High Level Design

Genomics Testing - Target Architecture

Document name:		Options Appraisal – Genomics Target Architecture
Programme/Project Name		Digital Interoperability
Project Manager		Jon Calpin
Author		Ravi Natarajan
Version	0.7	Date: 27/07/2022

Document management

Revision history

Version	Date	Summary of changes
0.1	30/05/2022	Initial draft based on future Informatics Team's review of Target Architecture Options Appraisal version 0.7.
0.2	31/05/2022	Updated version following internal review.
0.3	01/06/2022	Further edits to sections
0.4	06/06/2022	Architecture figure changes and major edits
0.5	09/06/2022	Updated addition figures and changes to sections
0.6	04/07/2022	Updates based on Jonathan Telfer feedback and further clean up
0.7	27/07/2022	Changes to sections to replace FHIR API aspects with FHIR Messaging

Approved by

This document must be approved by the following people:

Name	Signature	Title	Date	Version
Jon Caplin		Project Manager		

Related documents

Ref	Title	Owner	Location
1	Process Architecture Analysis		TBC
2	Genomics Testing Pathways Tube Matrix		Tube Matrix
3	Future Informatics Enterprise Architecture artefacts		TBC
4	Genomics Target Architecture Options Appraisal v0.7		Options Appraisal
5	GTODS Requirements Catalogue v0.5		GTODS

6	GMS Interoperability Programme Scope	Harini Nallapothola	

Document control

The controlled copy of this document is maintained by NHS England and NHS Improvement. Any copies of this document held outside of that area, in whatever format (e.g. paper, email attachment), are considered to have passed out of control and should be checked for currency and validity.

contents

Glossary

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. security-and-authorisation-user-restricted-apis
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.
FHIR UK Core	FHIR UK Core	HL7 FHIR® UK Core R4
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)
WGS	Whole Genome Sequencing	Whole-genome sequencing (WGS) is a comprehensive method for analysing entire genomes of patient or organism
IOP	Interoperability	Interoperability is the ability of diverse systems and organisations to work together seamlessly by exchanging information and using the information that has been exchanged.
Home GLH	Home Genomic Laboratory Hub	The national genomic testing service is delivered through a network of seven Genomic Laboratory Hubs (GLHs), each responsible for coordinating services for a particular part of the country.

1. Purpose

The purpose of this document is to capture 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 solution architecture, data architecture and patterns of message exchange; however, there may be some short-term solutions that act as enablers to meet the recommended option.

2. Problem Statements and Requirements

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

2.1. 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.

2.2. 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.

2.3. 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 aligned 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).

2.4. Non-Functional Requirements

The following requirements are a combination of strategic aims and other activities within NHS E&I:

2.4.1. 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.

2.4.2. 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 with FHIR wherever possible.

2.4.3. Scalability and Volume

The recommended solution must be capable of scaling to meet expected volumes for upcoming needs. For example there is an initiative to undertake WGS for all new-borns.

2.4.4. Strategic Aim for Central Portal/Repository

NHS E&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.

2.4.5. 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.

The sections expressed are generic and a much more detailed requirements gathering exercise would be undertaken to cover this area more in detail.

3. Scope

The scope of the Target Architecture includes Test Ordering flow, Status Updates (with Home GLH) and Test Results. Ref 6 captures the top levels in and out of scope as part of the technical architecture delivery.

a. In Scope

1. Bi-directional interface between requestors and Home GLH (Genomic Laboratory Hub)using:
    a. Portal (Time limited and would be sunset after 2 years post go live)
    b. FHIR Messaging - It is expected that the LIMS will have the capability to receive genomics Test Orders using nationally approved information standards (FHIR R4)
2. Status updates with Home GLH
   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.
3. Test results covering exchange of documents (pdf’s) and structured reports.
4. 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.

5. Central Genomic Test Repository
   A secure repository to capture requests and results for access by various stakeholders as appropriate.
6. Authorised report access via web portal and secure delivery to initial test requestors via appropriate methods (email, messaging etc).
   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.
7. Management Reporting around testing requests

b. Out Of Scope

1. Bioinformatics around DNA sequencing data analysis, inference and final report.
2. 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.

