

# High Level Design

Genomics Target Architecture

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**Related documents**

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| 1 | Process Architecture Analysis | Michael Price | TBC |
| 2 | Genomics Testing Pathways Tube Matrix | Adam Laurent | <https://future.nhs.uk/NHSgenomics/view?objectId=126420069> |
| 3 | Future Informatics Enterprise Architecture artefacts | Anne Crowther | TBC |
| 4 | Genomics Target Architecture Options Appraisal v0.7 | Danny Ruttle | <https://future.nhs.uk/NHSgenomics/view?objectId=132251973> |
| 5 | GTODS Requirements Catalogue v0.5 | Danny Ruttle | <https://future.nhs.uk/connect.ti/NHSgenomics/view?objectId=135800773> |

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**Glossary**

|  |  |  |
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| **Acronym/Term** | **Term** | **Description** |
| CIS2 | NHS Care Identity Service 2 | NHS Care Identity Service 2 (NHS CIS2), formerly known as NHS Identity, is a new, secure authentication service used by health and care professionals in England to access national clinical information systems. |
| CLOB | Character Large Object | A large block of text stored in a database in some form of text encoding (e.g. text containing a FHIR resource). |
| FHIR | Fast Healthcare Interoperability Resources | Global industry standard for passing healthcare data between systems. |
| GTODS | Genomics Test Order Data Set | The information that needs to be included to inform a laboratory of what is required for a given diagnostic genomics test. |
| iFrame |  | An inline frame used inside a webpage to load another HTML document inside it – within the context of this document this is how the Web Form application can be presented to the user. |
| MESH | Message Exchange for Social Care and Health | The main secure large file transfer service used across health and social care organisations, generally adopted for asynchronous exchange patterns. |
| OAuth2 |  | OAuth 2.0 is an industry-standard protocol for authorization, assignment of claims and permissions. |
| OIDC | OpenID Connect | OpenID Connect is a simple identity protocol and open standard that is built using the OAuth 2.0 protocol. |
| PLCM | Patient Level contract Monitoring | PLCM enables the interchange, in a uniform format, of monthly PATIENT level Contract Monitoring data between all purchasers and Health Care Providers |
| RBAC | Role Based Access Control | Role-based access control (RBAC) is a way of ensuring that users are suitably authorised. It is implemented across the NHS when accessing nationally hosted services. |
| SNOMED | Systemized Nomenclature of Medicine | Structured clinical vocabulary used across the NHS in electronic health records |
| TRE | Trusted Research Environment | TRE provides approved researchers with access to essential linked, de-identified NHS health data |
| TRUD | Terminology Release Update and Distribute | Service hosted by NHS Digital for the distribution of terminology and reference data |
| UTL | Unified test List | The national set for codes for standardised representation of names of testing being requested and reported on the national specification for sharing laboratory/pathology results between organisations (SNOMED) |

## Purpose

The purpose of this document is to present options to support improvements to the test ordering process for non-WGS (and WGS) genomics testing pathways identified to support piloting of a technical implementation to improve digital interoperability. This document discusses the target architecture and patterns of message exchange, however there may be some short-term solutions that act as enablers to meeting the recommended option.

## Problem Statements and Requirements

The problem Statements and Requirements covered by this document are discussed below.

### Genomics tests are ordered via paper forms

Unlike regular pathology test orders, genomics tests are usually recorded on paper and included with the sample(s). The details are then keyed into the LIMS at the first lab where the sample arrives. The test details are often scanned and attached to the order as a PDF; it isn’t possible to capture the scanned information in a structured form on the patient’s medical record from a scanned document.

Requirements:

As a clinician, I want to enter the Test Order details electronically when the order is created, to remove the need for re-keying, thus reducing data quality errors and improving patient safety.

As a clinician, I want the data entered to be structured in coded form so that it can be saved to the patient’s medical record for review.

### Lack of Management Information relating to “Send Aways”

Depending on the type of test being ordered, the order details and the specimen/sample can traverse multiple labs (e.g. wet labs, plating, etc..). Currently the status of Test Orders referred across multiple laboratories isn’t visible to the requestor.

Requirements:

As a clinician I require updates on the status of a test order to support patient care and aid with clinic planning.

As a Laboratory Manager, I want to see the status of a test order at every stage from receipt to results reporting so that I can manage clinician expectations and ensure turnaround is within our SLAs

### Genomics does not align with nationally approved information standards

Information transmitted via Genomics testing does not align with nationally approved interoperability standards.

Requirements:

As an informatics lead, I require metrics on all genomics Test Orders, to provide reports at lab, regional and national levels.

