Improved Handling of Synonymous SNOMED CT Concepts used in HL7 Version 3 by the Example of the Von Willebrand Disease

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Abstract

Background: In the context of synonymous concepts, errors and term duplications in SNOMED CT are likely. This affects in particular clinical ideas with multiple types of display, like the example of the Von Willebrand disease.
Objectives: It is required to ensure a common and correct interpretation of medical terms. Thus, it is necessary to distinguish between the clinical ideas of disease, disorder and syndrome in the Von Willebrand context. To perform a critical examination of the associated hierarchical order in SNOMED CT and above that, to follow the given rules of correct terminology binding.
Methods: The synonymous concepts problem is analyzed in reference to the Von Willebrand context in the SNOMED CT International Version 31012015. Improvements of handling are formulated based on the valid SNOMED CT compositional grammar and the SNOMED CT Editorial Guidelines. Terminology binding is examined regarding the prerequisites given in the "Using SNOMED CT in HL7 Version 3 Implementation Guide, Release 1.5
Results: Many Von Willebrand concepts in SNOMED CT are not fully defined but primitive, which means that their defining characteristics are not sufficient to distiguish synonymous meanings. Improved handling of synonymous concepts is proposed by the installation of a broader grouper concept and an adaption of the SNOMED CT hierarchical structure to avoid misinterpretations and duplications.
Conclusions: The terminology binding between the SNOMED CT semantics in the Von Willebrand context and HL7 V3 seems technically well described. It has been shown, that the problematic handling of synonymous terms is assigned to hierarchical structure problems out of SNOMED CT.

Keywords

SNOMED CT; HL7 Version 3; synonym; Von Willebrand disease

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1 Introduction

SNOMED CT is a terminology with approximately 350.000 concepts and over 800.000 synonyms [1]. The function of a synonym is the representation of a term that can be used to select a concept with most similar meaning. A concept may have plenty of synonyms. This allows the end-users to choose preferred terms that refer to special clinical ideas.

In the context of synonymous concepts, errors and term duplications in SNOMED CT are likely. Problems were described at an early stage by the merging of SNOMED RT and United Kingdom's National Health Service's Clinical Terms Version 3 into SNOMED CT. During the creation of the terminology individual modelers were allowed to merge several concepts from the source terminologies into one concept. A single concept then became the name of the SNOMED CT concept and the other(s) became a synonym. Significant errors that result from this process were due to wrong modeler assigning, e.g. a more general concept as a synonym of a more specific concept [2].

Given the problems of synonymous concepts, other relating errors like inconsistent hierarchical connections and term duplications occur. This affects in particular clinical ideas with multiple types of display, like the example of the Von Willebrand disease. As this disease has focus on the blood system, it touches a very broad range of medical disciplines and a great variety of stakeholders.

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Terminology users continue to encounter confusion about which concepts and codes to select when using Von Willebrand disease, Von Willebrand disorder or Von Willebrand syndrome because of unclear distinctions. Above that, end users describe problems and uncertainties to undertake correct classifications of several synonymous Von Willebrand 2 subtypes. Related duplications and overlaps can be found in the current SNOMED CT concept model. An unclear term interpretation and a lack of semantic interoperablity may imply negative consequences on clinical safety that should be avoided.

An uninterrupted electronic communication of medical content is made possible by the interaction of semantic and syntactic standards. The semantic standard SNOMED CT, which describes medical content, needs a "syntactic partner" capable of receiving such content and ensuring the exchange of data. Matching syntactic standards are in this context HL7 Version 2, HL7 Version 3 and HL7 CDA [3]. Markwell et al. (2008) refers to this specification as terminology binding. Terminology binding describes the preparation of a compound between terminology elements and an information model [4]. Above that, it is important to mention that HL7 V2 and V3 both provide syntax and semantics. This means on the one hand, that HL7 standards contribute to the definition of the meaning of the information exchanged, but on the other, there may insecurities what kind of semantic expression has to be chosen.

