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| --- | --- | --- | --- |
|  |  | Document Record ID | |
| Work stream | Informatics |  | |
| Programme Director | Tom Fowler | **Status** | DRAFT |
| Document Owner | Amanda O’Neill | **Version** | V2.0.0 |
|  |  |  |  |
| Document Author | Listed Below | **Version Date** | 10/03/2017 |

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GENOMICS ENGLAND

*100,000 Genomes Project Release Notes*

*FINAL*

*Rare Disease Model - Release v2.0.0*

**2.0.0**

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## Version History

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| --- | --- | --- | --- |
| Version | Author | Date | Description |
| 2.0.0 | A Milward | 10/03/17 | DRAFT |
|  |  |  |  |

## Distribution list

This document must be reviewed by the following:

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## 

## Approvers

This document must be approved by the following:

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| Name | Responsibility | Date | Version |
| Amanda O’Neill | Director of Clinical Data |  |  |
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# Introduction

## Purpose

The purpose of this document is to provide a brief summary of the DRAFT 2.0.0 Rare Disease Data Model Package.

## Audience

This document is primarily written for informatics leads within the GMCs and those involved in the collection and submission of data for the UK 100,000 Genomes Project.

# What’s New?

Summary of the main changes:

NOTE: specific changes can be found in the change tab of the Excel data specifications

The Rare Disease Model update introduces changes to the core components of the model focused on the components of the data set required for interpretation. The main aims of the update are: (1) to bring the rare disease model in line with components also present in the cancer model; (2) standardisation of the models to ensure, for example, standard patterns of metadata for all schemas; (3) modification of key fields required for interpretation; and (4) removal components no longer necessary or add components.

The update also separates the Sample Tracking component of the model. This brings the specification for the sample tracking part of the model into line with the Cancer model, and provides clearer guidance on the kinds of information needed with different types of sample.

## Additions

* Alignment with shared cancer models
  + Participant identifiers added for devolved nations
  + Consent and Consent update aligned with cancer
* Couple reports added
* Phenotype modifiers made more flexible

## Removals

* Sample Tracking removed from clinical data specification
* Family history requirements reduced
* Elements removed to align with cancer models

## Changes

* Withdrawals aligned with cancer models
* Diagnoses
* Bug fixes to death details
* Registration and consent bug fixes

## Note

New specifications for Pedigree reports, Interventions and Investigations are still under development.

# Package Contents

## Rare Disease Clinical Data Specifications for XML Submissions and Open Clinica Forms:

1. **Rare Disease Model-2.0.0.docx** - Rare Disease Clinical Data Specification as a Word Document describing the XML message definitions for Genomic Medicine Centres (GMCs) to provide clinical data for the programme from registration, to the description of their Rare Disease diagnosis and stage, through their treatment pathway.
2. **Rare Disease Model-2.0.0.xslx** - Rare Disease Clinical Data Specification AND CHANGE LOG as an Excel Document describing the XML message definitions for Genomic Medicine Centres (GMCs) to provide clinical data for the programme from registration, to the description of their Rare Disease diagnosis and stage, through their treatment pathway.

CHANGES: New Additions to the model are highlighted in Green, Removals are highlighted in Red, Superficial changes (such as descriptions) are highlighted in blue.

## Rare Disease Sample Tracking Data Specifications – CSV Submissions and examples:

1. **Rare Disease Sample Tracking-2.0.0.docx** - Rare Disease Sample Tracking Specification as a Word Document describing the csv message definitions for Genomic Medicine Centres (GMCs) to provide sample tracking data for the programme.
2. **Rare Disease Sample Tracking-2.0.0.xslx** - Rare Disease Sample Tracking Specification AND CHANGE LOG as an Excel Document describing the csv message definitions for Genomic Medicine Centres (GMCs) to provide sample tracking data for the programme. New CHANGES: Additions to the model are highlighted in Green, Removals are highlighted in Red, Superficial changes (such as descriptions) are highlighted in blue

# Known Deficiencies

## Inherent Defects

Specifications for Pedigree reports, and a detailed catalogue of Interventions and Investigations are under development. The timescales of these will be discussed during the consultation period of this release. Feedback on the proposed general model for Interventions and Investigations would be gratefully received.

# Impact of Release

## GMC Impact

Valid processing of XML submissions for against the published model.

## Biorepository Impact

None

## Sequencer Impact

None

## Support Impact

None anticipated beyond existing considerations.

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END OF RELEASE NOTE