

Rare Disease Common Data Model (RD CDM) v2.0

1 Formal Criteria

- ▶ 1.1 Pseudonym <string>
- ▶ 1.2 Date of Admission <date>

2 Personal Information

- ▶ 2.1 Date of Birth <date>
- ▶ 2.2 Sex at Birth <code>
- ▶ 2.3 Gender <code>
- ▶ 2.4 Country of Birth <code>

3 Patient Status

- ▶ 3.1 General Clinical State <code>
- ▶ 3.2 Time of Death <date>
- ▶ 3.3 Cause of Death <code>
- ▶ 3.4 Age Category <code>
- ▶ 3.5 Length of Gestation at Birth <string>
- ▶ 3.6 Undiagnosed Case <boolean>

4 Care Pathway

- ▶ 4.1 Encounter Start <date>
- ▶ 4.2 Encounter End <date>
- ▶ 4.3 Encounter Status <code>
- ▶ 4.4 Type of Encounter <code>

5 Disease

- ▶ 5.1 Disease <code>
- ▶ 5.2 Disease Subtype <code>
- ▶ 5.3 Verification Status <code>
- ▶ 5.4 Age at onset <code>
- ▶ 5.5 Date of onset <date>
- ▶ 5.6 Age at Diagnosis <code>
- ▶ 5.7 Date of Diagnosis <date>
- ▶ 5.8 Body Site <code>
- ▶ 5.9 Clinical Status <code>
- ▶ 5.10 Severity <code>

7 Consent

- ▶ 7.1 Consent Status <code>
- ▶ 7.2 Consent Date <date>
- ▶ 7.3 Health Policy Monitoring <string>
- ▶ 7.4 Agreement Contact for Research <code>
- ▶ 7.5 Consent Reuse of data <code>
- ▶ 7.6 Biological sample <boolean>
- ▶ 7.7 Link to a biobank <string>

8 Disability

- ▶ 8.1 Classification of functioning & disability <code>

6 Diagnosis

6.1 Genetic Findings

- ▶ 6.1.1 Genomic Diagnosis <code>
- ▶ 6.1.2 Progress Status of Interpretation <code>
- ▶ 6.1.3 Interpretation Status <code>
- ▶ 6.1.4 Structural Variant Analysis Method <code>
- ▶ 6.1.5 Reference Genome <code>
- ▶ 6.1.6 HGVS String <string>
- ▶ 6.1.7 Genomic DNA Change <code>
- ▶ 6.1.8 DNA Change <code>
- ▶ 6.1.9 Amino Acid Change <code>
- ▶ 6.1.10 Gene <code>
- ▶ 6.1.11 Zygosity <code>
- ▶ 6.1.12 Genomic SourceCclass <code>
- ▶ 6.1.13 DNA Change Type <code>
- ▶ 6.1.14 Clinical Significance <code>
- ▶ 6.1.15 Therapeutic Actionability <code>
- ▶ 6.1.16 Clinical Annotation Level Of Evidence <code>

6.2 Phenotypic Findings

- ▶ 6.2.1 Phenotype <code>
- ▶ 6.2.2 Determination Date <date>
- ▶ 6.2.3 Status <code>

6.3 Family Medical History

- ▶ 6.3.1 Propositus/-a <code>
- ▶ 6.3.2 Relationship to Propostius/-a <code>
- ▶ 6.3.3 Consanguinity <code>
- ▶ 6.3.4 Family Member Relationship <code>
- ▶ 6.3.5 Family Member Record Status <code>
- ▶ 6.3.6 Family Member Sex <code>
- ▶ 6.3.7 Family Member Age <Integer>
- ▶ 6.3.8 Family Member Date of Birth <date>
- ▶ 6.3.9 Family Member Deceased <Boolean>
- ▶ 6.3.10 Family Member Cause of Death <code>
- ▶ 6.3.11 Family Member Deceased Age <Integer>
- ▶ 6.3.12 Family Member Affected by RD <code>