A key factor of SNOMED CT is its big expressivity. This may lead to cases where overlaps occur with semantics that may also be represented by an information model such as the HL7 Reference Information Model (RIM). For example, a single SNOMED CT coded expression can represent a meaning that the HL7 RIM could also represent using a combination of several coded attributes or classes. Clear rules and guidance on these overlaps are needed to minimize ambiguity and erroneous interpretation.

This paper describes an approach for improved handling of complex synonymous terms in SNOMED CT taking over the Von Willebrand example and to provide solutions for correct terminology binding that the semantic meaning inside of the information model may be unambigous.

1.1 Disease, disorder and syndrome

Clinical findings represent observation results. The use of the terms disease, disorder and syndrome are connected to the description of abnormal clinical states but differ in their definitions:
A disease is a definite pathological process having a characteristic set of signs and symptoms. It may affect the whole body or any of its parts, and its etiology, pathology, and prognosis may be known or unknown [5]. A disorder is defined as a derangement or abnormality of function; a morbid physical or mental state [6]. A syndrome is a group of signs and symptoms that occur together and characterize a particular abnormality or condition [7].

Whether or not this designation is accurate, distinctions are definitely made in certain contexts. It is Parkinson disease, not Parkinson disorder; sleep disorder, not sleep disease. Specialist literature shows that disease is mostly connected to the influence of extrinsic factors. Disorder itself has a focus on condition of the individuals.

1.2 The Von Willebrand disease

The von Willebrand disease is defined as a bleeding disorder caused by a deficiency of the large and complex glycoprotein "Von Willebrand factor (VWF)". VWF itself is an essential factor in blood clotting. The Von Willebrand disease is an inherited bleeding disorder characterized by incomplete penetrance and variable expressivity.

There are 3 different types of von Willebrand disease:

Type 1: The predominant defect is a partial quantitative deficiency. There are two subtypes of Type 1, Type 1a and Type 1b. Most patients with type 1 Von Willebrand are able to live normal lives with only mild bleeding issues. The predominant effect is a quantitative deficiency of VWF.

Type 2: The predominant defect is a partial qualitative deficiency. Patients with type 2 are at greater risk for complications and experience mild to moderate bleeding. These individuals may suffer worse bleeding in the case of infection, surgery or pregnancy.

Type 3: The predominant defect is a complete deficiency (quantitative and qualitative). Patients with type 3 are at risk for severe bleeding as well as internal and gastrointestinal bleeding.

The Type 2 Von Willebrand, which represents 20-25% of all cases, has four different subtypes:

- Type 2A is the most common subtype. In Type 2A the blood platelets do not bind together well
- Type 2B is the next most common. In Type 2B, the VWF binds to platelets in the bloodstream, instead of binding at the site of the injury to the blood vessel
- Type 2N is much rarer. VWF also helps to carry around factor VIII in the blood and stabilize it so it can take part in the formation of a solid clot. In Type 2N the VWF does not transport factor VIII.
- Type 2M is an extremely rare sub-type. In Type 2M, binding of the VWF to platelets is impaired.

The etiology of the Von Willebrand disease is, that it can be acquired or hereditary. The acquired form is also called Pseudo-Von Willebrand's disease or platelet-type. It differs from the three hereditary forms (Type 1-3), that have already been described. In this context, the term hereditary is often used synonymously with congenital. Acquired Von Willebrand's is preferentially called "acquired Von Willebrand's syndrome" in medical specialist literature. The term von Willebrand's disease is often reserved for the congenital/ inherited form [8, 9].

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2 Methods

We analyze the synonymous concepts problem referring to the Von Willebrand context in the SNOMED CT International Version 31012015. The CliniClue-XPlore-Browser Version 2012.8.0270 and the IHTSDO SNOMED CT Browser Version 1.0 are used for content display.

Potential improvements in the handling of synonymous concepts are to be formulated according to the SNOMED CT compositional grammar. The terminology binding is checked regarding the prerequisites given in the "Using SNOMED CT in HL7 Version 3 Implementation Guide, Release 1.5" [10].