   The GTOD is a dataset which describes the Tests in sufficient detail for the receiving laboratory to understand what assay(s) and analysis(-es) it is being asked to perform. 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.
3. Interoperability for send aways between home GLH and subsequent labs (i.e. wet, dry etc)

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.

4. Interoperability between home GLH - Any subsequent communication between the GLH and downstream systems eg Wet and Dry Labs, other GLH systems) is out of scope. This includes decision support systems (Congenica, BSVI, Illumina). This also includes communication of test requests between different GLHS.

5. 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.

6. Distribution of test reports beyond initiating requestor.

7. Automated data flows into Patient Level Contract Monitoring systems (PLCM).

8. Integration into requesting systems to allow the testing report (pdf's or structured) embedded into their clinical systems is beyond the scope of this work.

4. Target Architecture

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

In the options appraisal, the central solution scored high in most areas and is presented below. All other options that were evaluated are available in Ref 4.

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 ordering 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:
		  o Direct EPR integration to FHIR API
		  o iFrame silent/cached login to Webform
		  o Separate login to Web Form
		  o Locally built Web Form integration
		• Medium component deployment count.
		• Rules associated with the Webform 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 with the concept of the GMS Coordination Platform discussed in the Future Informatics enterprise architecture (see Ref 2). The Test Ordering System needs to integrate with the proposed platform to enable the capabilities around a single API (and datastore).

The details of any genomic test order can be stored against the Patient's record within the Patient Care Management Platform aided by integration to the central solution.

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.

