Contents

1 Introduction

1.1 Purpose

This is the User Requirements Specification (URS) document for the `gel2mdt` project. The project aims to create a database to store data from the GeL CIP-API alongside patient demographic information and information about clinical workflow steps then subsequently display this data to clinical scientists thereby facilitating the reporting of variants.

1.2 Scope

This document outlines the requirements identified by proposed users of `gel2mdt`. The intended readership is West Midlands Regional Genetics Laboratories (WMRGL) clinical scientists, Great Ormond Street Hospital (GOSH) clinical scientists, and the project developers.

1.3 Definitions & Abbreviations

FRS	Functional Requirements Specification
GOSH	Great Ormond Street Hospital
KPI	Key performance indicator
RTM	Requirement Traceability Matrix
URS	User Requirements Specification
WMRGL	West Midlands Regional Genetics Laboratories

1.4 Named people

HA	Helena Ahlfors — Project Lead
SC	Samuel Clokie — Project Lead
ES	Edward Stone — Developer
PL	Patrick Lombard — Developer
TC	Theo Cole — Developer

2 User Requirements Specification

#	Requirement Title	Requirement Details	Trust
Functional			
1.1.1	Storage and display of all patients within the GeL CIP-API.	Data stored should include proband ID, family ID, interpretation ID, date last updated, CIP name, and the site code.	Both
1.1.2	Storage and display of case data for all with certain status types.	If the status type of a patient is 'sent to gmcs', 'report generated', or 'report sent' then case data should also be retrieved from GeL CIP-API.	Both
1.1.3	Demographic information should be stored.	(i) For patients described in 1.1.2, demographic information should also be pulled from LabKey or the the Trust's LIMS. (ii) Users need to be able to manually specify a lab ID number, clinical scientist name, CGU number, and comments about the patient.	Both
1.1.4	Flag high priority cases.	Cases with Tier 1, Tier 2, or Promoted variants should be flagged for review by clinical scientists.	Both
1.1.5	Facilitate variant review	Details required for variant review should be pulled from GeL CIP-API and other necessary sources to facilate the review of variants. These should include genomic position, nomenclature (including HGVS), tier, and pathogenic information.	Both
1.1.6	Confirm and report variants	(i) When reviewing variants, users need to be able to set a variant as 'Confirmed'. (ii) Additionally, users need to be able to set a variant as 'Included in Report'. (iii) Once a variant has been reviewed, a second clinical scientist user should be able to perform and record the results from a second check. (iv) Any variants that still require a second check should be flagged for checking.	WMRGL
1.1.7	Assign cases for MDT	Users should be able to assign particular patients for MDT then choose variants for discussion.	Both
1.1.8	View list of patients requiring MDT	Users should be able to quickly get a list of all patients who have been flagged for MDT and have yet to go through the MDT process.	Both
1.1.9	Manage MDT cases	Users should be able to view an MDT screen for each patient requiring MDT, where they can input decisions made at each MDT and record attendees of the MDT.	Both
1.1.10	Design primers	When viewing any particular variant, users should be able to press a button to design primers for that variant.	WMRGL
1.1.11	Provide audit trail	Any changes to information in the database should be recorded along with the user who made the change.	Both
Non-functional			
1.2.1	Availibility	The webapp should be available for use at all times during standard working hours.	Both
1.2.2	Accessibility	The webapp should be accessible from any trust computer without the need for installation of any software.	Both
Constraints			
1.3.3	Deployment	The webapp and database should be fully deployable following standard deployment procedures at both GOSH and WMRGL.	Both

3   Version Changes

0.93 – Added CS name, CGU number, and comments box requierment to 1.1.3(ii).
– Added variant information to 1.1.5.
– Renamed 'confirm' and 'report' to 'Confirmed' and 'Included in Report' in 1.1.6.
– Added flagging of second checks to 1.1.6.
– Created 1.1.11 (Audit).

0.92 – Reformatted document.
– Moved technical requirements to FRS.
– Added Trust column.

0.91 – Initialised document