As an informatics lead, I require all genomics Test Order details to align to a nationally approved standard, to support interoperability with other domains.

As a clinician, I require information to be structured and align to a nationally approved information standard, to improve the quality of information available to support the care provided to patients (this includes information sent in a Test Order and the Results returned following diagnostic analysis).

### Non-Functional Requirements

The following requirements are a combination of strategic aims and other activities within NHSE&I.

#### Align with NHS National Services

There is a strategic technical requirement to align with NHS National Services where this adds benefit to the end-to-end solution.

#### Support FHIR

HL7 FHIR is the international standard that is being adopted across all healthcare settings within NHS Health and Social Care to support interoperability. The proposed solution should align to FHIR wherever possible.

#### Scalability and Volume

There is an initiative to undertake WGS for all new-borns. The recommended solution must be capable of scaling to meet expected volumes.

#### Strategic Aim for Central Portal/Repository

NHSE&I has stated that all pathology tests are to be stored in a central repository available nationally to support Management Information. Genomics tests are a subset of pathology and should therefore align with the requirements laid out in this area.

#### Align to Pathology Strategy

The Pathology team at NHS Digital needs to implement pathology standards to support implementation of its own strategy. Work carried out within Genomics must align with this strategy where it is sensible to do so.

## Scope

The scope of the Target Architecture includes Test Ordering flow, Status Updates Test Results. For the avoidance of doubt the limits are set out below:

### Agreed Genomic Testing Pathways

See Ref 3 in the Related Documents section for the seven pathways that have been adopted to support discovery work.

### Ordering process to the first LIMS system

It is expected that the first LIMS will have the capability to receive genomics Test Orders using nationally approved information standards (FHIR R4).

### Status information on all steps

All systems involved in the end-to-end process need to support reporting of the status of both the Specimen and the Test Order as they move between different laboratories and “send away” LIMS.

### Test Results/outcome reporting

Work is underway within the North Thames GMSA to support the reporting of results in FHIR. This area will be addressed once the approach of this parallel piece of work is understood. The objective of this piece in the architecture is to allow relevant information to be returned to the requestor in coded (as well as free form summary) to be stored on the patient’s record.

### Genomics Test Order Data Set (GTODS)

Test Orders require a description of the test that needs to be carried out, in a format understood by the LIMS and which is also simple to interpret.

The Genomics Test Directory exists, however its structure lends itself to commissioning and decision support for clinicians. It can be used to derive the relevant set of genomics tests for a given patient presenting with clinical indications, but it isn’t presented in a format that is simple for LIMS to load the information as a set of reference data.

GTODS is a data set which describes the Tests in sufficient detail for LIMS to load the data. Whilst it shares some of its information with the Genomics Test Directory it has different requirements and it requires a separate specification. Alignment with the NHS Data dictionary and SNOMED as a national standard is something that should be pursued. A key decision requiring appraisal is around a separate list of Tests for Genomics vs inclusion in the pathology UTL (Unified test List). GTODS is focused on coding the Test Orders in SNOMED rather than Test Results.

### Specimen Identification

It is assumed that an identifier of some description is captured at the point where the specimen is taken – either a barcode or a QR code. A separate piece of work is required to investigate options for adoption of an appropriate standard and related set of processes. There are existing solutions which are used for regular pathology tests and it is expected that any solution proposed would align with these.

### Information Standards

All systems which receive the initial Test Order are required to support the ingestion of information using a nationally approved standard for information exchange, HL7 FHIR R4. All systems at each step of the process must have the capability to transmit status updates in HL7 FHIR R4.

### Out of Scope in the initial phase

#### Interoperability between different LIMS systems for “Send Aways”

Local processes between LIMS systems for transferring Test Order details and specimen/samples are unaffected by the proposed architecture. Currently the majority of LIMS systems support HL7v2.x messaging and this isn’t expected to change in the short term. In fact, it is very unlikely for some older systems to be able to change from HL7v2.

A separate piece of work is required to investigate options for adoption of an appropriate standard and related set of processes.

## Target Architecture

### Test Order

An architecture appraisal (see ref 4) focussing on the logical place in which to initiate Test Orders for genomics tests recommended a centrally hosted solution, which is described in detail below.

Web form and FHIR API components are hosted centrally on a single logical system acting as a message broker, with four options for integration to the Test Order system for Trusts, as follows:

1. Existing lab order component within the PAS integrates directly to the FHIR API.
2. iFrame integration with a web based PAS, supporting silent login/session management and click through of patient details from the PAS.
3. Separate log in to Web Form component/application hosted centrally.
4. Separate log in to Web Form component/application hosted locally which integrates with the FHIR API.