The architectural view of the proposed solution is shown below in Fig.1

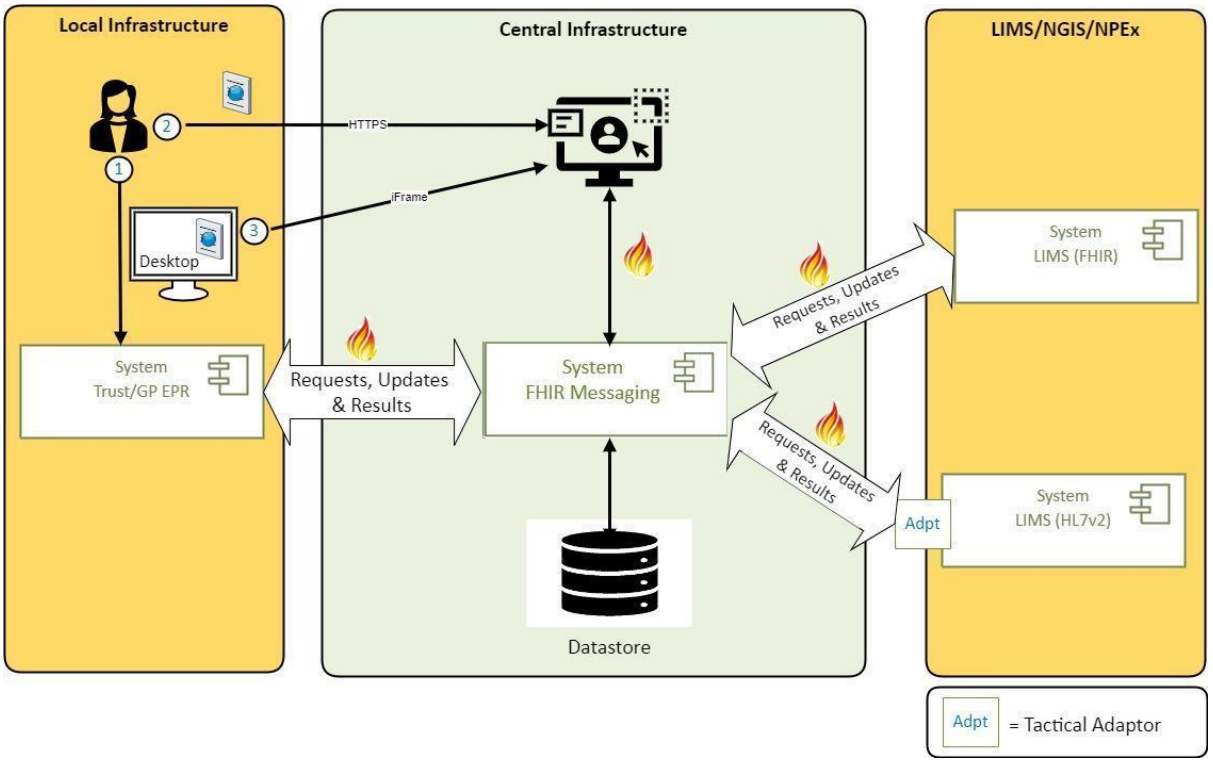

Figure 1 – Test Order, Data Capture Centralised – integration options 1 to 3

Webportal with dynamic forms and FHIR messaging components are hosted centrally on a single logical system acting as a message broker, with multiple options for integration to the Test Order system for Trusts, in the order of preference and digital maturity.

1. Integrates directly to the Test request via single FHIR messaging endpoint.
2. Direct access to the web portal/application hosted centrally.
3. Web page iFrame integration within a web based EPR, supporting transparent login/session management and click through of patient details from the EPR. **Note : This is the least favoured option due the security concerns around iFrame integration and browser compatibility issues.**

Integration options 1 to 3 are shown on Figure 2 above and the detailed architecture is shown on Figure 3 further below.

A 4th option exists wherein the trust uses a combination of the above 3 to achieve the required interoperability.

Users and systems authenticate using OAuth2 and CIS2 for RBAC. LIMS systems (Home GLH) integrate with a single endpoint 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.

