National Genomic Testing Process

Document Purpose

This diagram has been developed within the national interoperability project for the GMS. The purpose of the operational flow is to provide the visual reference points against which data requirements can be stated.

Synopsis – Principles Applied

This diagram illustrates a nationally applicable, generic process occurring between the requesting of genomic tests and the subsequent receipt of result reports. The scope of the diagram is held strictly between these events. To achieve this, the operational task sequence depicted is solution and location agnostic. Therefore, the diagram does not illustrate lower levels of detail inside organisational boundaries, which may differ between locations.

The diagram is presented from the perspective of the originating/Home GLH of the test request. Additionally, the process represents the task sequence applicable per test.

Finally, the diagram demonstrates how the Home GLH may be the facilitator of a specialist test or interpretation on behalf of a remote requestor.

Version Control

Version	Date	Modifier	Comments/Updates
0.1	11.02.22	M Price	Initial draft – SE_GLH process flow lifted into BPMN template- method and style adjustment. Process and sequence updates applied.
0.2	14.02.22	M Price	Interpretation & Report expanded sub processes added
0.3	17.02.22	M Price	Variant Identification process steps added, Reflex Testing placement adjusted. Validation of draft (with Subject Matter Experts) to commence from this version onwards. Extensive updates planned for subsequent drafts.
0.4-05	10.03.22	M Price	Review and updates via Clinical Directors (Cancer pathways) + Further informatics input driven amendment.
0.6	17.03.22	M Price	Error paths added/developed + minor amends. DPYD scenario added (new slide)
0.7	28.03.22	M Price	Copy is post Informatics review 28th Mar. WGS Scenario slide added. Further updates and validation now planned. Minor amendments include:- various terminology updates, additional decision point in error flow. Counterpart documents to update also.
0.8	04.04.22	M Price	Reference labels added and updated to align with data counterpart document
0.9	29.04.22	M Price	Minor amends following Scientific Advisors and Ops Directors reviews - 5.11.1 task added, 3.01 – 3.05 sequence revised.
0.9.1	06.05.22	M Price	Task 3.02 sequences updated, intermediate events adjusted.
0.9.2	26.05.22	M Price	DPYD & WGS copies uplifted to match current master diagram.
0.9.3	01.07.22	M Price	Copy Approved @ GOMT. Minor labelling amendment at 2.09.
1.0	**10.10.22**	**M Price**	**Baselined – draft stable**

BPMN & General KEY

Start Event: – Indicates the event that triggers the start of the process.

End Event – Indicating the event that occurs, marking the end points of either sub processes of the main process flow.

Intermediate Event – Represents any notable event that occurs between the start and end events.

Intermediate Timer – Indicates a time constraint applicable to a task.

Gateway: Parallel – Depicts commencement of tasks occurring in parallel. Follow-on instances ensure that parallel tasks complete prior to the next task in the flow commencing.

Gateway: Either/Or – Indicating the process flow will only follow one of the subsequent attached flows.

Gateway: Inclusive – Present when one or more flows can be followed optionally. The second, merging instance serves to ensure all active sequences complete prior to any subsequent task commencing.

Sub Task: Collapsed – Indicates a task that is comprised of a series of sub-tasks, which are linked to within the document and expanded in a separate page.

Sub Task: Expanded – Used to contain the expanded view of sub process tasks.

Sequence Flows:– Indicate and connect the general flow between tasks, gateways and events. Additional markers are used, such as the forward slash, which depicts the default flow, where multiple flows are present.

Message Flows: – Serves as the bridging link between tasks in separate pools or lanes, and indicates the flow of messages between tasks.

Document Validation

This document has been validated by providing communities-of-interest, with the opportunity to review and feedback. A combination of interviews and workshops has been held with Operational, Scientific and Informatics stakeholders. The following, non-exhaustive list of roles is applicable. The document has been exposed to multiple stakeholders from each region engaged in the GMS.