Integration options 1 to 3 are shown on Figure 1 and option 4 has been added to Figure 2. All integration options are shown on Figure 3.

Users and systems authenticate using OIDC and OAuth2/RBAC via CIS2. LIMS systems integrate with a single API service for receiving Test Orders and sending Status updates (and Results). This centralised topology removes the need to secure numerous point to point connections and allows any authorised user or system to access Management Information to support national level Management Information.



Figure – Test Order, Data Capture Centralised – integration options 1 to 3



Figure - Test Order, Data Capture Centralised – integration options 1 and 4



Figure - Detailed Interaction Diagram

### Components Overview

The options presented in the diagrams above leverage numerous components to support interoperability. Each component is described at a high level below to aid understanding.

#### Web Form Component

This is a web application, its primary function being to allow the person initiating the Test Order to capture all relevant information for a given test. Providing this capability removes the use of paper but not necessarily the re-keying of information, however it does act as an accelerator for clinicians to initiate the order electronically.

Note that the Web From Component is a **tactical tool** and as such a suitable retirement policy must be in place before it is released to users.

There are several options for implementation including:

1. Following the adaptor approach (see section 4.2.3), develop centrally and provide as a pre-assured self-contained component, which is hosted centrally (integration option 3)
2. Deploy the component to any infrastructure including the Trust’s own hardware or cloud hosting environments (AWS, Azure, GCP).
3. Publish a specification against which local implementations are assured and allow local Trusts to re-use any existing forms integration capabilities.

The implementation of all options would be assured by NHS Digital as a delivery partner.

All options must integrate directly with the interface exposed on the FHIR API component.

The Web Form application requires a rules/configuration subcomponent which will dynamically adjust the fields in the form to ensure the appropriate information is captured for a given genomic testing pathway (noting that this will change as the list of tests that can be ordered are extended or updated). It may be challenging to maintain a separate rules engine subcomponent that functions with local implementations discussed on 2 above. Depending on the option chosen this logic could be delegated to an API.

The component also supports an export mechanism where the details of any Test Order can be saved to the user’s local infrastructure. This is an attended process (a user is present) and allows details of the order to be stored against the patient’s record in the local PAS/EPR.

#### Test Order API Component

This is a Test Ordering component which exposes a FHIR API externally and manages the exchange of information to components which create or receive Test Orders and provide status updates as well as results. It also has internal API methods to support logic such as deriving a default route for the initial LIMS system and for transforming the data stored in its database to and from FHIR. The API’s key functionality is listed below:

* Accepts and validates a Genomics Test Order.
* Supports amendments to Test Orders. [[1]](#footnote-2)
* Provides access to the Genomics Test Order Data Set (GTODS)
* Manages access to Test Order records.
* Derives the default routing to the Initial LIMS (see section 10).
* Accepts Status Updates from LIMS initially but could be from any other downstream LIMS in the workflow).
* Provides status of all Test Orders including “Send Aways”.
* Provides logging of all activity to support Management Information.
* Transforms all information collected via the Web Form into the appropriate FHIR document.
* Manages access to its datastore.
* Delivers the validated FHIR document to the initial recipient using the appropriate method.
* Supports Publish and subscribe capability if this provides benefit.

#### Adaptor Component

All options described within this section require that the LIMS receiving the initial Test Order supports FHIR. In the short term this is unlikely to be the case; furthermore, to interoperate nationally this would require all LIMS across the GLH network to be compatible with FHIR on day one.

To mitigate this issue Adaptors are proposed which will translate between HL7 FHIR R4 and HL7v2.x in each direction as required, as shown in Figure 4 below. This approach has already been employed to support GP system suppliers with alignment to FHIR standards (and to simplify market entry for new suppliers in Primary Care). Adaptors are provided free of charge to suppliers under an open source licence (see: <https://opensource.org/osd>) with ongoing maintenance provided from the centre. The adaptors are supplied as pre-assured self-contained components hosted in containers and are deployable to any infrastructure including cloud hosting environments (AWS, Azure, GCP).



Figure - Tactical Adaptors

Developing the adaptors centrally provides benefits in terms of adding consistency for all parties and also allows functionality to be assured in one place.

With the central development approach there is effort required to carry out the mapping logic between the different standards and clinical coding between FHIR and HL7v2 and ongoing maintenance to support:

* Updates to support new FHIR versions
* Mappings of new coded values between SNOMED and other ontologies
* Security patches

There may be a need to constrain scope of legacy versions HL7 version 2 (i.e. V2.3 and V2.8) and also account for variations from the HL7v2.x standard such as Z Segments.

#### Lab System Component

The Lab System component represents any existing LIMS (Laboratory Information Management System).