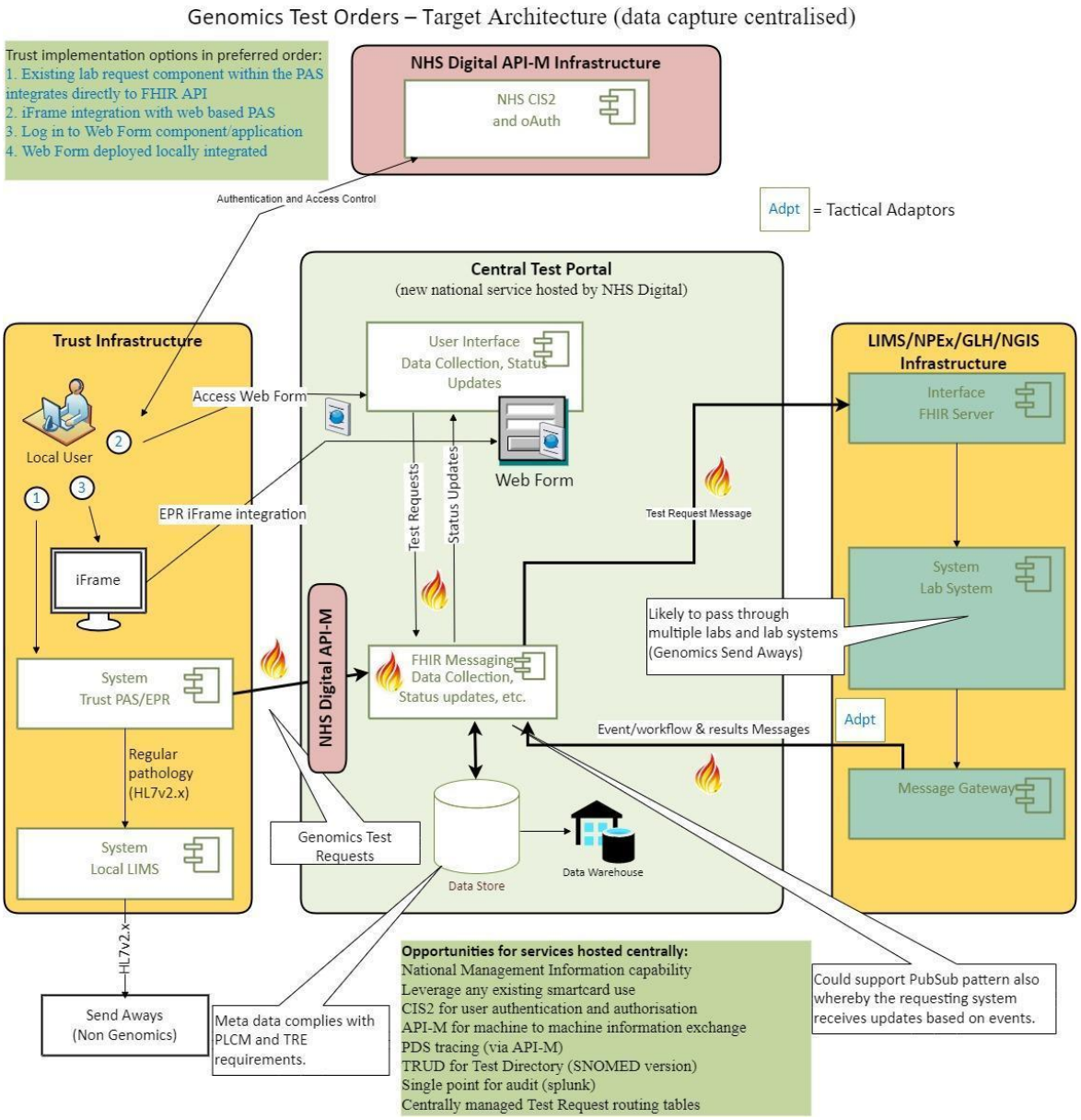

Figure 2 - Detailed Interaction Diagram

4.1. 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.

4.1.1. Web Portal

This is a web application, its primary function is 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 portal 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. Publish a specification against which local implementations are assured and allow local Trusts to re-use any existing 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 messaging component.

The web 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 in 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.

4.1.2. Test Order FHIR Messaging

This is a Test Ordering component which exposes a single FHIR messaging endpoint 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 FHIR messaging API's key functionality is listed below:

- Accepts and validates a Genomics Test Order.
- Supports amendments to Test Orders. [1]
- Manages access to Test Order records.
- Test reports access in pdf's and structured content via messaging.
- 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 in later phases.
- 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 portal into the appropriate FHIR document.
- Secure management access to datastore.
- Delivers the validated FHIR test result document to the initial recipient using the appropriate method.
- Supports Publish and subscribe capability if this provides benefit.
- Implemented as a synchronous pattern supporting REST architecture.

4.1.3. Adaptor

The target architecture fundamental requirement requires the LIMS receiving the initial Test Order, to support 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

[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 managed – amended record vs reject and initiate a new order.

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).