- Director of Operations
- Deputy Operations Lead for Clinical Bioinformatics
- GLH Bioinformatics Lead
- Deputy Directors to the GU
- Scientific Director
- Consultant Clinical Scientist Lead
- Principal Clinical Scientist
- Consultant Clinical Scientist
- Clinical Scientist
- Consultant in Clinical Genetics
- Laboratory Lead for Precision Medicine
- Cardiologist
- Scientific Advisor to the GU
- Informatics lead
- IT Manager

National Genomic Testing Process | – Master Diagram

V1.0

Ordering Entity

Referring Clinician

RoD/ Consent

Patient

Sample(s) S1

Genomic Test Required

1.01 Create Test Request (As NGTD)

Nominated Clinical Staff

Ordering Entity Notified of Process Failure

1.04 Store/Log Test Report

End

Test Report Received

Detail required locally or remotely? 1.03

Local

1.02 Provide MDT with Further Clinical Details

GLH (Home coordination point)

Send Away/ Specialist? 2.24

N

Y

Invalid Test

No

2.02 Test Request Valid

2.01 Process Test Request (Receipt/Entry)

Test Request Received

Yes

2.03 Require External Lab Sample

No

Yes

2.04 Request Sample (From external lab)

2.05 Log Sample Confirmation

Fail

2.23 Notify Requestor

Fail Test? 2.22

2.21 Determine Test Recoverability

Looping process to attain all required samples + request.

2.07 Forward Order Request

2.08 Forward Order Request

Technically Processing Genetic/ Genomic Data Locally? 2.09

No

Other GLH Processing Genetic/ Genomic Data? 2.10

Yes

No

Local Interpretation 2.11

Other GLH Interpreting 2.12

Require MDT 2.13

Yes

No

2.14 Genomic MDT (GTAB/GRDAB)

Require Further Clinical Detail 2.15

Local Interpretation 2.16

Reflex Test Required 2.18

2.17 Mark Test Complete

2.19 Distribute Report

2.20 Local/Remote Requestor

Local

Remote

Wet Lab (Local)

3.01 Book Sample(s) into Lab

3.02 Align Sample and Test Request/ Order

3.17

3.18

Sample or Request Not Present (Conditional, test-dependent time frames)

All Required Sample(s) & Request Received or Available

3.05 Send Away Sample

3.06 Process Sample *(Validate,Extract ,Plate, Sequence)*

3.07 Successful?

Send Away DNA/RNA? 3.08

3.09 Genetic/ Genomic data processing

Conducting Interpretation? 3.10

3.11 Interpretation

3.12 Successful?

3.13 Reflex/ Confirmatory Test?

3.14 Produce Initial Report

3.15 MDT Required

3.16 Finalise Report

Sample(s) S2

Dry Lab (Local)

Sample(s) S3

Sample icons are each potential start events

Sub Process Scenario Variables:

1) Sample validation

2) DNA Extraction

3) Plating

3) Sequencing

4.01 Genetic/ Genomic data processing

4.02 Conducting Interpretation?

4.03 Interpretation

4.04 Successful?

4.05 Reflex/ Confirmatory Test?

4.06 Produce Initial Report

4.07 MDT Required

4.08 Finalise Report

Remote GLH/Genetics Lab Send Aways & Specialist Tests

Request for Specialist Test Sent *(From a Remote GLH)*

RS1

5.01 Process Sample *(Validate,Extract ,Plate, Sequence)*

5.02 Successful?

5.03 Technically Processing Genetic/ Genomic Data?

5.04 Genetic/ Genomic data processing

Request for Specialist Interpretation *(From a Remote GLH)*

RS2

5.05 Conducting Interpretation?

5.06 Interpretation

5.07 Successful?

5.08 Reflex/ Confirmatory Test?

5.09 Produce Initial Report

5.10 MDT Required

5.12 Genomic MDT (GTAB/GRDAB)

Require Further Clinical Detail 5.11

5.11 Finalise Report

5.11.1 Distribute Report

Test Report Received

5.14 Store/Log Test Report

End

External Lab (Other/Sample Banks)

Ordering Entity Notified of Process Failure

6.01 Send Sample Availability Confirmation

Sample(s) S4

6.02 Sample(s) Available

Y/N

6.03 Identify Test/ Sample(s)

National Genomic Testing Process | – DPYD Scenario (Happy Path)

