Connectathon Brief

December 18th, 2024

Genomic Order Management Introduction

The scope of the core service is the digitisation of genomics test ordering within the whole of NHS-E, focusing solely on test order management (WGS and Non-WGS Test Requests) and the management of test results and reports. Key points in scope:

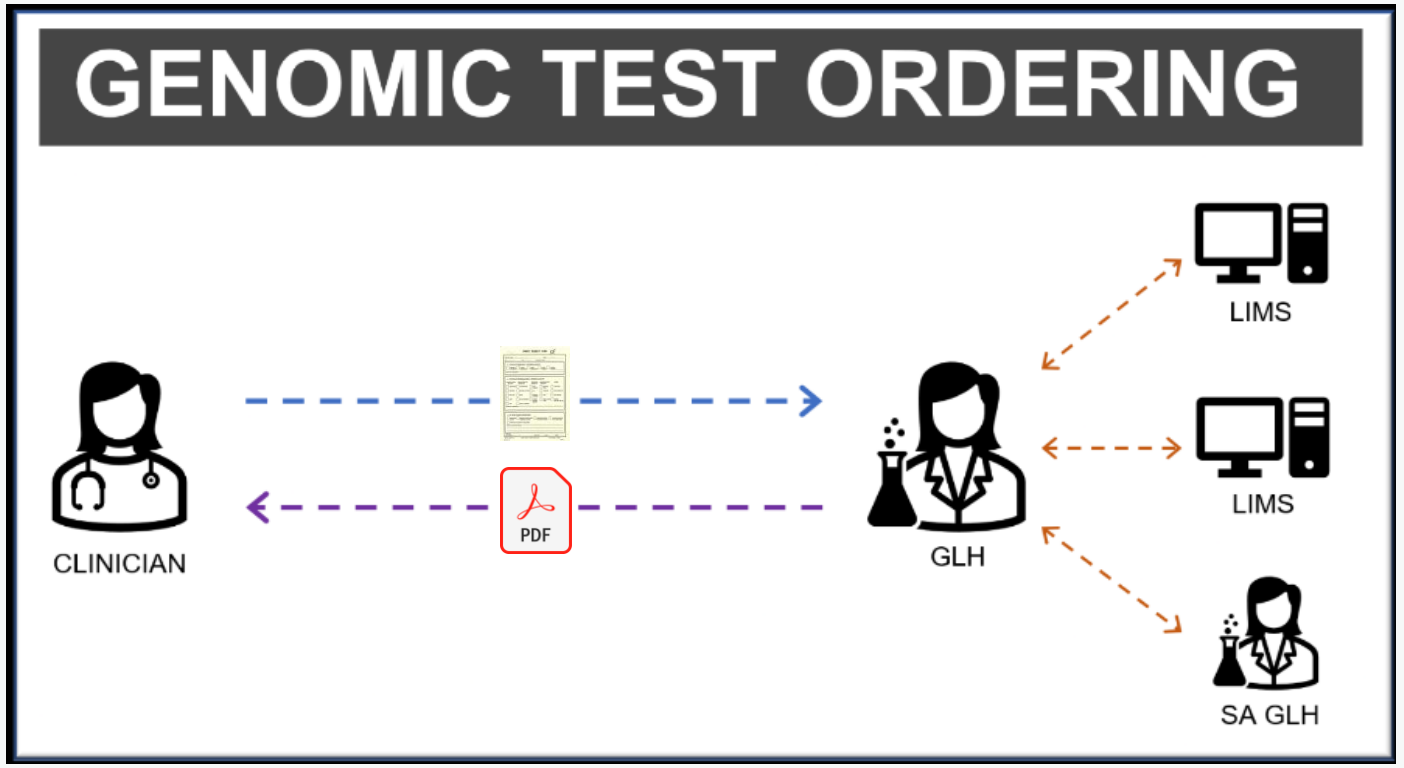
- it is a central national capability, which will standardise the data associated with each genomic test request and communication standards
- it is focusing on the relevant data flows between organisational boundaries
- with the end strategic goal being that a significant proportion of requesting locations have their systems (e.g., EHRs, Portals, LIMS, Order Comms) and receiving locations (e.g., Lab LIMS) integrated to central national capability

Benefits of the project include:

- Aligning the Genomic Test Request process with the Genomics strategy , Data and Digital Framework and GMS Architecture Vision
- Core foundation to the Genomics Service and operational data
- Reduction of transcribing the same test request, freeing resources and improving clinical safety
- Improving information around test requests and sample statuses

Problem Statement

- The existing process is predominantly **paper** driven.
- **Manual** entries leading to errors.
- **Consent management** is difficult.
- **No visibility** of status of the orders and samples.
- **Limited adherence** to national standards.
- **Lack of management information** across the pathways.
- **Coordination** of send aways is complicated.
- **Scalability** to accelerate the genomics services is difficult.
- **Alignment** to Accelerating GMS Strategy is restricted.