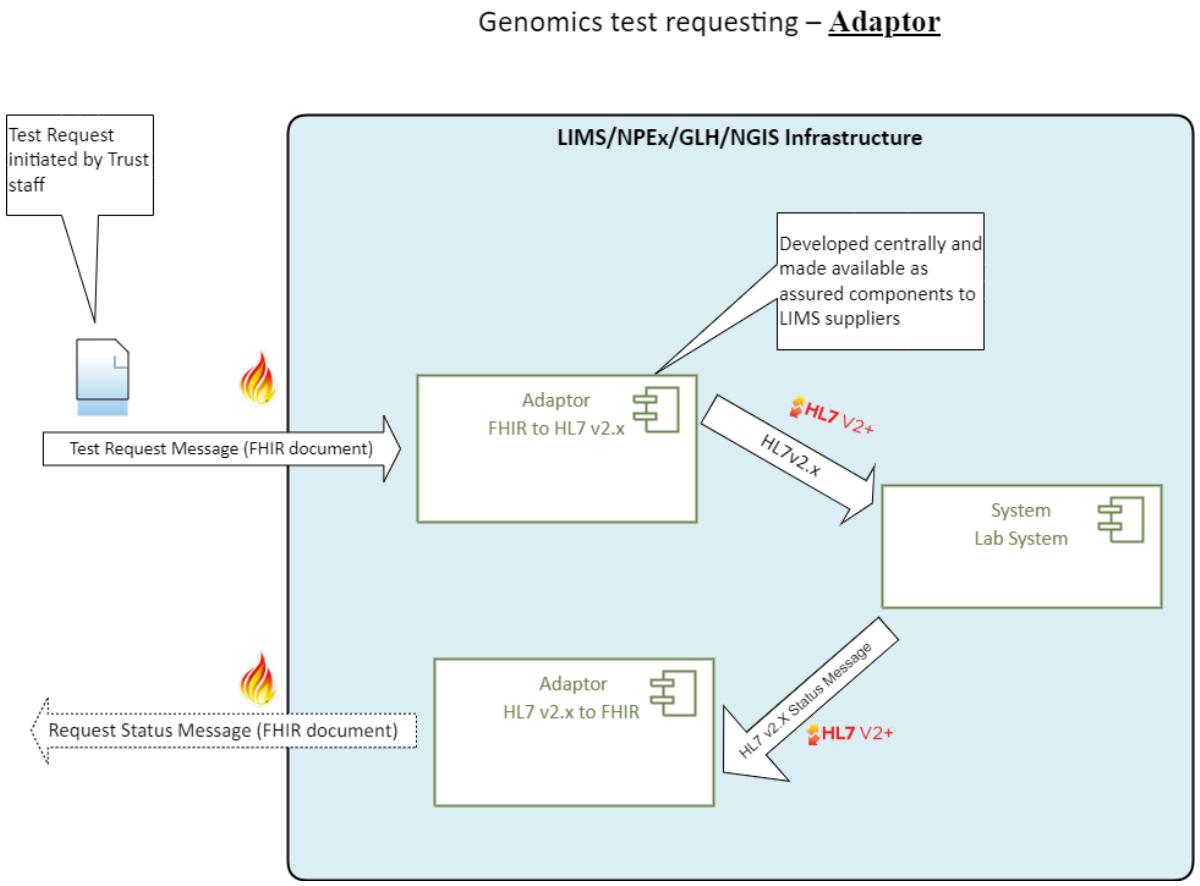

Figure 3 - Tactical Adaptors

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.

4.1.4. Lab System Component

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

4.1.5. 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.

5. Data Architecture

A genomic test request response 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 mandates FHIR and all information presented by the central API via externally facing services will present any output in FHIR format.

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.

5.1. 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.

5.2. 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.

5.3. 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.

5.4. 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 4 - Data Architecture Options

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

5.5. 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 NHS Record Management Code of Practice. 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).

5.6. 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.

5.7. 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.

5.8. Version History

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

6. 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.

6.1. Test Ordering

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

- Patient
- FamilyMemberHistory* (TBC)
- Practitioner
- PractitionerRole
- ServiceRequest
- Organisation
- Specimen
- Consent
- Observation
- QuestionnaireResponse

* - This resource is only available in the HL7 international FHIR specification and needs to be incorporated into the UK Core specification.

The ServiceRequest profile is the main FHIR Resource that will be generated to create a genomics Test Order and the other profiles listed are "referenced" within the ServiceRequest FHIR Resource – see Figure 5 below which illustrates where the contained resources are referenced. Organisation references or other Resources can also be referenced at a number of places depending on the context.

