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GENOMICS ENGLAND

*100,000 Genomes Project Release Notes*

*FINAL*

*Rare Disease Schemas 1.3.3*

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# Document Control

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## Version History

|  |  |  |  |
| --- | --- | --- | --- |
| Version | Author | Date | Description |
| V00\_01 | A Milward | 14/03/16 | Initial draft |
| V00\_02 | A Milward | 15/03/16 | Updated following consultation with RD team |

## Distribution list

This document must be reviewed by the following:

|  |  |
| --- | --- |
| Name | Organisation |
| Nathan Hicks | Informatics |
| Jim Davies | Informatics |
| James Thomas | Informatics |
| Atul Hatwal | Informatics |
| Reshmi Parag | PMO |
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| Grant Stapleton | Head of Service Management |

## 

## Approvers

This document must be approved by the following:

|  |  |  |  |
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| Name | Responsibility | Date | Version |
| Nathan Hicks | Software Development Manager |  | 2 |
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# Introduction

## Purpose

The purpose of this document is to provide a brief summary of bug fixes within the 100K Genomes Project Schemas that validate Rare Disease submitted via XML.

## Audience

This document is written for Genomics England general distribution and users of the software products providing data using XML format.

This release describes components of XML schemas deployed as part of 100k Genomes Project v1.7:

* Revised **Schemas v1.3.3**
* Revised **XML Data Capture to accommodate new schemas for Rare Diseases** via the sFTP file upload facility

These revisions to schemas will be deployed to the live environment as part of v1.8.2 release on the 24/03/2016 and are described within the release note to ensure consistency of the schemas published by NHSE and validated by the 100k Genomes Project live system.

Technical guidance and installation instructions are documented separately from this note and controlled internally by Genomics England.

These release notes reside in the Genomics England Huddle repository, under informatics\software development\implementation.

All QA Test Reports are included within JIRA for each of the components within this release.

# What’s New?

## Additions

* None

## Removals

* None

## Changes

As part of QC the following schemas have been updated:

* **“Registration and Consent RD-v1.5.2.xsd”** 
  + **New version released – “RegistrationAndConsent RD-v1.5.3.xsd”**
  + **Namespace updated to 1.3.3**
    - <xs:schema xmlns:xs="http://www.w3.org/2001/XMLSchema"  
       xmlns:vc="http://www.w3.org/2007/XMLSchema-versioning"  
       xmlns:gelRD="https://genomicsengland.co.uk/xsd/raredisease/1.3.3"  
       xmlns="https://genomicsengland.co.uk/xsd/raredisease/1.3.3"  
       targetNamespace="https://genomicsengland.co.uk/xsd/raredisease/1.3.3" vc:minVersion="1.1">
  + **Eligibility element made optional within schema**
    - <xs:element name="eligibility" type="eligibility-15016.2" minOccurs="0" maxOccurs="1"/>
    - **NOTE:** Aneligibility element must be provided for every Proband – xml submissions will fail if this is not included. Relatives do not have to provide this information
  + **Diagnosis details made optional**
    - <xs:element name="diagnosis" type="diagnosis-35968.1" minOccurs="0" maxOccurs="unbounded"/>
    - **NOTE:** A diagnosis element must be provided for every Proband – xml submissions will fail if this is not included. Relatives do not have to provide this information

# Known Deficiencies

## Inherent Defects

Software defects are logged as ‘bugs’ in the Genomics England JIRA system.

The following must be added to the investigation schemas:

* **“Investigation RD-v1.0.2.xsd”** 
  + **Genetic Results** 
    - <xs:element name="genetic-results" type="genetic-results-37489.1" minOccurs="0"  
       maxOccurs="unbounded"/>
  + **Genetic Reports** 
    - <xs:element name="genetic-reports" type="genetic-reports-37509.1" minOccurs="0"  
       maxOccurs="unbounded"/>
  + **Blood Pressure added (vital signs)**
    - <xs:element name="blood-pressure" type="blood-pressure-37510.1" minOccurs="0"  
       maxOccurs="unbounded"/>
  + **Weight (vital signs)**
    - <xs:element name="weight" type="weight-37512.1" minOccurs="0" maxOccurs="unbounded"/>
  + **Additional Body Measurements added (vital signs)**
    - <xs:element name="additional-body-measurements" type="additional-body-measurements-37511.1"  
       minOccurs="0" maxOccurs="unbounded"/>
  + **Autoantibodies** 
    - <xs:element name="autoantibodies" type="autoantibodies-37513.1" minOccurs="0"  
       maxOccurs="unbounded"/>

# Impact of Release

## GMC Impact

With this release GMCs will be able to submit XML for main programme rare disease in accordance with the new model.

## Biorepository Impact

None.

## Sequencer Impact

None.

## Support Impact

None anticipated beyond existing considerations.

The contact details for the Genomics England Contacts Desk are:

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END OF RELEASE NOTE