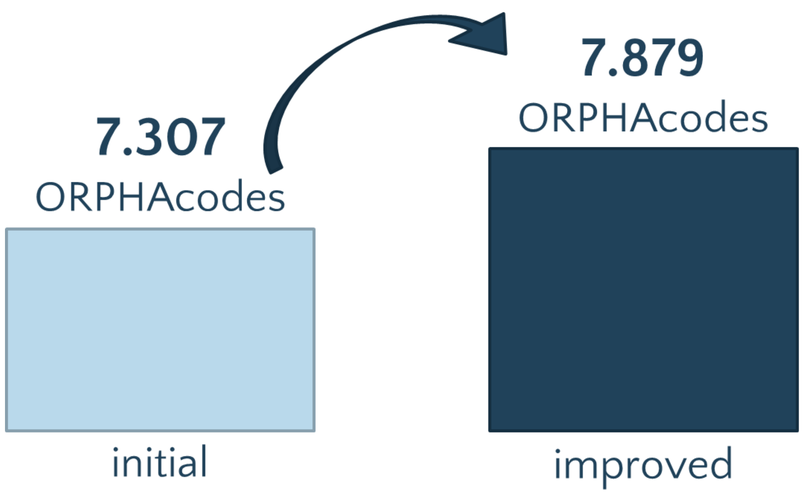
ORPHAcodes, which are unique identifiers for **rare diseases**, have been prepared as a new non-standard **vocabulary**.

Health Orphans – Rare diseases on their way to OMOP and into clinical trials

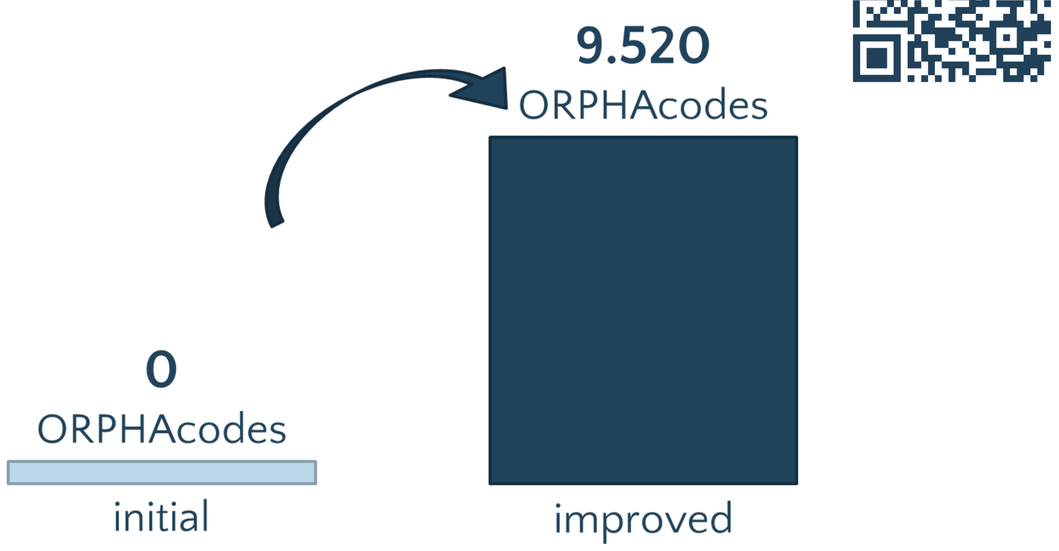
Background: Around 446 million people worldwide suffer form rare diseases. One major challenge is the lack of medical expertise and reliable information, partly due to inadequate coding for these conditions. ORPHAcodes serve as unique identifiers for rare diseases. To establish ORPHAcodes as a new non-standard vocabulary, it was essential to enrich their mapping to SNOMED, ICD10GM, and ICD10WHO.

Results

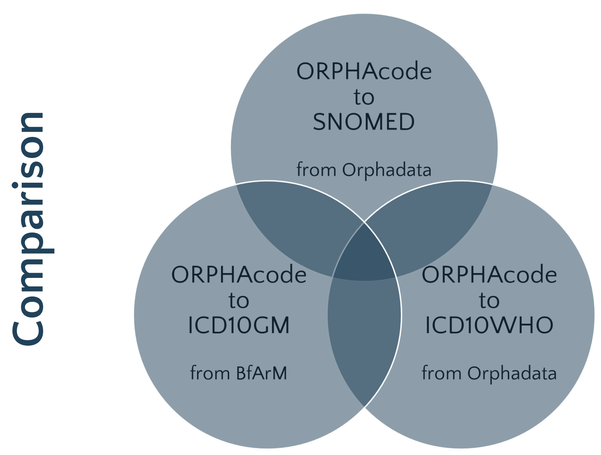
to SNOMED Mapping



to OMOP Mapping



Methods



Enrichment

USAGI

Preparation

- *concept*
- *concept_relationship*
- *synonyms*

▶ GitHub-Issue #1010

Limitation: The medical validation of the additional mapping within *Usagi* is still pending. Would you like to enhance the evaluation and validate the results further? If so, please get in touch with us.

OHDSI

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