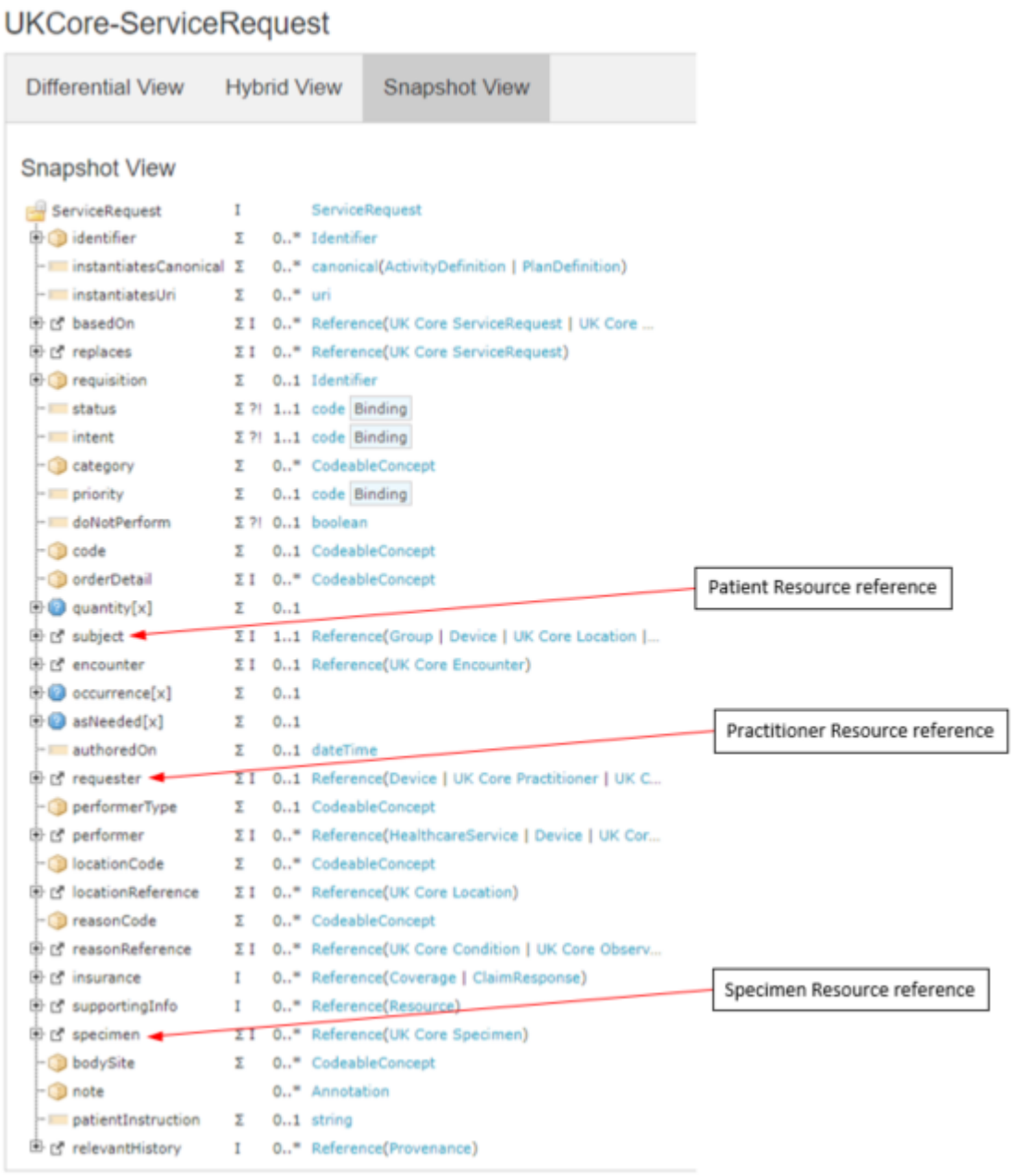

Figure 5 - Pathology ServiceRequest FHIR Profile with Contained Resources

6.2. Test Results

For Test Results the pathology standard defines the following profile:

- DiagnosticReport
- Observation
- MolecularSequence

- This profile managed in the HL7 international diagnostic reporting domain is to describe an atomic sequence which contains the alignment sequencing test result and multiple variations.

However there are numerous activities in relation to FHIR Test Results which are of interest to the genomics interoperability work.

