SNOMED CT to Orphanet Map package Production Release Notes - July 2021

Date	**20211031**
Document Version	**1.0**
Release Status	**PRODUCTION**

Introduction

The SNOMED CT to Orphanet Map Release is the product of a joint project carried out under a collaboration agreement between Institut national de la sante et de la recherche medicale (INSERM) and SNOMED International. Based on an agreed priority set, new concepts for rare diseases as defined in Orphanet have been added to SNOMED CT. A map from SNOMED CT to Orphanet has been created of this priority set.

One of the key use cases for this standardised map is to meet EU requirements to implement ORPHA codes in health systems for Rare Diseases epidemiology and research, including use in registries, enabling linkage from SNOMED CT enabled EHRs, and cross-border interoperability with ICD-based coding systems. Details are found in following links:

- Recommendation on ways to improve codification for rare diseases in health information systems (2014) European Commission Expert Group on Rare Diseases 2014: https://ec.europa.eu/health/sites/default/files/rare_diseases/docs/recommendation_coding_cegrd_en.pdf
- eHealth Network guidelines on Patient Summary : https://ec.europa.eu/health/sites/default/files/ehealth/docs/ehn_guidelines_patientsummary_en.pdf
- European Common Semantic Strategy 2019 (section 2.1 of the cover notes and deliverables document, in a ZIP file): https://ec.europa.eu/health/ehealth/events/ev_20191128_en

This document is intended to provide a brief description, background context and explanatory notes on the SNOMED International/INSERM collaborative work and the resulting SNOMED CT to Orphanet map. The map is being published as the Production release at this time with the intention of publishing annually in October. This document is not intended to provide technical specifications about SNOMED CT or Orphanet. Editorial guidance for SNOMED CT content can be found in the SNOMED CT Editorial Guide. Editorial guidance for Orphanet Nomenclature of Rare Diseases can be found in Orphanet (https://www.orpha.net/consor/cgi-bin/Education_Procedure.php?lng=en) and Orphadata (http://www.orphadata.org/cgi-bin/ORPHAnomenclature.html)

Audience

The audience for this document includes National Release Centers, vendors of electronic health records, terminology developers, researchers, genomics experts and individuals who wish to have an understanding of the content of the SNOMED CT to Orphanet map.

Background

SNOMED International (then IHTSDO) and INSERM first started exploring opportunities for linking SNOMED CT and Orphanet Rare Disease in 2012 because of the drivers to link between clinical data using SNOMED CT and the mandated use of Orphanet for rare disease research purposes in Europe and other countries outside Europe. As a consequence, an agreement was signed in 2014 and a priority set of rare diseases was agreed. Since then, the two organisations have worked closely as follows to:

1. Undertake a gap analysis for the defined set.
2. Develop textual definitions for the rare diseases based on expert input through the INSERM community.
3. Create new content in SNOMED CT.
4. Create a map between existing and new content in SNOMED CT and Orphanet.
5. Ensure throughout the development period that new SNOMED CT content aligns with changes in Orphanet.

3533 new rare disease concepts were added to SNOMED CT as part of this collaboration and the resulting map product contains 5,652 SNOMED CT and Orphanet concepts/terms.

In parallel to the content development and mapping work, the operational aspects have been agreed to ensure that the map is maintained and updated according to changes within the 2 terminologies. As a consequence, an updated Agreement was signed in March 2020 signalling both organisations commitment to maintaining and updating the content and map. Both organisation recognise the need for remaining in scope Orphanet content to be added to this map and this is to be done between now and October 2023 based on priorities of users.

Scope and Purpose of Collaborative Work

Under the terms of the Collaboration agreement between SNOMED International and INSERM, the Map will be issued annually by both SNOMED International in RF2 format and INSERM in spreadsheet format. Documented quality assurance processes will ensure that the content of both formats are the same and supporting materials are produced jointly. In addition, criteria for extending the Map will be agreed and managed through agreed processes to ensure that the product meets the needs of the different stakeholder groups based on use cases.

Design

Content Changes for Production Release

The key changes made to content in readiness for the move from the Alpha release to the Production release are:

1. Update to map for content that had been originally mapped to morphologic abnormality concepts.
2. Update to map for content that has been inactivated in SNOMED CT.
3. Removal of map from production release for entries that have been marked obsolete or deprecated in Orphanet.

Map

Decisions about maps between pre-existing content in SNOMED CT and Orphanet, the creation of new content for SNOMED CT and also Orphanet, and the validation of the map between SNOMED CT and Orphanet were made by consensus between both organisations.

Version

The version of Orphanet used is July 2021.

The version of SNOMED CT used is the July 2021 International Release.

Source for Map

The source data is a subset of 5652 SNOMED CT concepts.

Direction of Map

The direction of the map is from SNOMED CT July 2021 International Release to Orphanet July 2021.

Cardinality of Map

The cardinality in the table is one-to-one, one SNOMED CT concept maps to one Orphanet entry.

Obtaining the Release package

Access within SNOMED International member countries is provided by the Member National Release Centre in each country, via the relevant Member page. Affiliates of SNOMED International in non-member countries can access the map file through their Member Licensing and Distribution Service (MLDS) account. Please contact info@snomed.org for more information if required.

The Map in spreadsheet form is available for download from the Orphanet website: www.orphadata.org

If you're having any problems getting hold of the Orphanet Map Package, please contact info@snomed.org.

Users of the map must comply with licensing arrangements for both SNOMED CT and Orphanet.

Feedback

Feedback should be sent jointly to info@snomed.org and contact.orphanet@inserm.fr.

Feedback should include any issues relating to implementation, suggestions for future content inclusion or general comments regarding the map.

Technical Notes

RF2 package format

The RF2 package convention dictates that it contains all relevant files, regardless of whether or not there is content to be included in each particular release. Therefore, the package contains a mixture of files which contain both header rows and content data, and also files that are intentionally left blank (including only a header record). The reason that these files are not removed from the package is to draw a clear distinction between:

1. ...files that have been deprecated (and therefore removed from the package completely), due to the content no longer being relevant to RF2 in this or future releases, and
2. ...files that just happen to contain no data in this particular release (and are therefore included in the package but left blank, with only a header record), but are still relevant to RF2, and could therefore potentially contain data in future releases.

This allows users to easily distinguish between files that have purposefully been removed or not, as otherwise if files in option 2 above were left out of the package it could be interpreted as an error, rather than an intentional lack of content in that release.

"First Time Release"

As this is the first time that we are publishing the Orphanet Map package, per the current RF2 packaging standards the Delta, Full + Snapshot files are identical. This assists those users who update using Delta files, as they are relative to a blank Snapshot as there is no previous published release package.