



SNOMED CT to Orphanet Map package Production Release

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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. The first release, in October 2021, was based on an agreed priority set, with new concepts for rare diseases, as defined in Orphanet, added to SNOMED CT. Work is now complete between SNOMED CT and rare disease content in the Orphanet nomenclature, taking into account the differences in structure and rules of each terminology. A maintenance and updating plan is now in place to ensure alignment is maintained overtime.

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
- Guideline on the electronic exchange of health data under Cross-Border Directive 2011/24/EU Patient Summary. Release 3.3, June 2023: https://health.ec.europa.eu/system/files/2023-10/ehn_guidelines_patientsummary_en.pdf

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 with the plan 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 in 2021 contain 5,652 SNOMED CT and Orphanet concepts/terms.

An updated Agreement was signed in March 2020 signalling both organisations commitment to maintaining and updating the content and map. A further 1025 Orphanet entries were reviewed for inclusion in the map.

This October 2023 release now represents an alignment between SNOMED CT and Orphanet for Rare Diseases where in scope and a maintenance and updating process is being put in place to ensure this alignment is maintained as changes take place in both terminologies and the map updated accordingly.



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.



Design

Content Changes for 2023 Production Release

The key changes made to content for the 2023 Production Release:

1. Update to map for content that has been inactivated in SNOMED CT.
2. Removal of map from production release for entries that have been marked obsolete, deprecated or non-rare in Orphanet.
3. Total New mapped concepts this release - 346
4. Total inactivated mapped entries this release - 187

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 2023.

The version of SNOMED CT used is the July 2023 International Release.

Source for Map

The source data is a subset of 6555 SNOMED CT concepts.



Direction of Map

The direction of the map is from SNOMED CT July 2023 International Release to [Orphanet July 2023](#).

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.com

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.

RF2 Delta files removed from the package

Delta files have been removed from both International and Managed Service release packages, and are now being removed from all Derivative products in line with this new Standard.

The Delta files have therefore been removed from this 2023 Orphanet Release package. Please contact support@snomed.org if you have any questions about these improvements.