- Genomics
- Genomics Reporting

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 STU1 profiles	Need to review the outputs from this work. Also see eMERGE referenced in the project documentation for North Thames GMSA. Note : This work was implemented using STU1 profiles. Any consideration of this work needs to accommodate the recent FHIR standard which is R4.
Phenotype profiles	• GA4GH (Global Alliance for Genomics and Health) • FHIR R4 profiles	See GA4GH 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] The DiagnosticReport and Observation profiles could be extended to support Test Results.
HL7 Genomics	• Trial use specification still under development • FHIR R4	See FHIR Genomics and Genomics Reporting for more information. It's unclear whether this covers all requirements for genomics reporting.

Figure 6 - Test Results Methods Summary

[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.

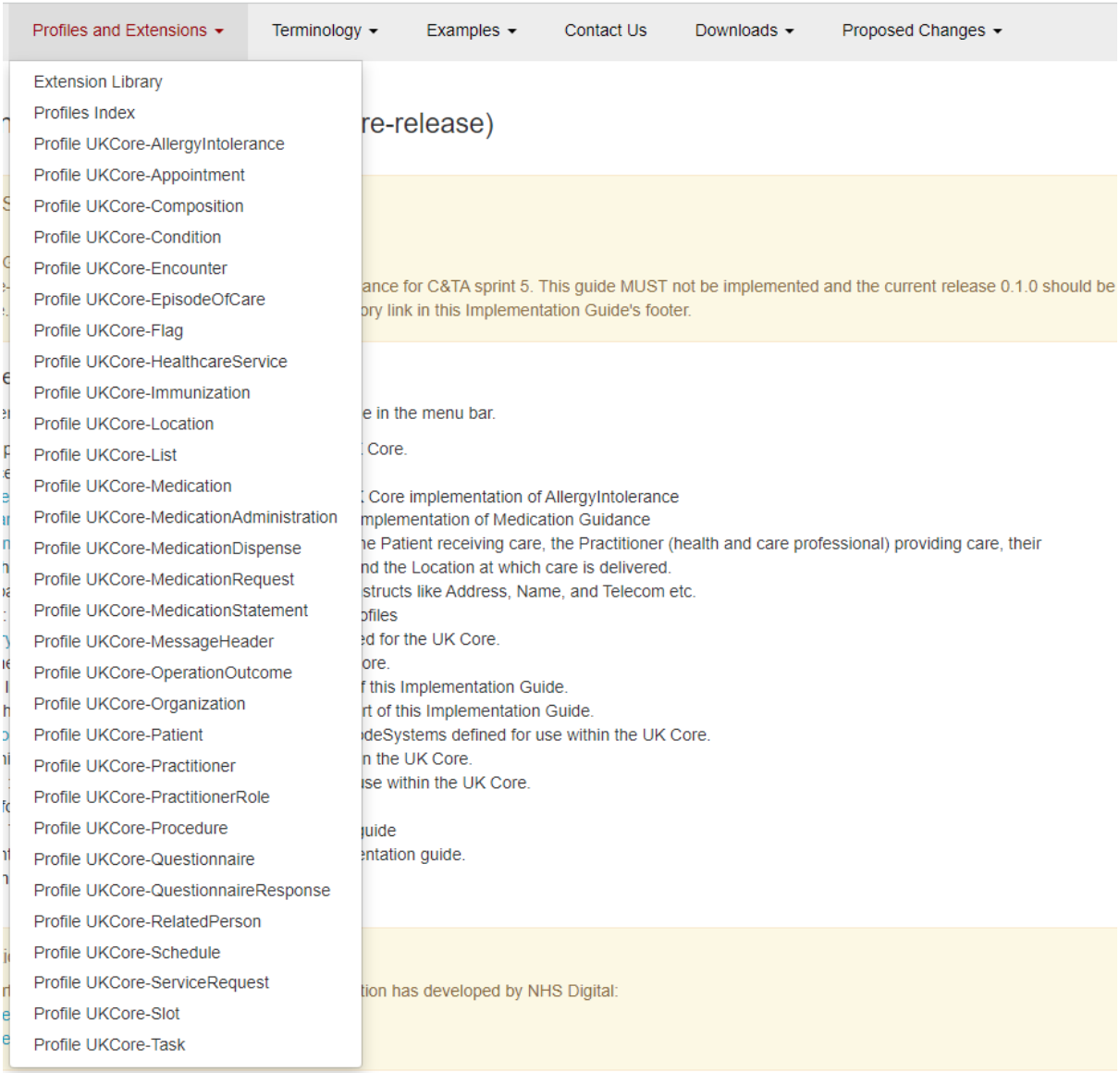

Figure 7 - Current FHIR UK Core Profiles

6.3. Considerations for Genomics

6.3.1. 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.

