

# Title: Interoperability of Disease Concepts in Clinical and Research Ontologies - Contrasting Coverage and Structure in the Disease Ontology and SNOMED CT

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## Audience

Developers and users of SNOMED CT interested interoperability with research datasets.

## Summary

While SNOMED CT is used to record diseases in health records, the Disease Ontology is used instead in many research projects. We investigate the coverage and structure of these two ontologies as a contribution to interoperability between the two domains.

## Abstract

**Objectives.** To contrast the coverage of diseases between the Disease Ontology (DO) – a publicly available ontology of diseases (about 7000 diseases) used in many research projects – and SNOMED CT (over 100,000 clinical findings), and to compare the hierarchical structure of the two ontologies.

**Methods.** We establish a reference list of mappings. We characterize unmapped concepts in DO semantically and structurally. Finally, we compare the hierarchical structure between the two ontologies. The August 2016 release of the Disease Ontology and the March 2016 release of SNOMED CT (US Edition) were used in this investigation.

**Results.** Overall, 4478 (65%) the 6931 Disease Ontology concepts are mapped to SNOMED CT. The cancer and neoplasm subtrees of the Disease Ontology account for many of the unmapped concepts. The most frequent differentiae in unmapped concepts include morphology (for cancers and neoplasms), specific subtypes (for rare genetic disorders), and anatomical subtypes. Unmapped concepts usually form subtrees, and less often correspond to isolated leaves or intermediary concepts.

**Conclusion.** This detailed analysis of the gaps in coverage and structural differences between the Disease Ontology and SNOMED CT contributes to the interoperability between these two ontologies and will guide further validation of the mapping.

## References

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