Connectathon

NHS England

Connectathon

- **Purpose**
  - To demonstrate digital interoperability capabilities of the Genomic Order Management API and its utilisation by NHS system providers.
  - Unique testing opportunity (Similar to IHE event baring the certification)
  - Gain feedback about
    - Robustness of the API's
    - Improvements needed to the Genomics FHIR IG specification via simplifier.
  - Understand the system vendors technical challenges
    - Help in shaping the next stages around private beta, beta and live
  - Improve visibility of the GU digital initiatives
  - Not a **procurement activity**

Conectathon - Supplier Prerequisites

- Extensive healthcare experience
- Understanding of NHS policies , procedures and standards etc
- Experience in digital systems deployment within NHS
- Hands on development team who can
  - Undertake rapid development and deployment
  - Restful API's integration
    - JSON specification
  - HL7 FHIR / V2, Terminology , Code systems and Valuesets etc.
- Understand the needs of the application role demonstrated (Consumer, Provider ...)
- Pre-Onboarded to APIM for Genomic Order Management API
  - API key allocated – See Video Enclosed
- **BYOD** (Bring your own device) which can connect to Genomic Order Management API over internet.
- **BYOTS** (Bring your own Test System) - A robust supplier test system pre-configured with accounts with features to demonstrate quick configuration, code deployments etc

Scenarios for Connectathon

Scenario 1: Generate FHIR- based genomic test request (Non WGS) on a singleton (single)patient with a single sample required and submit test request to the core broker. Perform validation as well as measure commonality and ingest of each FHIR bundle created. Query broker for test request/sample status update. If status marked as complete, query final report from broker

Scenario 2: Generate FHIR-based genomic WGS test request, sample required and Record of Discussion and submit test request to the core broker. Perform validation as well as measure commonality and ingest of each FHIR bundle created. Query broker for test request/sample status update. If status marked as complete, query final report from broker

Scenario 3: Generate FHIR-based Non WGS fetal test request, indicate additional samples required (mom+dad) and submit test request to the core broker. Perform validation as well as measure commonality and ingest of each FHIR bundle created. Query broker for test request/sample status update. If status marked as complete, query final report from broker

Scenarios Links

Scenario 1: Generate FHIR- based genomic test request (Non WGS) on a singleton (single) patient with a single sample required and submit test request to the core broker.

- Non WGS Singleton Test Request

Scenario 2: Generate FHIR-based genomic WGS test request, sample required and Record of Discussion and submit test request to the core broker

- WGS Test Scenario

Scenario 3: Generate FHIR-based Non WGS fetal test request, indicate additional samples required (mum+dad) and submit test request to the core broker

- Fetus Management

Pre- conditions (All Scenarios)

- EPR/Test Order Portal Suppliers/ Middleware/ LIMS suppliers: API connection to the core broker (INT environment).
- API Int Endpoint: https://int.api.service.nhs.uk/genomic-order-management-service
- Configured with the structured definitions within Genomics IG: FHIR Genomics Implementation Guide

Dependencies

- Digital Test Directory - API
  - To enable validation of inbound tests for their accuracy
  - To provide routing information for the tests ordered
- National Services
  - PDS – To gather demographic information for patient
  - ODS – To identify organisation information with NHS

Core Components

- **Core Broker** – Receive, Manage and Deliver digital communication for genomic orders across NHSE
- **Genomic Testing Consumer** – Manage Genomic test requests, sample information based on interoperability standards, data sets and integrate to Core Broker.
  - EHR
  - LIMS
  - Web Portal ...
- **Genomic Testing Provider** – Deliver testing services for genomic orders and integrate into core broker
  - LIMS