3 Results

3.1 Positioning of Von Willebrand related concepts in SNOMED CT

The SNOMED CT International Edition 20150131 uses | 128105004 | von Willebrand disorder | as a grouper concept for Von Willebrand related concepts. | 128105004 | von Willebrand disorder | is defined as:

|128105004 |von Willebrand disorder| **is a**
|64779008|blood coagulation disorder| **is a**
|362970003|disorder of haemostatic system| **is a**
|64572001|disease| **is a**
|404684003|clinical finding|

All of the von Willebrand disorder related concepts are child concepts of | 128105004 | von Willebrand disorder | ; with the Fully Specified Name (FSN) | 128105004 | von Willebrand disorder (disorder) |.

Furthermore, | 128105004 | von Willebrand disorder | contains 10 different synonyms.

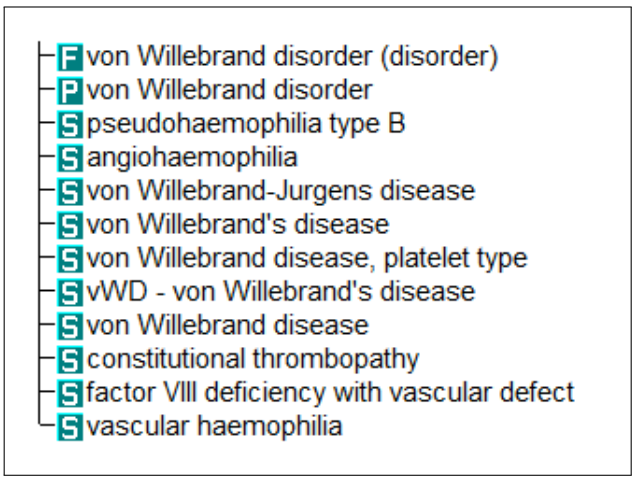

Figure 1: FSN and synonyms of | 128105004 | von Willebrand disorder | .

It is noted that there are incompatible synonyms attached to 128105004 | von Willebrand disorder (disorder) | such as | von Willebrand disease, platelet type | . The latter provides an inappropriate specification on the given hierarchical level.

| 128105004 | von Willebrand disorder | has 29 child concepts.

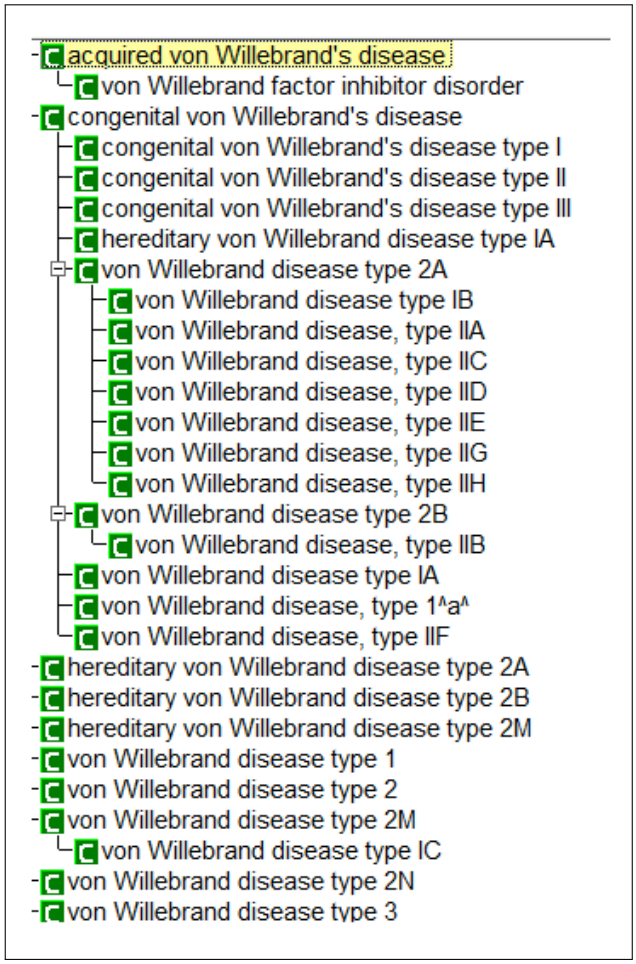

Figure 2: Child concepts of | 128105004 | von Willebrand disorder | .