#### PAS/EPR Component

This component is the system deployed at a given Trust and supports the administration of patient healthcare and storing/management of patient records. There are many different systems used by different NHS organisations all with differing levels of maturity and integration capabilities. In some cases the PAS/EPR component could represent a LIMS within the Trust.

In the options appraisal the central solution scored high in most areas and is presented below.

|  |  |  |
| --- | --- | --- |
| Measure | Score | Notes |
| Meets core requirements | 5 | All requirements are covered. This also aligns to GEL and GMS-wide objectives for a clinician to be able to access the status of a test order from any location.  The home lab’s responsibility of reporting status and outcome becomes redundant as the odering system has access to all information related to the Test Order. |
| Cost effective | 4 | Single API and Web Form component |
| Complexity of implementation | 4 | * Single API acts as a broker for all stakeholders. * All connections to support FHIR exchange are secured to a single service. * Trusts can integrate at 4 levels:   + Direct EPR integration to FHIR API   + iFrame silent/cached login to Webform   + Separate login to Web Form   + Locally built Web Form integration * Medium component deployment count. * Rules associated with the Web Form component maintained on one system. |
| Aligns with national standards | 5 | FHIR server exchanges information with external parties using FHIR |
| Leverages national services | 5 | Consumes CIS2 via API-M for user and system authentication  Opportunities to integrate with other national services including:   * TRUD (for UTL or test directory represented as SNOMED). * Host Routing for Test Orders (new service). * Consume NHS Digital API Management for other national services which can provide benefit. |
| Supports national Management Information | 5 | Central system audits all Test Orders for Management Information. |
| Service Management | 4 | Central portal and associated capabilities managed to NHS ITIL standards. Simplifies SLA monitoring. |
| **Total Score** | **32** |  |

This option aligns to the concept of the GMS Coordination Platform discussed in the Future Informatics enterprise architecture (see Ref 2) however, the Test Ordering System needs to move from the Patient Care Management Platform to sit alongside Test Order Tracking System – see Figure 5 below. Both capabilities are then satisfied by a single API (and datastore). However, the details of any Test Order can be stored against the Patient’s record within the Patient Care Management Platform.

Additionally, the status updates for send aways and order/sample tracking will need to flow into the Patient Care Management System within the Patient Care Management Platform.



Figure – Futures Architecture Vision Update

### Test Results

Options related to Test Results are discussed within 6.2 which discusses FHIR in more detail.

## Data Architecture

A datastore is central to the architecture. No detailed requirements exist to define the scope of what will be stored in the database, however from an informatics perspective it is clear that details relating to Test Orders (and associated Updates and Test Results) created within the system will need to be stored and also exposed to support operational and management information reporting use cases.

The interfaces exposed to clinical systems which need to connect to the service all mandate FHIR and all information presented by the central API via externally facing services will present any output to FHIR.

This doesn’t mean that FHIR is the only “currency” for the format of the data and there may be internally facing services to support reporting that require more direct access to the data. There may also be a need to support later versions of FHIR or updated FHIR profiles over time. However without a defined set of use cases for data-related capabilities it is difficult to propose suitable solutions. For now, only a general discussion at a high level is possible. The options below relate to “data at rest”, i.e. how the data is stored once it has been received by the system.

### Store as FHIR Documents

The FHIR server exposes API methods which accept HTTP requests as FHIR content and also returns the information as FHIR documents. All content (Test Orders, Results and Workflow resources) is initially created via HTTP POST so all information exchanged with connecting clients can be stored as FHIR CLOBs. As a minimum it would be expected that the FHIR Resources are indexed by a system generated GUID and identifiers such as the Test Order ID and the patient’s NHS number.

### Store FHIR Documents with Key Information Indexed

The Test Orders are stored and indexed as FHIR documents as above, however more of the content is indexed, such as the “contained” Resources such as the Practitioner, Specimen ID(s) and any Organisation references, etc. The indexed attributes will be determined based on use cases.

### Normalised Database

FHIR resources are parsed and the content is stored in a set of core normalised database tables representing each FHIR Resource with relationships/references included as required.

### Options Appraisal – Data Architecture

|  |  |  |
| --- | --- | --- |
| Solution | Benefits | Drawbacks |
| FHIR Documents | Simple to implement. | Meets a limited set of FHIR-centric use cases.  Reporting requirements are likely not to be supported. |
| FHIR Documents with Key Information Indexed | Simple to implement.  Can meet a wider set of use cases beyond FHIR information exchanges. | Use cases need to be understood early in order to avoid rework.  Limited support for reporting requirements. |
| Normalised Database | Normalised data is likely to support reporting capability.  More flexible to handle changes such as FHIR versions or Profile changes (as the data remains the same and the transform is updated as required – MVC pattern). | More complex to implement as all FHIR content needs to be mapped into the database.  The data needs to be transformed to FHIR on the way out when queried by client systems. |

Figure - Data Architecture Options

Use cases need to be understood before any of the options above can be recommended.

