Terminologies in the World Health Organisation Family of International Classifications (WHO-FIC)

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SUMMARY

As part of its strategic workplan, the World Health Organization Family of International Classifications (WHO-FIC)Family Development Committee (FDC) commissioned work to understand better the relationship between WHO-FIC classifications and terminologies on the one hand and other health terminologies on the other. Members of the FDC and others developed a series of local, national, and international use cases that demonstrated: 1) the parallel use of WHO-FIC classifications and terminologies and other terminologies (with automated and manual approaches to mapping, both nationally and locally); 2) the potential enrichment of WHO-FIC classifications and terminologies with other terminologies; and 3) the potential enhancement of the practical application of WHO- FIC classifications and terminologies through the use of other terminologies. The use cases illuminated issues around the concurrent use of different terminologies, leading to recommendations that ensure optimal deployment in the WHO-FIC context.

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1 BACKGROUND

This discussion paper describes work conducted by members of the World Health Organization Network for the Family of International Classifications (WHO-FIC) Family Development Committee (FDC) to clarify, as part of its strategicworkplan, the relationship between terminologies within the WHO-FIC, traditionally referred to as reference, derived and related classifications [1], and other terminologies used within health care.

2 INTRODUCTION

Health and healthcare data are paramount for WHO to fulfil its global mission. The reality of data collection has changed dramatically over the past 30 years in the sense that data on health and health care is no longer documented, collected and reported solely through aggregating terminologies (or classifications). The number of terminology ecosystems has expanded enormously, and the average number of terminologies within a single terminology ecosystem has also increased. For those terminology ecosystems to thrive, one must understand how their constituent terminologies relate.

WHO Member States have clearly expressed their wish to clarify the relationships between terminologies by adopting the resolution on ICD-11 in May 2019. The resolution included an article requesting that the WHO Director-General should "*implement a regular updating process for ICD-11 and to develop further and implementthe family of disease- and health-related classifications, with the International Statistical Classification of Diseases and Related Health Problems as the core classification linked to other related classifications, speciality versions and terminologies*". [2]

3 TERMINOLOGY ECOSYSTEMS

Healthcare terminologies are "artefacts that provide the standardized meaning of human language expressionsused in oral or written communication within a given domain. Multiple terminologies have been developed in multiple contexts of use. Together, these terminologies can form a terminology ecosystem'. Sometimes such ecosystems can be highly fragmented and serve different functionalities, such as reimbursement, monitoring, mortality and morbidity statistics, quality assessment, registries, or screening programmes" [3]

Multiple terminologies have been developed in various contexts of use, forming different terminology ecosystems. At the same time, policymakers are confronted with challenges in interoperability when trying to digitalize health care and exchange health information. Often, health information is fragmented in silos. To overcome this fragmentation, policymakers are redesigning their terminology ecosystems and formulating semantic strategies to facilitate health information exchange and interoperability between systems (FAIRprinciples). Part of this process involves the selection of a terminology or a set of terminologies.

4 HISTORY AND FUNCTIONS OF TERMINOLOGIES

While the historical origins of health classifications are overwhelmingly statistical [4], contemporary applicationsof ontologies, terminologies, and classifications range from clinical operations and reimbursement to translational

research [5] and biomedical discovery [6]. These changes in usage patterns imply commensurate changes in modern classifications' content, structure, and underlying architecture. Lists of diseases served adequately for 16th-century mortality statistics [7], but enumerated lists cannot serve the spectrum of current demands. Hierarchical terminologies with permanent codes, such as ICD-10, expanded classification capabilities to accommodate aggregation within coding levels (e.g., all children of a 3-digit code). This enabled such systemsto function for reimbursement and clinical operations, though with granularity and detail limited to enumeratedstatistical rubrics.