In SNOMED CT, the parent concept of all kinds of Von Willebrand disorder,- disease or syndrome concepts is | 128105004 | Von Willebrand disorder | . | 128105004 | Von Willebrand disease is a synonym of | 128105004 | Von Willebrand disorder | and it has no parent or grouper concept function.

All of the child concepts of | von Willebrand disorder | , except | congenital von Willebrand´s disease | , are not fully defined but primitive which means that their defining characteristics are not sufficient to uniquely distiguish its meaning from other similar concepts.

Figure 3 shows the concept model of | 128107007 | von Willebrand disease type 2 | in a detailed view.

The concept is primitive, with the FSN at | 128107007 | von Willebrand disease type 2 (disorder) | , the synonym | 128107007 | hereditary von Willebrand disease type 2 | and has no child concepts.

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Figure 3: Summary of | 128107007 | von Willebrand disease type 2 (disorder) | .

In the 201501131 Version of SNOMED CT

- | 359729006 | von Willebrand disease type 2M (disorder) |
- | 359732009 | von Willebrand disease type 2N (disorder) |
- | 359711001 | hereditary von Willebrand disease type 2A (disorder) |
- | 359717002 | hereditary von Willebrand disease type 2B (disorder) |
- | 359725000 | hereditary von Willebrand disease type 2M (disorder) |

are child concepts of | 128105004 | von Willebrand disorder | . Above that, the upper five concepts are originally kinds of | von Willebrand disease type 2 | , which hasn´t been modelled in SNOMED CT so far.

3.2 Improvement approach on Von Willebrand related concept structuring

The proposed design tries to improve the structure of the Von Willebrand type disorders and to simplify navigation, especially for the stakeholders. We take a four step,top-down approach with regard to the hierarchical structure of SNOMED CT.

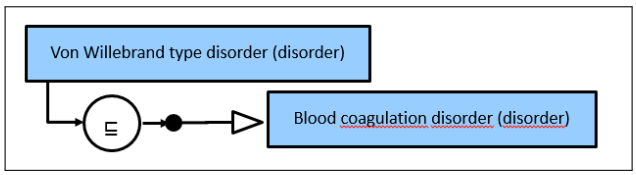

Figure 4: Concept model design for | von Willebrand type disorder (disorder) | .

Step 1: A new and broader grouper concept named | von Willebrand type disorder (disorder) | shall be created. The first draft of a concept model for | von Willebrand type disorder (disorder) | is shown in Figure 4

| Von Willebrand type disorder (disorder) | is displayed as a child concept of | 64779008 | Blood coagulation disorder (disorder) | via an "is_a" relationship.

Step 2: Switch the hierarchy from | 128105004 | von Willebrand disorder (disorder) | to | von Willebrand type disorder (disorder) | . This will be a primitive grouper concept and all existing children will need to be given a stated "is a "relationship to this new grouper concept.

The SNOMED CT Editorial Guide January 2015, Section 7.6 Naming Convention for the disorder hierarchy states that "*In the disorder hierarchy, the word 'disorder' in singular should be used. When the concept is a general grouper of disorders of a body system, body site, or other broad general category, the word 'disorder' should be used in preference to 'disease' for the FSN. This rule in favour of 'disorder' over 'disease' applies only to broad groupers, and is not applied at 'leaf' level*"[11].

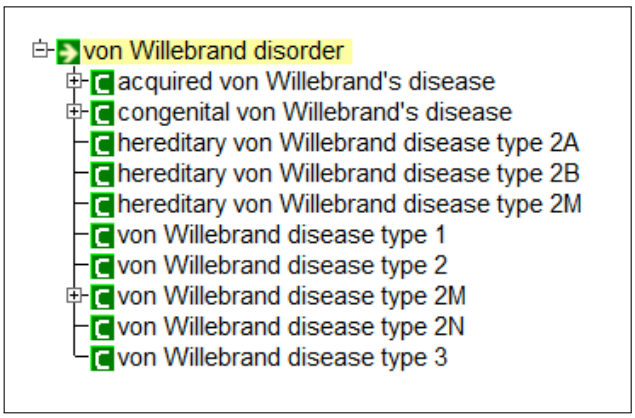

Figure 5: 128105004 | von Willebrand disorder (disorder) | as a grouper concept.