Success Criteria

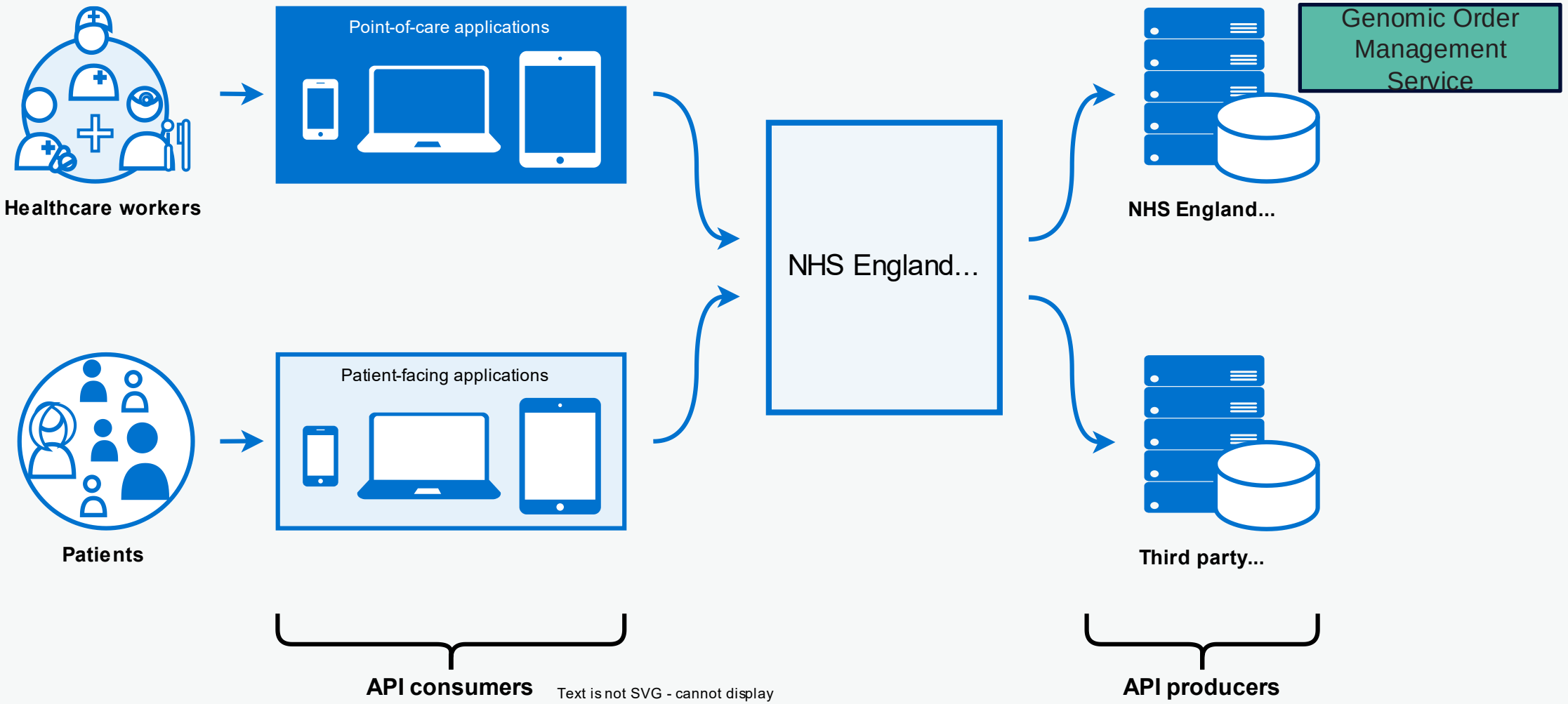
- EPR/Test Order Portal/ LIMS actors will successfully submit Non WGS & WGS Test Request FHIR resource to the central broker. Example will correspond to the single patient data example provided

- EPR/Test Order Portal/ LIMS actors will successfully validate on test and sample status updates. If test marked as complete, actors to query the broker for final report (PDF) and display report natively within their clinical application

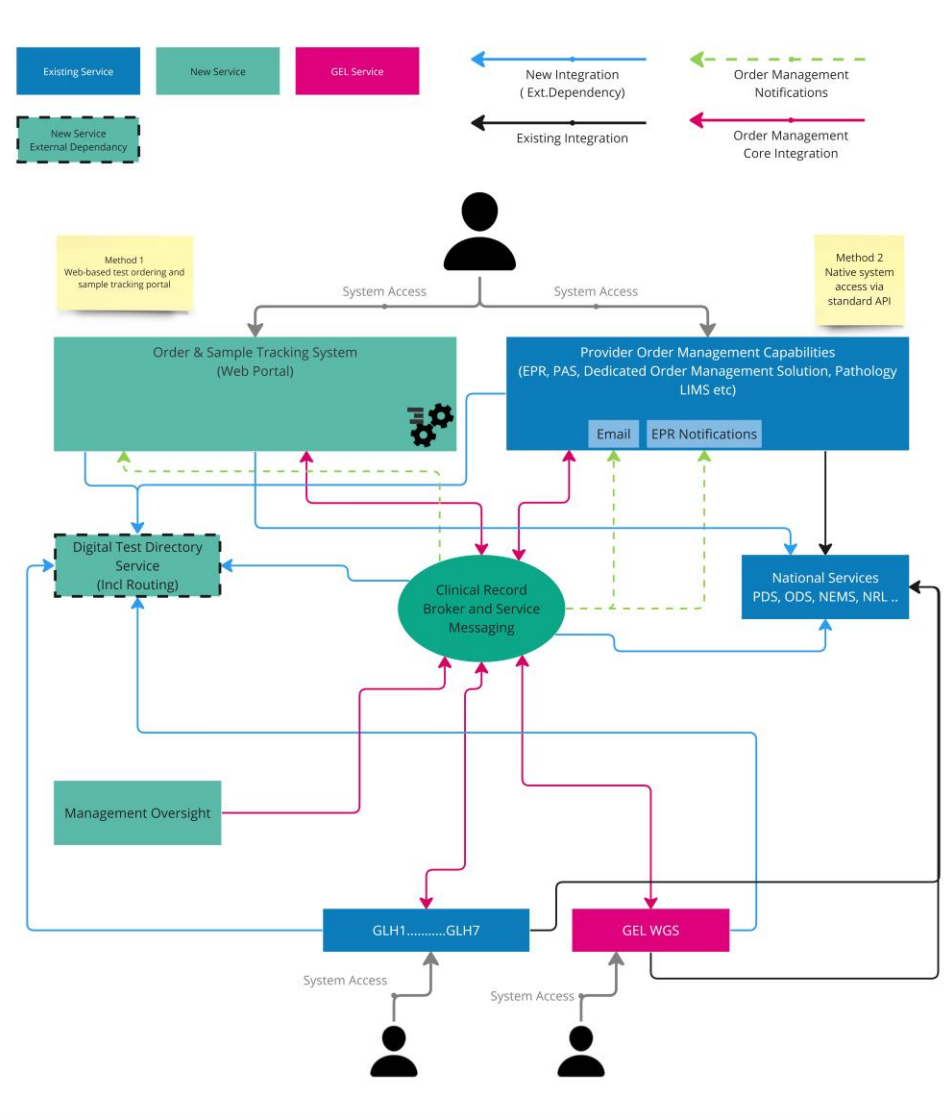
- Participants (actors) and observers (SMEs) will provide feedback on the IG