### Data retention Policy

Any policy adopted needs to align with existing policies relating to clinical data. As a minimum it should meet the requirements set out in the [NHSX Record Management Code of Practice](https://www.nhsx.nhs.uk/information-governance/guidance/records-management-code/). The closest example to genomics is Cancer/oncology records which states 30 years, or 8 years after death, but unlike cancer genomics related diagnoses aren’t necessarily terminal (obviously some cancers aren’t either).

### Audit and Logging

Requirements need to be defined for this area.

* Messages received by the service (POST, PUT and GET) must be logged.
* Error messages are logged (and these must be indexed to support root cause analysis).
* Audit must include sufficient context to meet IG requirements

### Record Locking

FHIR supports Etag which is the standard HTTP method which supports optimistic record locking. Version IDs (often represented as time stamps) exposed within the FHIR responses must be observed to ensure that client systems have the latest version of the resource before updating any existing FHIR resources.

### Version History

All versions of any record must be maintained in the system, with appropriate audit in place.

## Considerations for FHIR

From a NHS England policy perspective healthcare applications must use FHIR R4 (i.e. use profiles which comply with release 4 of the FHIR specification). The IOPS team at NHS Digital (in collaboration with HL7 UK) maintains a set of FHIR profiles for use in England (and the home countries) known as FHIR UK core. FHIR profiles defined in this release should be adopted where possible.

Early analysis seems to suggest that Genomics can align with the Pathology domain.

Currently there are three sets of pathology FHIR profiles published by IOPS:

* STU3 profiles adopted by the GPIT futures team (but with little live implementation). These are also referenced in the DAPB4017 Pathology standard issued in August 2021.
* “National Pathology” R4 profiles published by the NHS Digital IOPS team. These are separate to the UK Core as a standard (possibly pre-date UK Core) and have no live implementation.
* A subset of the profiles required to support pathology are now available in UK Core.

The STU3 Pathology profiles are to be deprecated and the IOPS team is planning to bring all the FHIR profiles required to support National Pathology profiles within the scope of the UK Core release.

There are two main information flows involved in the Pathology (and Genomics) process - **Test Ordering** and receipt of **Results**.

### Test Ordering

**For Test Ordering** extended versionsof the following FHIR profiles used within the Pathology domain could align to Genomics:

* Patient
* Practitioner
* ServiceRequest
* Organization
* Specimen

The **ServiceRequest** profile is the main FHIR Resource that will be generated to create a genomics Test Order and the other profiles listed are “contained” within the ServiceRequest FHIR Resource – see Figure 7 below which illustrates where the contained resources are referenced. Organization references or Resources can also be referenced at a number of places depending on the context.

Graphical user interface

Description automatically generated with medium confidence

Figure - Pathology ServiceRequest FHIR Profile with Contained Resources

### Test Results

**For Test Results** the pathology standard defines the following profile:

* DiagnosticReport
* Observation

However there are numerous activities in relation to FHIR Test Results which are of interest to the genomics interoperability work. Some effort is required to establish the most appropriate method (or methods) for returning Results. Figure 8 below summarises the current activities.

|  |  |  |
| --- | --- | --- |
| Area | FHIR specifics (Test Results) | Notes |
| National Pathology | * No Live implementation * FHIR R4 profiles | Propose deprecation and extend as required in UK Core. |
| North Thames | * Profiles based on the EMERGE work from the US * FHIR R4 profiles | Need to review the outputs from this work. Also see [EMERGE](https://emerge-fhir-spec.readthedocs.io/en/1.0/artifacts/index.html) referenced in the project documentation for North Thames GMSA. |
| Phenotype profiles | * GA4GH (Global Alliance for Genomics and Health) * FHIR R4 profiles | See [GA4GH](http://phenopackets.org/core-ig/) for further information. Supports rare and infectious diseases currently – status of cancer diagnosis needs to be verified. |
| FHIR UK Core | * Strategic Target for UK wide FHIR implementations * FHIR R4 | Does not include all FHIR profiles required for Genomics (or Pathology). Please see **Figure 9** below for details of what is currently supported.[[2]](#footnote-3)  The current DiagnosticReport and Observation profiles could be extended to support Test Results. |
| HL7 Genomics | * Trial use specification still under development * FHIR R4 | See [FHIR Genomics](https://www.hl7.org/fhir/genomics.html) and [Genomics Reporting](http://hl7.org/fhir/uv/genomics-reporting/index.html) for more information. It’s unclear whether this covers all requirements for genomics reporting. |

