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 from 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**
  - Initial: 7,307 ORPHAcodes
  - Improved: 7,879 ORPHAcodes

- **to OMOP Mapping**
  - Initial: 0 ORPHAcodes
  - Improved: 9,520 ORPHAcodes

**Methods**

- Comparison
- Enrichment
- 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.