✓	**Consistent set of products**	✗	**May be a lot of effort particularly for Test Results**

✓	Under our control	✗	May not align to international activities
✓	constrains FHIR profile proliferation		

6.3.2. 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).

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

6.3.3. Workflow

A light touch workflow is proposed for communicating Status Updates for Send Aways. This is not domain-specific and the FHIR task profile could be adopted to support this requirement – see: https://build.fhir.org/workflow.html. This method doesn't have any live deployments; however it may be a good fit for the Genomics work.

7. Terminology

The Health and Social Care Act published in 2012 requires all clinical systems to use SNOMED to represent clinical information relating to a patient's conditions, treatments and diagnosis. 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). There's a historical work that resulted in creation of content specific to WGS tests with being grouped together under a simple refset within the UK extension. The refset is titled "**National Health Service England National Genomic Test Directory whole genome sequencing test simple reference set (foundation**

metadata concept) - 71681000000103". Any work needs to consider refreshing the refset with associated changes to content i.e. retired tests, adding new tests etc.

- Analysis of the clinical content required for the Test 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 is being specified and delivered by NHS England.

To accelerate the adoption of SNOMED for the Test Results ,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.

8. 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 9 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	RESTful call (synchronous if polling for updates)

	Asynchronous	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 9 - Candidate message patterns for further discussion

8.1. 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

8.2. 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

8.3. 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

8.4. 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.

Its current use cases which NEMS supports include updates to patient demographics, new born events, immunisation 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.

c.

9. 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 9 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.

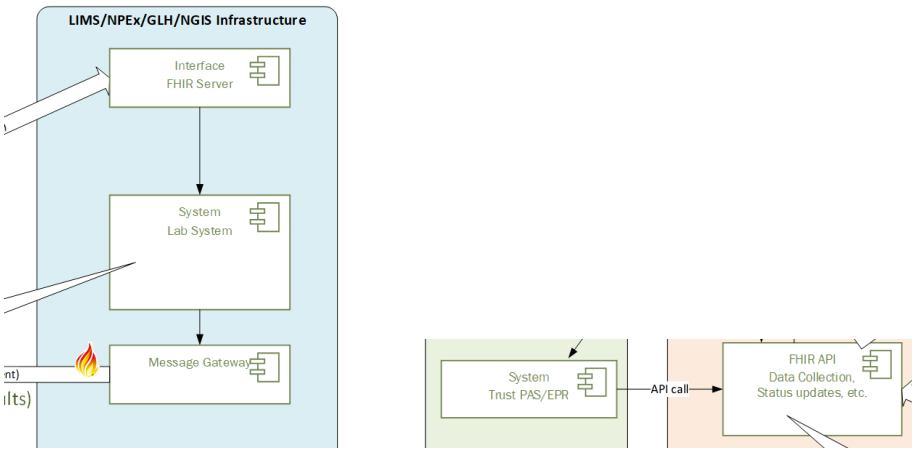

Figure 9 - Excerpt, FHIR Messaging Capability

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.

10. Genomics Test Routing

Currently each home 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 11 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 11 - 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:

2. Establish a national database which national Genomics API(s) can use to support routing decisions
3. Try to adapt rerouting patterns implemented for GP Connect and other national information systems like NEMS.
4. The structured information MUST use ODS codes[3]

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

[3] ODS – Organisation Data Service is a central team responsible for maintaining organisation codes and reference data to support Health and Social Care.