Figure - Test Results Methods Summary

Graphical user interface, text, application, email

Description automatically generated

Figure - Current FHIR UK Core Profiles

#### FHIR Considerations for Genomics

##### Use Common Set published within FHIR UK Core

This solution uses a common set of FHIR profiles for Test Ordering and Test results, developed with the IOPS team at NHS Digital. These would be extended from existing National Pathology profiles and then published to the appropriate FHIR UK Core release(s) to be used by both Pathology and Genomics.

|  |  |  |  |
| --- | --- | --- | --- |
| Badge Tick1 with solid fill | Consistent set of products | Badge Cross with solid fill | May be a lot of effort particularly for Test Results |
| Badge Tick1 with solid fill | Under our control | Badge Cross with solid fill | May not align to international activities |
| Badge Tick1 with solid fill | constrains FHIR profile proliferation |  |  |

##### Combine FHIR UK Core with other Genomics Initiatives

Extended versions of the Test Request profiles and publish within FHIR UK Core. Adopt Test Results profiles from EMERGE/North Thames, GA4GH or FHIR Genomics (or combinations as required to meet requirements).

|  |  |  |  |
| --- | --- | --- | --- |
| Badge Tick1 with solid fill | May reduce the work for Test Results. | Badge Cross with solid fill | Inconsistent, with products produced by different teams/bodies. |
| Badge Tick1 with solid fill | Will align to international activities (particularly Test Results). | Badge Cross with solid fill | Partially under our control (Test Ordering). Results governed by international bodies. |
| Badge Tick1 with solid fill | Constrains FHIR profile proliferation. |  |  |

##### Workflow

A light touch workflow which is proposed for communicating Status Updates for Send Aways. This is not domain-specific and the FHIR Workflow profile could be adopted to support this requirement – see: <https://build.fhir.org/workflow.html>.

## Terminology

The Health and Social Care Act published in 2012 requires all clinical systems to use SNOMED to represent clinical information relating to patient’s conditions, treatments and diagnoses. There are a number of challenges in achieving this in both Test Ordering and Test Results as follows:

* Alignment of Genomics Test Orders to SNOMED.
  + This may involve re-classification of the tests defined in the Genomics Test Directory to support interoperability (see document reference 5)
* Analysis of the clinical content required for the results.
  + What is required over and above the human readable summary and what is the variation in structure for the different types of test?
  + What is required to be stored against the EPR?
* Potential mapping of existing genomics terminologies (MONDO, HPO and LOINC (HGVS)) to SNOMED (depending on the amount of detail required).
* How much of the information that needs to be communicated for genomics is already coded in SNOMED?

The adoption of SNOMED for Test Orders is more controllable as the system greenfield is being specified and delivered by NHS England.

To accelerate the adoption of SNOMED for the Test Results the LIMS could continue to use existing terminologies and transform/map to SNOMED at the point at which the Results are sent to the FHIR API, i.e. embed the capability within the adaptors.

The mapping work could follow the Genomics pathways agile approach and the SNOMED terms would then build up over time.

## Information Exchange Patterns

The FHIR standard focuses on the structured content within a message to represent events within healthcare. The actual transmission of the FHIR content depends on the implementation and is driven by the capabilities of the organisations and systems involved The appropriate methods adopted are governed by the message pattern (sync/async), as well as the capabilities of the organisations and systems involved in the exchanges. Figure 10 summarises functional capabilities against potential messaging patterns and messages. This is heavily caveated as the actual system capabilities and requirements are not yet known. The methods are summarised below.

|  |  |  |
| --- | --- | --- |
| Functional Capability | Pattern | Methods for Consideration |
| Test Order | Synchronous | RESTful API |
| Status Updates (inbound to central API service) | Asynchronous | MESH NEMS FHIR tasks profile |
| Status updates (inbound to ordering system) | Synchronous, or  Asynchronous | RESTful call (synchronous if polling for updates)  NEMS Pub/Sub notification |
| Results Reporting (inbound to central API service) | Asynchronous | MESH NEMS FHIR tasks profile ADT (FHIR) |
| Results Reporting (inbound to ordering system) | Synchronous, or  Asynchronous | RESTful call (synchronous if polling for updates)  NEMS Pub/Sub notification |

Figure - Candidate message patterns for further discussion

### RESTful API

This method supports calls to a FHIR server to create, update and retrieve FHIR resources (provided by a tactical Adaptor where required).