Architecture

Order Management Service - Overview

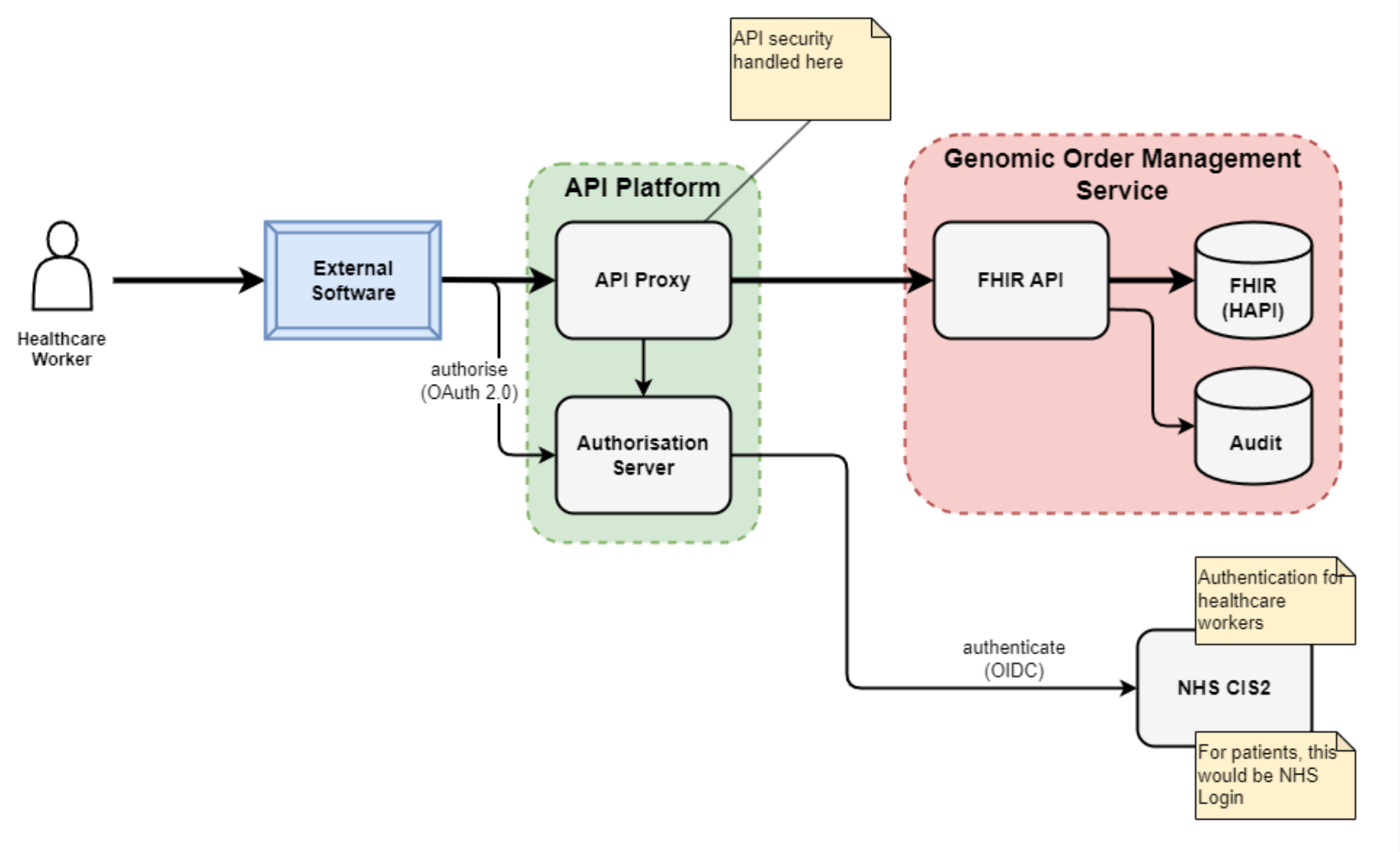


Architecture - ALPHA

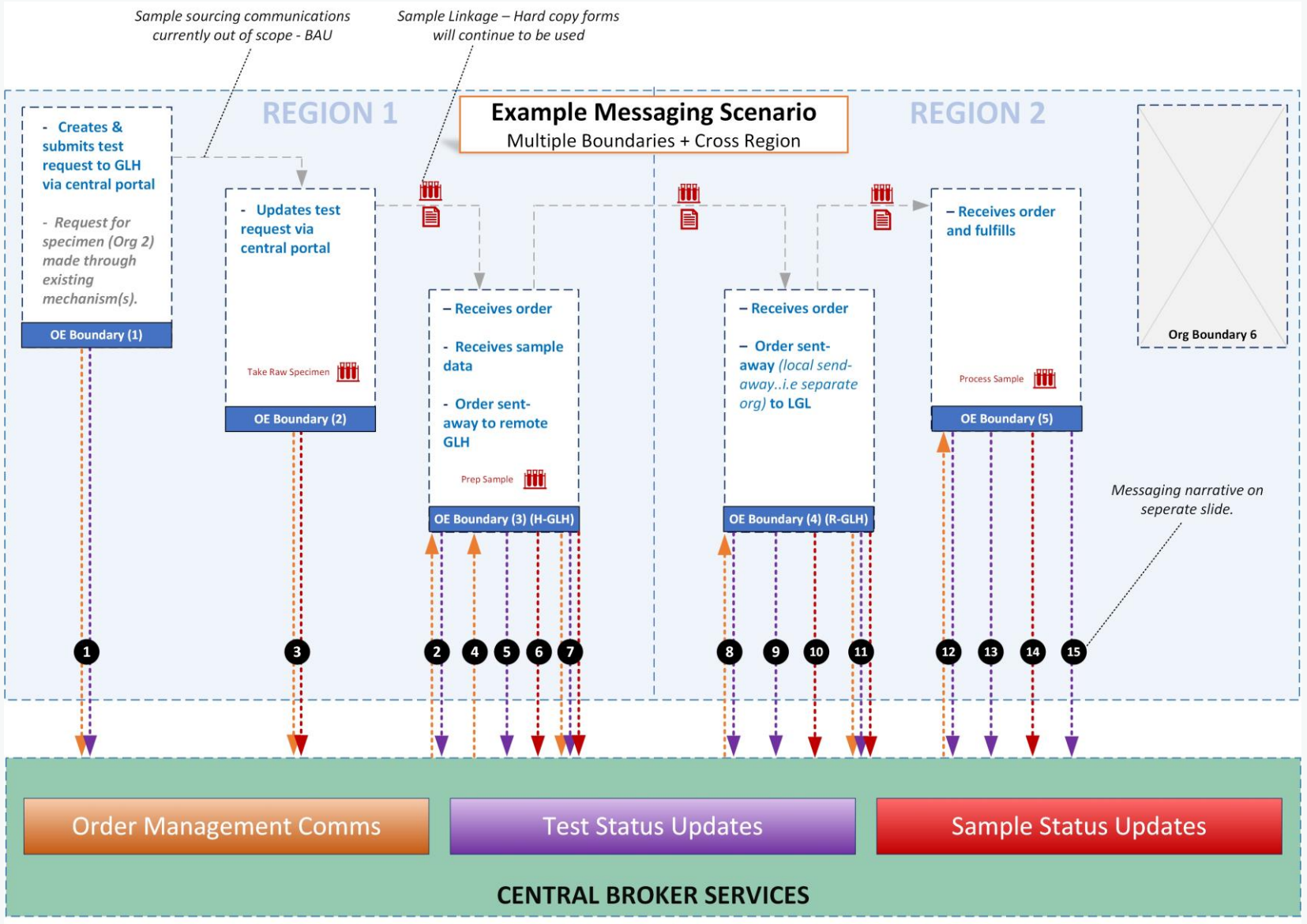


- Adheres to original architecture vision
- Delivering to meet the needs BRD and User requirements.
- UGR out of scope
- Limited scope for management oversight
- Core broker with end-to-end orchestration features
- Delivered in a NON-PRODUCTION environment using synthetic patient data
- NHSE API - https://digital.nhs.uk/developer/api-catalogue/genomic-order-management-service-fhir
- NHSE FHIR IG - https://simplifier.net/guide/fhir-genomics-implementation-guide?version=0.4.2

