

# 201942 The Value of SNOMED CT Description for the Mapping of Chinese Rare Diseases Ontology

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## Summary

The author and the team built an ontology based on SNOMED CT design for rare diseases in China. They did cross mappings for all the concepts in order to knowledge linking for CDSS. The descriptions of SNOMED CT helped the mapping between terminologies.

## Audience

Clinical, Technical

## Learning Objectives

1. Situations of Rare Diseases in China;
2. New development of Chinese Rare Diseases Ontology;
3. Function of knowledge linking for CRDO.

## Abstract

Rare diseases (RDs) are rare, but rare disease patients are numerous. More and more interests are paid on RDs in China. A list containing 121 rare diseases (Rare Diseases List, RDL) was published by five governmental departments of China, including National Health Commission, Ministry of Science and Technology, Ministry of Industry and Information Technology, National Medical Products Administration, State Administration of Traditional Chinese Medicine, in June of 2018. National Rare Diseases Registry System, containing 128 rare diseases now, is a platform for the data registry of rare diseases in China. Since China is not a member country of SNOMED CT, NRDRS obtained a first affiliate license of SNOMED CT in China. The author and his team were authorized to use this license to build up an ontology named CRDO, Chinese Rare Diseases Ontology, based on the 208 rare diseases from RDL and NRDRS.

One of the use cases of CRDO is knowledge linking. The Each concept in CRDO has an English name, which was used for linkage with Radiology Gamuts Ontology (RGO) that is a domain application ontology for radiology and a knowledge resource for radiology diagnosis. Since the concepts were mapping with SNOMED CT, which contained a large number of synonyms in SNOMED CT description, the synonyms in SNOMED CT were also used for augmenting the



matching between CRDO and RGO for those that don't have an exact match (indirect exact match) between concepts in CRDO and RGO.

There are 92 CRDO concepts exactly mapping with RGO, in which 77 were direct and 15 indirect. The method was also used in the mapping between CRDO and other terminologies, including MeSH, ICD-10, ICD-11 and OMIM. There were 66 direct and 12 indirect for mapping to MeSH, 9 direct and 12 indirect for ICD-10, 59 direct and 3 indirect for ICD-11 and 49 direct and 12 indirect for OMIM.

The plenty of synonyms in SNOMED CT is a wonderful bridge between terminologies.

#### Reference Documentation

1. Drug Project: <https://confluence.ihtsdotools.org/display/IAP/Reference+Documentation+-+Drug+Model>
2. Substance Project: <https://confluence.ihtsdotools.org/display/IAP/Reference+Documentation+-+Substances>