* Most common and preferred method
* Mature

### Delivery via MESH mailboxes

MESH (Messaging for Social Care and Health) is a national service provided by NHS Digital to support information exchange between systems.

* Supports asynchronous message patterns
* MESH is mature across the NHS
* Some transmission of FHIR documents exists

### ADT (admissions, discharge, transfer of care) messaging

The payload (containing the FHIR document) is wrapped in ITK FHIR using FHIR messageHeader extensions to denote sender and receiver.

* Some implementations use FHIR (preferred)
* The pattern has been adopted for ADT, previously for HL7v3

### NEMS Publish and Subscribe Pattern

NEMS (National Event Management Service - see <https://digital.nhs.uk/services/national-events-management-service>) uses a publish and subscribe pattern, whereby subscribers can request updates to certain events. Events are published by systems and subscribers receive information on certain events, e.g. for Genomics this could be a status update on a Test Order or the Test Results being available.

It’s current use cases which NEMS supports include updates to patient demographics and the National record Locator (<https://digital.nhs.uk/services/national-record-locator>). Currently the service leverages MESH mailboxes to make information available to subscribers. An appropriate asynchronous message pattern is required which could be MESH mailboxes (NEMS supports this) or a Service hosted by an API service to receive inbound events.

### FHIR Task Profile

FHIR R4 includes a task resource/profile which supports workflow and event management. This method doesn’t have any live deployments; however it may be a good fit for the Genomics work.

### OpenEHR

See <https://www.openehr.org/>). OpenEHR is a technology for e-health consisting of open platform specifications, clinical models and software IHE is an enterprise-wide approach to healthcare informatics

* Unlikely to be supported by LIMS or PAS/EPR systems currently
* Requires further investigation to determine implementation maturity

## National Integration Patterns

Historically national messaging has been implemented across Spine and has been conceived as complex and expensive. However in recent years the bar for integration has been lowered significantly with the introduction of internet facing capabilities which hide the complexity of message exchanges. This has been driven by initiatives such as GP Connect which opens access to patient data in GP systems.

National messaging capabilities include:

1. FHIR messaging
2. MESH transport
3. ITK used for ADT
4. NEMS Publish and subscribe
5. HL7v3 messaging (out of scope - for legacy implementations only)

Items 1 to 4 are listed as options in the Information Exchange section above. Many of the target architecture options proposed in this document require support for FHIR components deployed to the Trust/LIMS/GLH/NPEx/NGIS infrastructure (as shown in the excerpt in Figure 11 below). In most cases the components representing the FHIR Server and the Message Gateway will be a single component. Existing PAS/EPR systems will require configuration to support Genomics messages.

Diagram

Description automatically generated Chart

Description automatically generated with medium confidence

Figure - Excerpt, FHIR Messaging Capability

Further work is required to define the topologies required within Genomics, which should then be reviewed against the patterns defined in the Integration Patterns Book published by NHS Digital at: <https://digital.nhs.uk/about-nhs-digital/our-work/nhs-digital-architecture>. A candidate pattern that is likely to align is an intermediary API pattern which is currently used for patient referrals.

## Genomics Test Routing

Currently each GLH manages local reference data based on the type of Test that is being ordered and the Trust ordering the test. Within these two axes there is a lab specified for processing the test and a lab responsible for reporting the results. Figure 12 below shows an example of how this is structured. This is based on information shared by North East and Yorkshire GLH but has pseudo codes substituted).

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| ***Test Order*** | **ODS Trust A** | | **ODS Trust B** | | **ODS Trust C** | | **ODS Trust D** | |
| Process | Report | Process | Report | Process | Report | Process | Report |
| *Test Type 1* | ODS1 | ODS15 | ODS29 | ODS43 | ODS57 | ODS71 | ODS85 | ODS99 |
| *Test Type 2* | ODS2 | ODS16 | ODS30 | ODS44 | ODS58 | ODS72 | ODS86 | ODS100 |
| *Test Type 3* | ODS3 | ODS17 | ODS31 | ODS45 | ODS59 | ODS73 | ODS87 | ODS101 |
| *Test Type 4* | ODS4 | ODS18 | ODS32 | ODS46 | ODS60 | ODS74 | ODS88 | ODS102 |
| *Test Type 5* | ODS5 | ODS19 | ODS33 | ODS45 | ODS61 | ODS75 | ODS89 | ODS46 |
| *Test Type 6* | ODS6 | ODS15 | ODS24 | ODS46 | ODS62 | ODS76 | ODS90 | ODS45 |
| *Test Type 7* | ODS7 | ODS11 | ODS15 | ODS49 | ODS63 | ODS77 | ODS91 | ODS17 |
| *Test Type 8* | ODS8 | ODS7 | ODS6 | ODS5 | ODS64 | ODS78 | ODS45 | ODS18 |
| *Test Type 9* | ODS29 | ODS3 | ODS23 | ODS49 | ODS75 | ODS101 | ODS127 | ODS153 |

Figure - Indicative example of Test Routing table

This is a very good reference and provides a good basis for expanding routing as a concept to be managed at a national level, however it seems that the different GLHs hold the information in different ways and the data may support other use cases. This requires further analysis and identification of the key requirements to support Test Routing as a capability.