Genomic Order Management – ALPHA APIM



Example Communication

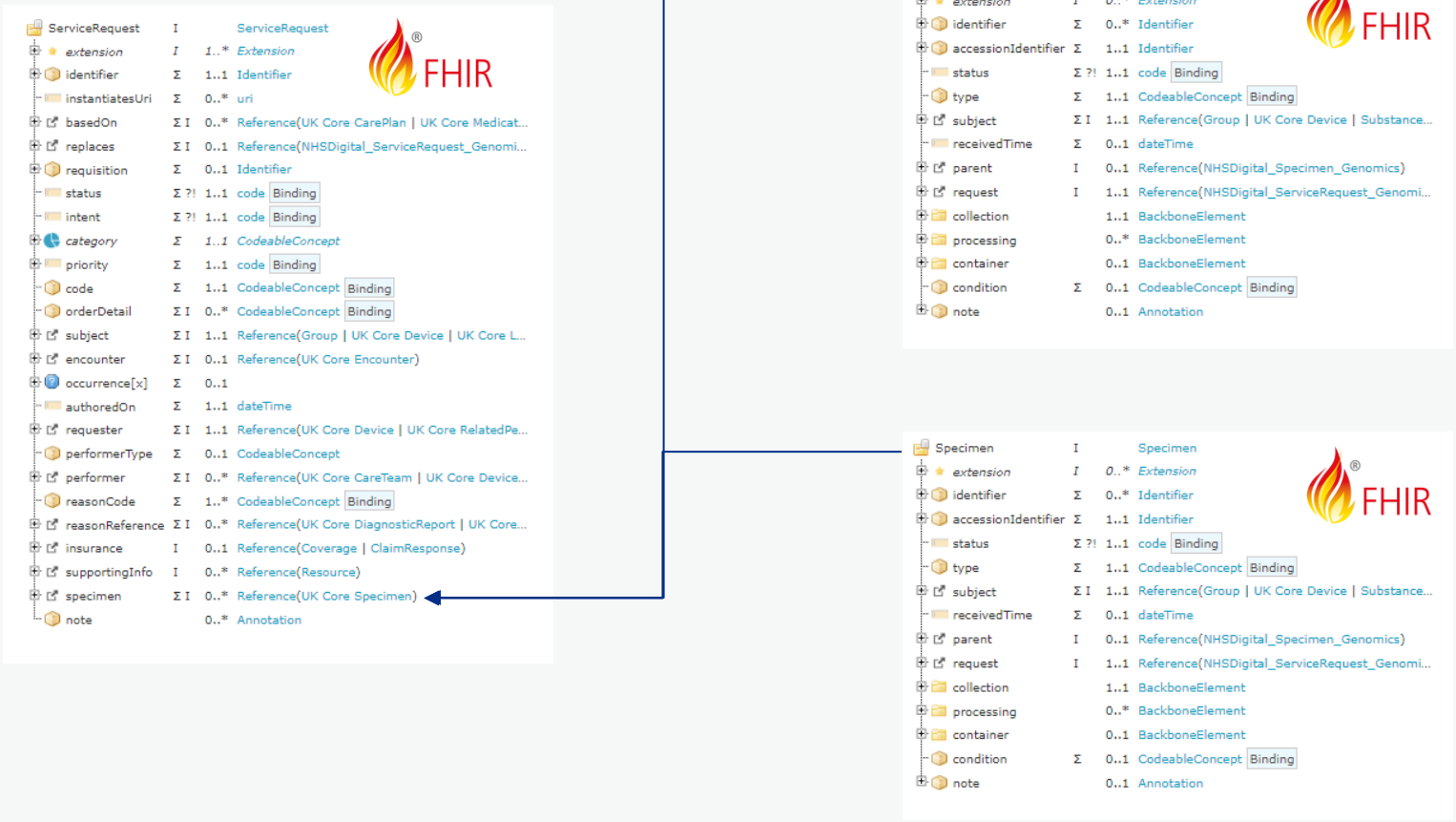


Entities in FHIR Service Request

ServiceRequest	I		ServiceRequest
extension	*I*	*1..**	*Extension*
identifier	Σ	1..1	Identifier
instantiatesUri	Σ	0..*	uri
basedOn	Σ I	0..*	Reference(UK Core CarePlan | UK Core Medicat...
replaces	Σ I	0..1	Reference(NHSDigital_ServiceRequest_Genomi...
requisition	Σ	0..1	Identifier
status	Σ ?!	1..1	code Binding
intent	Σ ?!	1..1	code Binding
category	*Σ*	*1..1*	*CodeableConcept*
priority	Σ	1..1	code Binding
code	Σ	1..1	CodeableConcept Binding
orderDetail	Σ I	0..*	CodeableConcept Binding
subject	Σ I	1..1	Reference(Group | UK Core Device | UK Core L...
encounter	Σ I	0..1	Reference(UK Core Encounter)
occurrence[x]	Σ	0..1	
authoredOn	Σ	1..1	dateTime
requester	Σ I	1..1	Reference(UK Core Device | UK Core RelatedPe...
performerType	Σ	0..1	CodeableConcept
performer	Σ I	0..*	Reference(UK Core CareTeam | UK Core Device...
reasonCode	Σ	1..*	CodeableConcept Binding
reasonReference	Σ I	0..*	Reference(UK Core DiagnosticReport | UK Core...
insurance	I	0..1	Reference(Coverage | ClaimResponse)
supportingInfo	I	0..*	Reference(Resource)
specimen	Σ I	0..*	Reference(UK Core Specimen)
note		0..*	Annotation