The functions of a clinical nomenclature, where detailed characterization of a clinical case advanced in ICD-11, with the adoption of post-coordinated expressions and the ICD-11 foundation component. Detailed clinical sentences comprising multiple stem codes enhanced with modifiers and qualifiers are possible in ICD-11. More detail is possible directly using the foundation component of ICD-11 in conjunction with uniform identifiers. Additionally, an arbitrary number of statistical tabulations or linearizations from the underlying semantic network of all concepts and their multi-parented relationships, the Foundation, permits flexibility in the way statistical aggregations occur for different use cases, such as public health, clinical practice, translational research, and of course the primary linearization - Mortality and Morbidity Statistics (ICD-11 MMS). As the Foundation, including ICD, ICF and ICHI and a set of extension codes, increasingly populates the completeness of the definitions and relationships for each of the increasing number of entries, particularly multi-hierarchical parenting, WHO Family of Classifications based on the Foundation become better suited for sophisticated data science inferencing on a larger spectrum of clinical and biomedical data. Such enhancements improve the promise of WHOFIC classification serving a broad range of terminology and classification use cases.

The application areas highlighted previously - statistics, clinical operations, reimbursement, translational research, and biomedical discovery - have guided the structural evolution of contemporary terminologies, permitting the grouping of terminologies into structural types, such as lists, hierarchical terminologies, and computable ontologies. The same application areas have required terminologies to fulfil a range of different purposes or functions, which has resulted in terminologies exhibiting particular properties, such as multiple parenting or specification of attributes. These properties can be used to group terminologies also into functional types such as interface terminologies, reference terminologies, and aggregating terminologies.

- Interface terminologies refer to collections of terms used in written and oral communication within a group of users, for example, in a data entry form. Entries in interface terminologies need to be described in terms of the natural language they belong to and by dialect, time, clinical speciality and professional group. The implicit meaning within terms drawn from interface terminologies generally manifests through linkages to reference terminologies.
- Reference terminologies describe the meaning of terms of a domain, together with the properties or attributes of the objects that these terms denote, in a relatively neutral sense, i.e., uncommitted to any specific context.
- Aggregating terminologies are systems of non-overlapping classes in single hierarchies used for data aggregation and ordering. Typically, these involve classifications such as the WHO-FIC.

5 CATEGORIZING CLASSIFICATIONS

The World Health Organization Family of International Classifications (WHO-FIC) provides a suite of health-related classifications, traditionally referred to as reference, derived and related classifications. The application area of these aggregating terminologies covers the entire healthcare spectrum.

5.1 REFERENCE CLASSIFICATIONS

Reference classifications are the global standards for health information, clinical documentation, and statistical aggregation. They include:

- the International Classification of Diseases and Related Health Problems (ICD)
- the International Classification of Functioning, Disability and Health (ICF), and
- the International Classification of Health Interventions (ICHI)

All three reference classifications are available as linearizations in the WHO-FIC Foundation. The WHO-FIC Foundation is a multidimensional collection of interconnected entities representing diseases, disorders, injuries,external causes, signs and symptoms, functioning descriptions, interventions, and extension codes. Each reference classification has a reference guide outlining rules for using the content.

5.2 DERIVED CLASSIFICATIONS

Derived classifications were extensions (deepenings) of reference classifications developed for use within a speciality setting and derived from the common reference classification. The content of derived classifications has been integrated into the WHO-FIC Foundation and replaced by the opportunity of generating special linearizations, if necessary.

The derived classifications in WHO-FIC were the following:

- International Classification of Diseases for Oncology, 3rd Edition (ICD-O-3)
- The ICD-10 Classification of Mental and Behavioural Disorders: Clinical Descriptions and diagnostic guidelines
- The ICD-10 Classification of Mental and Behavioural Disorders: Diagnostic Criteria for Research
- International Classification of External Causes of Injury (ICECI)

5.4 RELATED CLASSIFICATIONS

Related classifications are complementary to reference and derived classifications, and cover speciality areas not otherwise described by these WHO-FIC reference or derived classifications. Content in these related classifications can overlap with reference or derived classifications. In that case, the reference or derivedclassifications prevail, in that respective order. The related classifications in WHO-FIC are the following:

- International Classification of Primary Care, 2nd edition (ICPC-2)
- Technical aids for persons with disabilities - Classification and terminology (ISO9999)
- The Anatomical Therapeutic Chemical Classification System with Defined Daily Doses (ATC/DDD)
- International Classification for Nursing Practice (ICNP)
- Verbal autopsy standards: ascertaining and attributing causes of death

6 CURRENT LIMITATIONS REGARDING CATEGORIZING CLASSIFICATIONS AND TERMINOLOGIES

In contemporary terminological practice, attempts to characterize terminologies by structure or function become increasingly difficult. For example, a terminology that forms a computable ontology also comprises a plethora of classifications and a myriad of potential lists. And a single terminology can, at the same time, exhibit interface, reference, and aggregating properties.

The quest continues; the aspiration for a unified, universal terminology that can comprehensively address all healthcare necessities continues to be a challenging and unattained objective. Still, to support information exchange and system interoperability, there remains a need to understand the interplay between different terminologies – the purpose of this work.

7 PRACTICAL USE OF MULTIPLE TERMINOLOGIES

To better understand the issues around the use in practice of multiple terminologies, the WHO-FIC FDC, in collaboration with the WHO, drew together a team to help source and collate a wide range of use cases that they felt would adequately cover the problem space: 1) the parallel use of WHO-FIC terminologies andother terminologies (with automated and manual approaches to mapping, both nationally and locally); 2) the potential enrichment of WHO-FIC terminologies with other terminologies; and, 3) and the potential enhancement of the practical application of WHO-FIC terminologies through the use of other terminologies.

7.2 PARALLEL USE OF WHO-FIC TERMINOLOGIES AND OTHER TERMINOLOGIES

7.2.1 USE CASE – SPAIN, NLP MAPPING OF REFERENCE TERMS TO BOTH SNOMED CT AND ICD

The Hospital Clínic-University of Barcelona seeks to bring automated coding of health problems as close as possible to the clinical process. A coded health problem list will be built online with the help of Natural Language Processing (NLP) and machine learning (ML) algorithms supervised and monitored by expert coders in a regional scenario combining a University Hospital and twenty Primary Care centres, covering half a million inhabitants in the city of Barcelona.

This work builds on previous experience using NLP for the automated coding into ICD-10 of discharge reports [8, 9]. The present study will draw on textual clinical descriptions in referral letters written by family physicians and in clinical notes written by hospital specialists, using NLP for additional automated coding into SNOMED-CT and ICD-11.

Once the triple codes have been assigned automatically to health problems, the three terminologies will be explored for similarities and differences, revealing a set of 'natural' mappings between SNOMED-CT and ICD. Experts will compare these mappings with pre-existing mappings [10, 11], and any differences will be subjected to further investigation.

7.2.2 USE CASE – NETHERLANDS, MANUAL MAPPING OF REFERENCE TERMS TO BOTH SNOMED CT AND ICD

In the Netherlands, the Diagnosis Thesaurus is used in most hospitals to document clinical diagnoses and diagnosis-treatment combinations in medical records and the National Hospital Discharge Registry.[12] The thesaurus covers all medical specialities and comprises an extensive, searchable collection of over 35,000 preferred clinical terms; healthcare providers use these to document patient contact. In this respect,the Diagnosis Thesaurus can be regarded as interface terminology.

Concepts within the Diagnosis Thesaurus are linked to the nearest equivalent concept within SNOMED CT (drawing on, for example, definitions within the Diagnosis Thesaurus and semantic tags within SNOMED CT). In this context, SNOMED CT, therefore, acts as a reference terminology. Concepts within the Diagnosis Thesaurusare also linked to concepts within an aggregating terminology, ICD-10-NL. All linkages (between interface terminology and reference terminology and between interface terminology and aggregating terminology) were done manually. The de facto linkages between reference terminology (SNOMED CT) and aggregating terminology(ICD-10-NL) were subsequently validated against the internationally available SNOMED-ICD-10 2014 mappings.