Points for further discussion:

1. Establish a national database which national Genomics API(s) can use to support routing decisions
2. The structured information MUST use ODS codes[[3]](#footnote-4)

Beyond MVP for Test Routing consider expansion to support capacity management, service availability and Management Information to inform SLAs.

1. **NHSE Interoperability Review**

Summary of discussion with Jonathan Telfer (Interoperability and Standards lead at NHS England) – 17/05/2022.

Generally, we seem to be doing the right thing, particularly from the standards perspective (SNOMED, FHIR and the pathology UTL).  See consultation:  <https://facultyofclinicalinformatics.org.uk/blog/faculty-of-clinical-informatics-news-1/post/how-standards-will-support-interoperability-90>

Some of the comments items raised have already been incorporated into the document.

**Adaptor**

The interop strategy is not to support translators between different standards “at a national level” however the adaptors we are proposing will be deployed locally to each LIMS (where required).

Jonathan’s view was that adaptors (in general) give suppliers a bye in terms of us doing the work for them, but coupled with a retirement strategy our proposal should be fine.

An adaptor is classed as a medical device and attracts a high level of attention for clinical safety.

We need to work on this early on in the process to ensure all supplier concerns are addressed.

Options for providing:

1. NHS Digital builds and supports:

We need to be clear that this will be supported for a limited period, after which the suppliers can maintain their own code base (code is open source).

This also applies to older products that will never be able to support FHIR natively.

1. We provide mappings/specifications and let the market plug the gap with solutions (could be innovators or the Trusts configuring their own integration engines).

For option 1, thinking about suppliers maintaining their own code base, we need to survey the supplier base and work out which is the most commonly used application programming language in use.  We would only consider building the adaptor using a modern programming language such as Python, Java or .NET.  Other languages such as Delphi, Visual Basic and less common languages will not be considered.

**Workflow for send aways and results**

We need to look at the options for this (FHIR workflow/existing events monitoring such as NEMS) and the method of transfer (asynchronous pub/sub, MESH, etc…).  Basically patterns and Jonathan talked about authoritative  sources for these, which I think is identifying the organisation/system that provided the update.  We need to identify the points in the process flow where we would want supplier systems to ping an event update back to the centre.

**LIMS Re-procurement**

We need to engage with the pathology team and work out where this is.  If possible get some requirements built in to the specification to support the patterns which genomics and pathology require.

**Centralised Architecture**

Generally the strategy will not support centralised technology unless there are tangible benefits.  We think that the MI requirement, the standard processing of orders and the event management for sendaways and results justifies this.

Jonathan mentioned that diagnostic imaging has a centralised broker for event handling/monitoring on their strategy, so we could bring their requirements into scope.  We know that regular pathology has a desire for this too.

**Web Portal**

The web portal component for ordering the tests (to support organisations whose supplier doesn’t integrate with the FHIR API) could potentially be provided by innovators who then sell their products to Trusts and CCGs.  This should be feasible particularly if we make the Genomics Reference Set (AKA new TD) available via an internet facing service/API.

**FHIR Profile Proliferation**

This is basically each system creating a modified FHIR profile to support its own very local requirements.  We should avoid this if possible and modify the UK Core FHIR profiles with optional content for Genomics (and possibly diagnostic imaging??).

Currently there is a gap in the architecture for Patient Case Management, which will be addressed via the current consultation.  Anne Crowther has a logical component for this, but other than the results finding their way back in to that we don’t have a lot of overlap from an interop perspective.

1. There is a scenario whereby the GLH can modify the incoming test Order and substitute different tests. The behaviour of this interaction needs to be carefully manged – amended record vs reject and initiate a new order. [↑](#footnote-ref-2)
2. The IOPS team at NHS Digital maintains a backlog of activities relating to future profiles for inclusion within FHIR UK Core. DiagnosticReport and Observation profiles are not yet in the release but are available in draft. [↑](#footnote-ref-3)
3. ODS – Organisation Data Service is a central team responsible for maintaining organisation codes and reference data to support Health and Social Care. [↑](#footnote-ref-4)