Genomics England

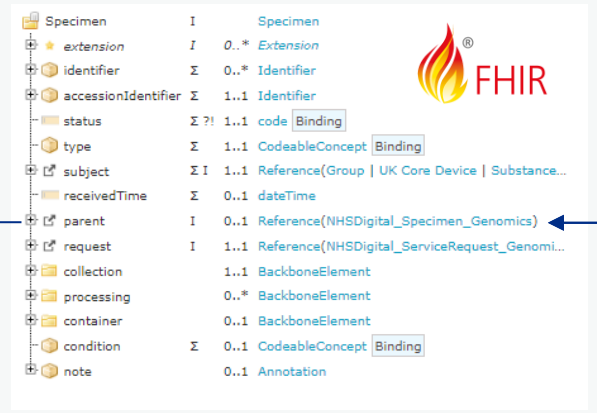
FHIR Service Request and Specimen



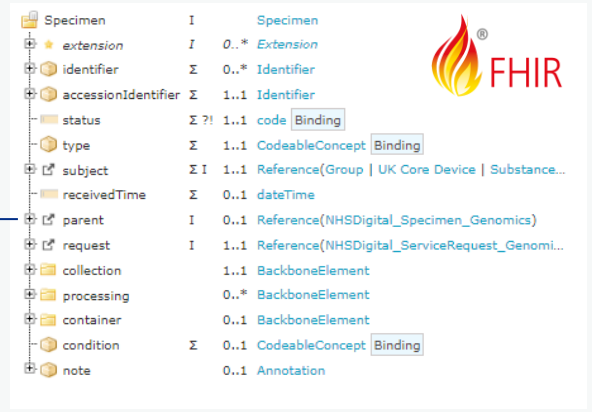
FHIR Specimen Modelling

Raw Specimen 1..* (NHS Trust)

DNA Sample 1..* (GLH)



