an agile approach so iterative updates to content will be added on a regular basis. It is anticipated that the specification and implementation guidance will be updated further following First of Type (FoT) implementation.

For further information please contact: england.contactus@nhs.net

The **FHIR Genomics IG** Specification defines a set of draft HL7 FHIR R4 profiles to support the exchange of genetic testing requests and results. **This release focuses on supporting improvements to the test ordering process for non-WGS and WGS genomic testing pathways identified to support piloting of a technical implementation to improve digital interoperability. Guidance on the content of results is restricted to return of a PDF using DiagnosticReport at this early stage, future releases will expand on structured genetic resulting data.**

This Specification supports the ability to request genomic testing and receive results in NHS primary and secondary care settings. The scope of this guide is NHS England, though the rules and constraints defined are expected to apply to UK-wide Genomic Testing.

This Implementation Guide provides additional guidance, rules and constraints which extends the [UK Core Implementation Guide](#), currently STU3, to allow exchange of structured electronic test requests and responses. It is anticipated that the messaging specification and implementation guidance will be updated following First of Type (FoT) implementation.

Potential future releases may include support for:

- Other genetic pathways
- Structured reporting of results
- Pharmacogenomic data items
- Secondary use

Questions?



Wrap up



Thank You



@nhsengland



company/nhsengland



england.nhs.uk