Figure 5 shows the original hierarchy design with | 128105004 | von Willebrand disorder | , with the FSN | 128105004 | von Willebrand disorder (disorder) | , as a grouper concept:

Figure 6 shows the new concept von Willebrand type disorder (disorder) with the switched hierarchy from | 128105004 | von Willebrand disorder (disorder) | :

As von Willebrand type disorder is expected to be a primitive concept, it shall be pointed out that normally the use of intermediate primitives is prohibited [11]. There are few exemptions when the use of intermediate primitives may be allowed, as

- There is no other option and the concept is clinically necessary
- The impact of adding the concept in question has been fully explored and understood
- The impact is deemed manageable and a plan for management has been determined

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In this special case, the use of an intermediate primitive seems to be acceptable, as the specific child concepts are known or searchable, and that the switch of the hierarchy to Von Willebrand type disorder has additionally been proposed by IHTSDO terminology experts.

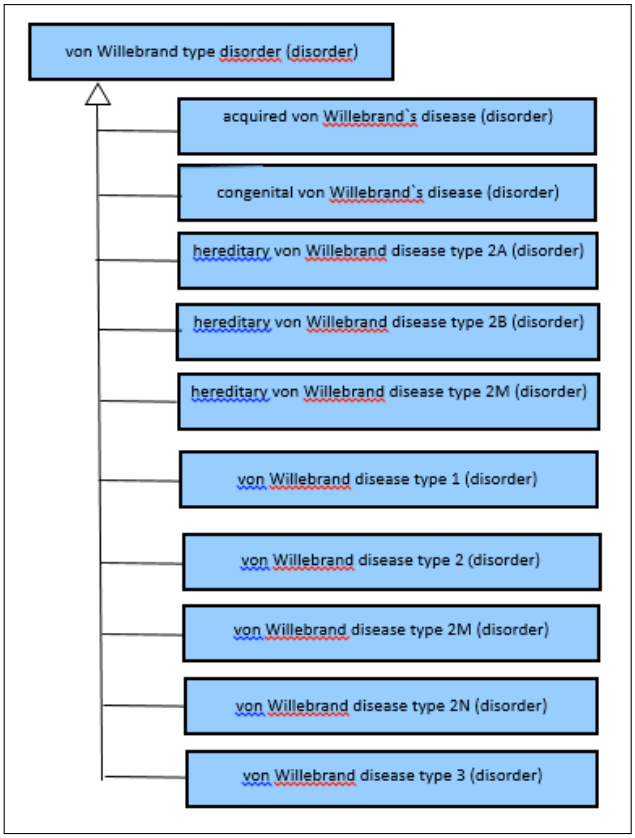

Figure 6: Von Willebrand type disorder (disorder) with subhierarchy in diagramming style.

Step 3: Retire | 128105004 | von Willebrand disorder (disorder) | as a grouper concept

Step 4: Re-organization of subtypes of | 128107007 | von Willebrand disease type 2 (disorder) | We provide a preliminary design of six new concept models for "is_a" relationships in Figure 7

In this context and to avoid duplications in future SNOMED CT editions, it is also possible to retire:

- | 359729006 | von Willebrand disease type 2M (disorder) |
- | 359732009 | von Willebrand disease type 2N (disorder) |
- | 359711001 | Hereditary von Willebrand disease type 2A (disorder) |
- | 359717002 | Hereditary von Willebrand disease type 2B (disorder) |
- | 359725000 | Hereditary von Willebrand disease type 2M (disorder) |

from the current position (see Figure 2), as they are now defined as child concepts of | 128107007 | von Willebrand disease type 2 | .