DNA Sample1

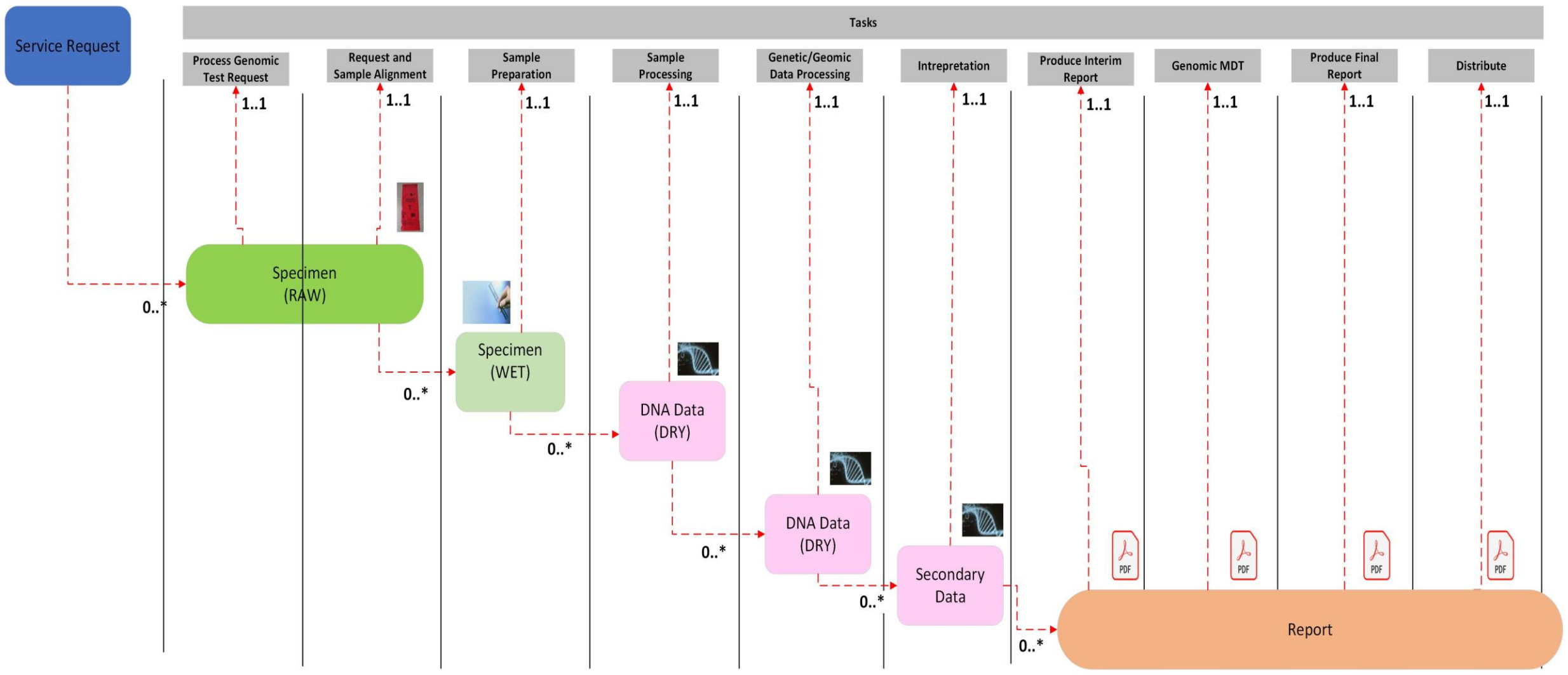


DNA Sample2

FHIR Resource Identifiers

- Identifiers
  - HL7 Standard - https://www.hl7.org/fhir/resource.html#identification
  - NHS FHIR Policy - https://nhsconnect.github.io/fhir-policy/identifiers.html
  - FHIR for FAIR - https://build.fhir.org/ig/HL7/fhir-for-fair/FHIRidentifiers.html
- Each FHIR resource has a “Resource.**id**” element which captures the logical identity of the resource.
  - Assigned by the server responsible for storing it.
  - This identifier will differ if the resource is moved or persisted on another server (it is analogous to the primary key in a database).
  - This id is the key to manage CRUD operations against the services.
- Resource will also have one or more “business” identifiers captured within Resource.**identifier**.
  - These have a business meaning outside the FHIR server (e.g. an NHS number or Organisation Identifier)
  - Organisation are allowed to pass in business identifiers with namespace and update them as appropriate.
- Central Broker will centrally allocate logical id values for all FHIR resources passing through the services during persistence. This is for
  - Consistent identification/versioning/ management
  - Avoiding collision of identifiers across similar resources sent from multiple NHS organisations.
  - API operations on the resources are based on the logical id to
    - Ensure consistency
    - Prevent collisions
    - Prevent data mix up leading to clinical issues
  - Adopting exiting NHS practices within national services.
- Systems interacting with the central services API's need to store the logical id locally for interactions.

Orders vs Specimen vs Reports vs Tasks



Order Scenarios vs Task Resource Cardinalities

Type	No. Test Requests	No. Primary Sample	No of DNA Samples	Task	No. Test Reports	Notes
Singleton / Duo /Trio	1	1..* Proband	1..* Proband	Process Genomic Test Request [1..1] [D] - Service Request Request & Sample Alignment [1..*] **/ Sample** Sample Preparation [1..*] **/ Sample** Sample Processing [1..*] **/ Sample** Genetic/Genomic Data Processing [1..*] **/ Sample** Interpretation [1..*] **/ Sample** Produce Interim Report [1..*] **/ Sample** Genomic MDT [1..1] [D] - - Service Request Produce Final Report [1..1] [D] - Service Request Distribute Report [1..1] [D] - Service Request	1..*	DNA Samples would be modelled as FHIR Specimen resources. Represented as parent child relationship. Interim reports are shared in the flows and eventually a final report is made available to clinician.
Duo	1	1..* Proband [P] 1..* Consultand C1	1..* Proband [P] 1..* Consultand [C1]	Process Genomic Test Request [1..1] [D] - Service Request Request & Sample Alignment [1..*] **/ Sample** Sample Preparation [1..*] **/ Sample** Sample Processing [1..*] **/ Sample** Genetic/Genomic Data Processing [1..*] **/ Sample** Interpretation [1..*] **/ Sample** Produce Interim Report [1..*] **/ Sample** Genomic MDT [1..1] [D] - - Service Request Produce Final Report [1..1] [D] - Service Request Distribute Report [1..1] [D] - Service Request	1..*	
Trio	1	1..* Proband [P] 1..* Consultand [C1] 1..* Consultand [C2]	1..* Proband [P] 1..* Consultand [C1] 1..* Consultand [C2]	Process Genomic Test Request [1..1] [D] - Service Request Request & Sample Alignment [1..*] **/ Sample** Sample Preparation [1..*] **/ Sample** Sample Processing [1..*] **/ Sample** Genetic/Genomic Data Processing [1..*] **/ Sample** Interpretation [1..*] **/ Sample** Produce Interim Report [1..*] **/ Sample** Genomic MDT [1..1] [D] - - Service Request Produce Final Report [1..1] [D] - Service Request Distribute Report [1..1] [D] - Service Request	1..*	

[D] - Task Created on Default , Tasks related to specimen are created when specimen is made available to Core Broker

Demonstration of Concept

Next Steps

Next Steps

- Register for the Connectathon:
  - January 15th: NHS Genomic Medicine Service – Connectathon
  - Feb 21st: Register here: Connectathon
- Prep and Attend the Connectathon at TechUK
- Provide Feedback
- Attend Connectathon Lessons Learned Session:
  - Teams Meeting Invite to be circulated following Feb session

Thank You

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