7.2.3 USE CASE – UK, NATIONAL MAPPING SNOMED CT TO ICD-10

The United Kingdom (UK) has mandated SNOMED CT as the terminology to support the direct management of care in clinical and pathology systems across the NHS. The SNOMED CT UK Edition contains UK extensions that provide terms specific to UK requirements, and its implementation is underway across devolved nations, although most advanced in England.

In 2018, the primary care sector in England transitioned to SNOMED CT for term choice, interoperation between GP practices and for national data collections. IT systems either use SNOMED CT exclusively or map from legacy Read or local codes to SNOMED CT. Secondary care Electronic Patient Record (EPR) systems are generally less mature with respect to SNOMED CT, and the pace of adoption is variable.

NHS Digital provides national cross-maps from SNOMED CT to ICD-10 and the UK procedure classification OPCS-4. These support local clinical coding processes and semi-automation. AI and machine learning pilots are exploring how data could be used. NHS Digital undertook a small exploratory project to investigate how NLP technology might support the clinical coding process and the more general process of assigning SNOMED CT, ICD-10, and OPCS-4 codes to clinical text. The project built an open-source NLP pipeline to annotate syntheticdischarge letters using SNOMED CT, ICD-10, and OPCS-4 and began to assess how well the NLP system performed its annotation of the discharge letters. It was intended to document and publish a report of the project's findings, but the project is paused due to priority COVID-19 work.

The UK is developing a national ecosystem of clinical information standards that will provide richness in recording and sharing patient interactions across clinical settings and flow this information to secondary-use consumers. Several clinical data set collections have been redesigned to request data using SNOMED CT and, in England, the Admitted Patient Care Commissioning Data Set has been updated to allow the optional data flow of SNOMED CT (including diagnosis, procedure) for national reporting in parallel with existing mandatory data flows using ICD-10 and OPCS-4.

NHS Digital's strategy is to reduce the burden of data collection on front-line staff by collecting 'raw' data from the NHS, which is then curated, coded, and classified centrally by NHS Digital. Clinical data will continue to be represented in SNOMED CT to support interoperability and data sharing. The classifications (OPCS-4, ICD-10 and, in the future, ICD-11) will be vital in classifying patient episodes.

The successful uptake of ICD-11 in the UK depends on its ability to embed it within the information ecosystem. Rich point-of-care information in patient records must be linked with ICD-11 and the procedure classification, but developing and maintaining this linkage require considerable resources. Many countries share this requirement. The flow of SNOMED CT data and classifications as part of national collections allows one to explore possible ways of achieving the strategy to reduce the burden and determine whether developments in AI and machine learning can assist this work and inform the implementation of ICD-11.

7.2.4 USE CASE – CANADA, LOCAL MAPPING SNOMED CT TO ICD

The Canadian Institute for Health Information (CIHI) maintains the standards for morbidity data reporting in Canada and maintains, distributes, and supports the application of ICD-10-CA (the Canadian modification of ICD-10). The Canadian Classification of Health Interventions (CCI), developed by CIHI, is the national standard for classifying healthcare procedures. All acute care hospitalization data and most emergency department data are coded with ICD-10-CA and submitted to CIHI. Statistics Canada supports the mortality application of ICD-10. Note that ICD-9 is used in physician billing in Canada.

Canada Health Infoway, rather than CIHI, is the National Release Centre (NRC) for SNOMED CT and supports the Canadian adaptation, SNOMED CT CA.