The described design tries to improve the structure of the von Willebrand type disorders and to simplify navigation, especially for the stakeholders.

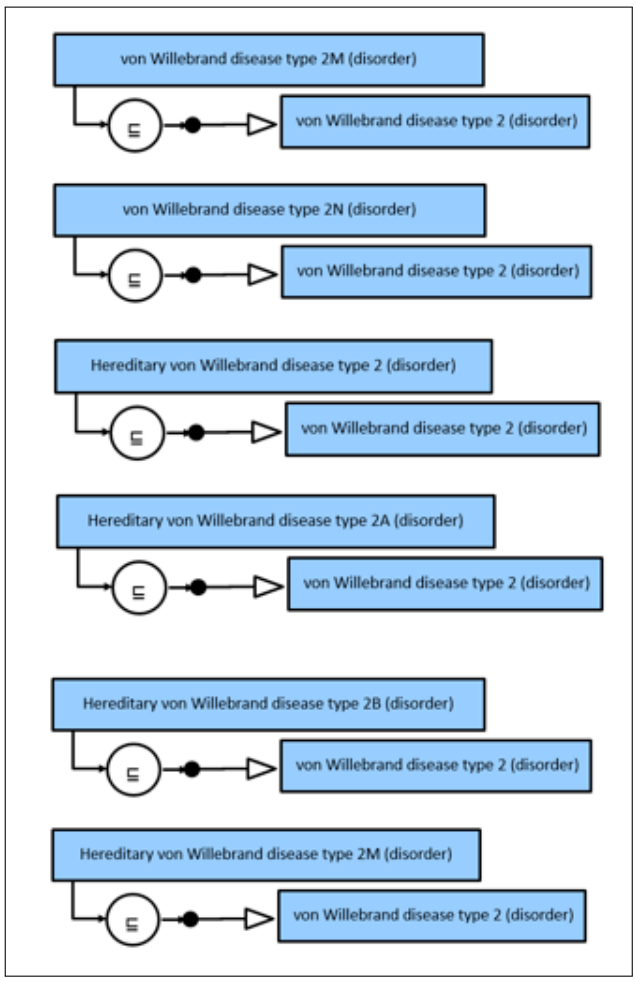

Figure 7: Possible "is_a" relationships of von Willebrand disease type 2 (disorder).

3.3 Terminology binding in the Von Willebrand context

The HL7 implementation guide "Using SNOMED CT in HL7 Version 3" offers recommendations how to provide semantic interoperability through the harmonized interaction of terminology and information model.

In HL7 V3, observation is displayed as an isolated event, whereas an HL7 V3 condition is an ongoing event. Symptoms and findings, e.g. the Von Willebrand disease, are observations. There is a distinction in SNOMED CT between "clinical findings" and "diseases" where the latter is necessarily a pathological condition. The SNOMED CT clinical finding/disease distinction is orhtogonal to the HL7 observation/condition distinction. This means that

```
<observation classCode="OBS" moodCode="EVN">
   <code code="ASSERTION" codeSystem="2.16.840.1.113883.5.4"/>
     <text> von Willebrand disease type 2 </text>
       <value xsi:type="CD" code=" 128107007 | von Willebrand disease type
         2 |" codeSystem="2.16.840.1.113883.6.96">
          <displayName value=" von Willebrand disease type 2 "/>
       </value>
</observation>
```

Figure 8: Assertion of the clinical finding "Von Willebrand disease type 2".

```
<act classCode="DOCSECT" moodCode="EVN">
  <code code="8646-2" codeSystem="2.16.840.1.113883.6.1" codeSys
     temName="LOINC"/>
     <title>Hospital Admission Diagnosis</title>
        <text>Hospital admission diagnosis </text>
           <actRelationship typeCode="COMP">
             <observation classCode="OBS" moodCode="EVN">
                <code code="ASSERTION" codeSystem="2.16.840.1.113883.5.4"/>
      <value xsi:type="CD" code="128107007|von Willebrand disease type 2 |"
         codeSystem="2.16.840.1.113883.6.96">
        <displayName=" von Willebrand disease type 2"/>
      </value>
    </observation>
  </actRelationship>
</act>
```

Figure 9: Context dependent assertion of the diagnosis "Von Willebrand disease type 2".

a SNOMED CT finding or disease can be an HL7 observation or condition.