V0.9.3

Ordering Entity

GLH (Home coordination point)

Wet Lab (Local)

Dry Lab (Local)

Remote GLH/Genetics Lab Send Aways & Specialist Tests

External Lab (Other/Sample Banks)

Example Scenario 1 - DPYD

- Sample and request provided locally
- Local web-lab test execution
- No MDT required

Referring Clinician

Patient

RoD/ Consent

Sample(s) S1

Genomic Test Required

1.01 Create Test Request (As NGTD)

Nominated Clinical Staff

2.01 Process Test Request (Receipt/Entry)

2.02 Test Request Valid

Test Request Received

Yes

2.03 Require External Lab Sample

No

3.01 Book Sample(s) into Lab

3.02 Align Sample and Test Request/ Order

3.03

3.04 All Required Sample(s) & Request Received or Available

3.05 Send Away Sample

No

3.06 Process Sample *(Validate,Extract ,Plate, Sequence)*

3.07 Successful?

Yes

3.08 Send Away DNA/RNA?

No

3.09 Genetic/ Genomic data processing

3.10 Conducting Interpretation?

Yes

3.11 Interpretation

3.12 Successful?

Yes

Reflex/ Confirmatory Test?

No

3.14 Produce Initial Report

3.15 MDT Required

No

3.16 Finalise Report

End

1.04 Store/Log Test Report

Test Report Received

Local

2.15 Reflex Test Required

No

2.19 Distribute Report

2.20 Local/Remote Requestor

2.17 Mark Test Complete

Sub Process Scenario Variables:

1) Sample validation

2) DNA Extraction

3) Plating

3) Sequencing

National Genomic Testing Process | – WGS Scenario (Happy Path)

V0.9.3

Example Scenario 2 - WGS

- Fresh tissue sample provided by ordering entity
- Send-Away DNA scenario
- Local Dry Lab Interpretation
- MDT required

Ordering Entity

GLH (Home coordination point)

Wet Lab (Local)

Dry Lab (Local)

Remote GLH/Genetics Lab(s) Send Aways & Specialist Tests

External Lab (Other/Sample Banks)

Patient

Referring Clinician

RoD/ Consent

Sample(s) S1

Genomic Test Required

1.01 Create Test Request (As NGTD)

Nominated Clinical Staff

Ordering Entity Notified of Process Failure

2.01 Process Test Request (Receipt/Entry)

Test Request Received

2.02 Test Request Valid

2.03 Require External Lab Sample

Yes

No

2.01 Book Sample(s) into Lab

3.02 Align Sample and Test Request/ Order

3.03 All Required Sample(s) & Request Received or Available

3.05 Send Away Sample

3.06 Process Sample *(Validate, Extract, Plate, Sequence)*

3.07 Successful?

Yes

2.08 Forward Order Request

3.08 Send Away DNA/RNA?

Yes

No

Sub Process Scenario Variables:

1) Sample validation

2) DNA Extraction

3) Plating

3) Sequencing

5.01 Process Sample *(Validate, Extract, Plate, Sequence)*

5.02 Technically Processing Genetic/ Genomic Data?

Yes

5.03 Genetic/ Genomic data processing

5.04 Conducting Interpretation?

Yes

No

5.05 Successful?

Yes

Local Interpretation

2.11

No

Other GLH Interpreting

2.12

No

4.03 Interpretation

4.04 Successful?

Yes

4.05 Reflex/ Confirmatory Test?

No

4.06 Produce Initial Report

4.07 MDT Required

Yes

No

2.13 Genomic MDT (GTAB/GRDAB)

Require Further Clinical Detail

2.14

No

2.15 Local Interpretation

No

2.16 Reflex Test Required

No

2.19 Distribute Report

2.20 Local/Remote Requestor

Local

2.17 Mark Test Complete

4.08 Finalise Report

Test Report Received

1.04 Store/Log Test Report

End

Request for Specialist Test Sent *(From a Remote GLH)*

5.06

Request for Specialist Interpretation *(From a Remote GLH)*

5.07

Identify Test/ Sample(s)

1.02

Send Sample Dispatch Confirmation

Sample(s) S1

Sample(s) Available