Currently, there are no comprehensive maps from SNOMED CT to ICD-10-CA. To date, mapping has been done on a case-by-case basis (e.g., subset lists mapped by CIHI or hospitals mapping their subsets), jeopardizing a standardized approach. With the proliferation of EHR implementations (which frequently use SNOMED CT in Canada), CIHI has been approached to address this gap and to provide support for stakeholders doing their mapping. EHR vendors include maps from SCT to ICD-10/ICD-10-CM but not ICD-10-CA or CCI. Facilities are not always aware of the versioning of SNOMED CT embedded in their implementations (SNOMED CT or SNOMED CT CA). Some jurisdictions have procured a clinical terminology tool to provide clinicians with a way to enter a clinical diagnosis at the point of care. Terms used in the tool map to ICD-10-CA and SNOMED CT. These maps are proprietary and have not been validated by CIHI.

Agreed maps from SNOMED CT to ICD-10-CA that can be used consistently across Canada in the electronic health record and electronic medical record systems would be of great benefit. Such maps would facilitate the re-use of point-of-care data captured by clinicians, enable best coding practices, reduce coder burden, and potentially expedite quality data submissions to CIHI from different healthcare settings.

CIHI has procured a mapping tool to enable a semi-automated solution for creating maps from SNOMED CT to the Canadian health classifications. SNOMED International currently provides maps from SNOMED CT to ICD-10, and CIHI is using these as a starting point to develop maps for ICD-10-CA. Mapping to CCI is a more complex task, and for the time being, priority will be given to developing SCT to ICD-10-CA maps.

7.3 POTENTIAL ENRICHMENT OF WHO-FIC TERMINOLOGIES WITH OTHER TERMINOLOGIES

7.3.1 USE CASE – LOINC AND NPU; THE CONCEPT OF SIGNPOSTING

In addition to the use cases presented previously that describe the parallel use of WHO-FIC terminologies and other terminologies, a further use case involves the potential enrichment of WHO-FIC terminologies with other terminologies, i.e., to capture clinical data in health records using the WHO-FIC terminologies, but to augment this data using other terminologies to add further detail. The level of detail found in other terminologies may extend beyond that needed for statistical reporting. In this case, other terminologies may be used to provide additional detail to the WHO-FIC terminologies. Note that the general availability of other terminologies is an important consideration in the context of the international information ecosystem. Any gaps in availability may negatively impact data exchange across organizational and geographical boundaries.

This differs from other use cases as, although the resulting information may be connected, there is no direct interaction between the terminologies themselves, and any links are maintained by an external artefact such as an information model. It is important to note that the added detail goes beyond the scope of the more general Extension Codes in the Foundation, such as specific anatomical location. The content within these other terminologies may be related to content within the Foundation but do not form part of the Foundation itself.

For example, a medical specialist treating a young female patient with a very specific iron overload disease (ICD- 11 5C64.10) called Pantothenate-kinase-associated neurodegeneration asked her blood to be examined. A light microscope blood test revealed the presence of acanthocytes, and electrophoresis determined the level of pre-beta lipoprotein in her blood serum (very low). This helped determine the specific form of iron overload disease, HARP syndrome. Now, these two tests' results and methods may constitute information that is very relevant for

the clinical geneticist treating this patient and should therefore be documented in her electronic health record. In this case, two other terminologies, LOINC [13] and NPU [14], both offer relevant additional detail. For example, LOINC offers 7789-1 (Acanthocytes [Presence] in Blood by Light microscopy) while NPU offers: NPU17074 (B?Acanthocytes; arb.c.(proc.)).

This demand for documentation of additional detail beyond the level of detail that is offered through the WHO-FIC classifications Foundation Component is preferably gatewayed through the Foundation Component, but without actually incorporating additional terms in the WHO-FIC classifications. This is the concept of signposting.

7.4 POTENTIAL ENHANCEMENT OF THE PRACTICAL APPLICATION OF WHO -FIC TERMINOLOGIES THROUGH THE USE OF OTHER TERMINOLOGIES

It requires communication between the WHO-FIC Foundation Component and the target terminology. The operationalization requires further clarification.