The distinction between an HL7 observation and HL7 condition is made by setting the Act.classCode to "OBS" or "COND". The distinction between a SNOMED finding and SNOMED disease is based on the location of the concept in the SNOMED CT hierarchy [12].

Figure 8 shows the assertion of the clinical finding "Von Willebrand disease type 2":

A diagnosis like Von Willebrand disease has clinical meanings as it is the result of a process whereby for example symptoms are determined to describe the condition afflicting a patient.

Figure 9 shows the context dependent (hospital admission related) assertion of the diagnosis "Von Willebrand disease type 2":

A concern is something that a clinician is particularly interested in and wants to track.in terms of patient history. As a defeciency of the blood clotting system and possible bleeding issues, all information on Von Willbrand disease presence are important in this case.

The HL7 Patient Care Technical Committee is developing a formal model for the tracking of conditions. In that model, a problem is wrapped in an act with a new Act.classCode "CONCERN". The focus is on the use of SNOMED CT, where the Patient Care condition tracking model is the definitive source for the structure of a problem list [10].

4 Conclusions

As there is a great variety of SNOMED CT concepts connected to the Von Willebrand disease context, meaningful concept structuring inside of the terminology is one of the major goals. We tried to construct an appropriate hierarchical order concerning the concept | 128105004 | von Willebrand disorder | . In this context and according to stakeholder needs, a revised and functional organization of subtypes of | 128107007 | von Willebrand disease type 2 (disorder) | has been provided as well.

Some irregularities have to be taken into account, like f. e. incompatible synonyms or duplications. The risk of addressing this problem is that | 128105004 | von Willebrand disorder (disorder) | has currently the function of a grouper concept. If the descriptions are changed to a new grouper concept like | 128105004 | von Willebrand type disorder (disorder) | this may cause miscoding. Above that, it is possible that the end users may choose this grouper concept rather than a more granular child concept.

Other relating problems and exemptions, that have to be taken into account in the future, raise from the terms "congenital" and "hereditary" in connection to von Willebrand diseases. As specialist literature states that the von Willebrand disorders or – diseases type 1 up to 3 are all hereditary it may be irritating why it is necessary to have a concept | 359729006 | von Willebrand disease type 2M (disorder) | on the one hand and | 359725000 | hereditary von Willebrand disease type 2M (disorder) | on the other. There is a need for further refinement of the hierarchy respective these attributes [9]. Regarding these

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problems, it seems to be appropriate to check the current hierarchy first in terms of duplication. It needs to be clarified, whether there is a need to keep | 359725000 | hereditary von Willebrand disease type 2M (disorder) | if | 359729006 | von Willebrand disease type 2M (disorder) | may mean exactly the same. Additionally, it should be addressed whether "congenital" and "hereditary" may have a synonymous meaning together with the Von Willebrand disease, and if they do, duplicate concepts may be retired. The idea is to keep the more common and frequently used term "congenital Von Willebrand disease" or "hereditary Von Willebrand disease".

The terminology binding between the SNOMED CT semantics in the Von Willebrand context and HL7 V3 seems to be well described. It has been shown, that the problematic handling of synonymous terms is assigned to hierarchical structure problems out of SNOMED CT. HL7 standards rely on the "model of meaning" that states a common understanding of a certain context. It is crucial to provide fully defined SNOMED CT concepts in the Von Willebrand context whenever possible and to structure the assigned hierarchical order.

The Von Willebrand example seems to be suitable to show how small semantic differences lead to big effects. It is a demanding task to identify and to work on comparable synonymous issues in SNOMED CT for effecient content improvement, with the overall objective to facilitate terminology usage and – in the end – patient safety.

4.0.1 Acknowledgements

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