7.4.1 USE CASE – ICNP AND DRGS

The final use case differs from previous use cases as, rather than exploring the interface between different terminologies, it considers the possible enhancement, through the use of other terminologies, of the application of WHO-FIC terminologies in practice. WHO-FIC terminologies are also used in many countries to inform patient classification systems, such as Diagnosis Related Groups (DRGs), where an ICD diagnosis is used with other information, such as procedures, sex, or age, in the assignment of patients into categories or groups that can be used for example to support reimbursement. Reimbursement for nursing care is often included as part of a fixed daily ‘room rate’. However, this has been shown to underestimate actual nursing costs [15]. The use of nursing data in combination with DRGs has been shown to improve the explanation of variance across a range of indicators, sometimes dramatically [16]. The use of a WHO-FIC terminology (in this case, ICD), in combination with another terminology (such as the International Classification for Nursing Practice, which is, in fact, a Related Classification within WHO-FIC), therefore, has the potential to increase the specificity of DRGs. This demonstrates the wider utility of other terminologies in the context of the WHO-FIC terminologies.

9 CONCLUSIONS

Work commissioned by the WHO-FIC FDC has resulted in use cases that demonstrate

1. the parallel use of WHO-FIC terminologies and other terminologies (with automated and manual approaches to mapping, both nationally and locally);
2. the potential enrichment of WHO-FIC terminologies with other terminologies; and,
3. and the potential enhancement of the practical application of WHO-FIC terminologies through the use of other terminologies.

There is a strong case to support the use of WHO-FIC terminologies to report health data and facilitate comparison; there is also a case to support the use of other terminologies to record clinical data and to facilitate data exchange across organizational and geographical boundaries. The use cases have demonstrated the value of WHO-FIC and other terminologies in local, national, and international contexts. However, they have also revealed a degree of duplication in attempts to harmonize both terminology sets and a need for coordination.

11 RECOMMENDATIONS

1. WHO-FIC reference classifications should be used whenever possible for recording and reporting purposes, locally, nationally, and internationally.
2. For specific purposes, and where they exist, derived classifications[1] could also be used; Where reference and derived classifications are unable to capture point-of-care information at the level of specificity needed, other terminologies (ideally related classifications/terminologies) should be used to complement reference and derived classifications. Still, they should not replace reference and derived classifications for reporting purposes. The related classifications and terminologies must be mapped to the reference classifications[2] to ensure unambiguous and comparable terms, concepts and classifications.
3. WHO-FIC should determine an approach to recognizing a set of other terminologies that might complement the use of reference and derived classifications, to ensure consistent use, prevent duplication of effort and better coordinate mapping activity.
4. WHO-FIC should explore further the relationship between the Foundation and relevant content from recognized terminologies.
5. WHO-FIC should develop and put in place robust governance arrangements with strong strategic oversight to support the integration of recognized related classifications or terminologies with the Family, to ensure consistency in application, to prevent duplication of effort, and to better coordinate mapping activity.
6. The principles that guide the governance of WHO-FIC classifications might also apply to other recognized terminologies, thereby informing any recognition process:
    - Recognized terminologies should be available internationally and useful across a wide range ofcultures.
    - Recognized terminologies should be made available as an international public good on the same basis as the reference classifications.
    - Recognized terminologies should be maintained regularly with a transparent and inclusive approach to governance.
    - Stakeholders should be willing to contribute to an ongoing shared mapping effort coordinated by WHO-FIC to prevent duplication of effort and inconsistencies in reporting.
    - The results of any coordinated mapping effort between recognized terminologies and WHO-FIC terminologies should be made available under the same terms of the WHO-FIC reference
    2 WHO: levels of mapping or alternatively integration rather than mapping will need to be discussed in the Network

[1] WHO: In the context of ICD-11, ICF and ICHI, a derived classification would be derived from a common foundation, e.g. a speciality linearization

[2] WHO: levels of mapping or alternatively integration rather than mapping will need to be discussed in the Network

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