

PROPOSAL— REMOVING THE DISORDER DISTINCTION

26/3/2019

Matt Cordell – Australian Digital Health Agency

Keith Campbell - Veterans Health Administration

PROBLEM STATEMENT

Since the creation of SNOMED CT, there has existed an apparent partition between Disorder and other Clinical Findings.

Finding	Disorder
<ul style="list-style-type: none">▪ May be normal (but not necessarily)▪ May exist only at a single point in time (e.g. a serum sodium level)▪ Cannot be temporally separate from the observation (one cannot observe them and say they are absent, nor can they be present when they cannot be observed)▪ Cannot be defined only in terms of an underlying pathological process that is present, when the observation itself is not present	<ul style="list-style-type: none">▪ Always and necessarily abnormal▪ Necessarily have an underlying pathological process▪ Have temporal persistence (may be under treatment, in remission, or inactive, even though they are still present)▪ May be present as a propensity for certain abnormal states to occur, even when treatment mitigates or resolves those abnormal states

[^] <https://confluence.ihtsdotools.org/pages/viewpage.action?pageId=71172245>

The existence of such a distinction has introduced several problems:

- A duplicity of concepts such as:
 - 425558002| Azoospermia (disorder) | vs 48188009| Azoospermia (finding) |
 - 211402004 | Abrasion of foot (disorder) | vs 781488002 | Abrasion of foot (finding) |
 - 403165009| Cat scratch injury (disorder) | vs 283112002 | Cat scratch - wound (finding) |

This duplicity manifests as duplicates for clinical end users when searching fields limited to clinical findings.

- Extensive inconsistency across the terminology for otherwise similar concepts:
 - 422411000|Surgical scar (disorder)| vs 249433005|Laryngectomy scar (finding)|
 - 300131008|Lump in ear canal (disorder)| vs 299704007|Lump on face (finding)|
 - 102031000119109|Paratesticular mass (disorder)| vs 271860004|Abdominal mass (finding)|
 - 164277005|On examination - dysphonia (disorder)| vs 162890008|On examination - dyspnea (finding)|
 - 238969001|Burning scrotum (disorder)| vs 36031001|Burning feet (finding)|
 - 74788000| Tongue absent (disorder)| vs 246916009| Absent eyeball (finding)|
- The designation of a finding as normal or abnormal can be subjective and dependent on cultural or moral positions and these can also change over time. The most obvious areas associated with this are the findings associated with sexual behaviour and identity, and mental health. Additionally, the reference point of what is normal can change with age. Normal behaviour for a child can be considered abnormal in an adult. There are many disorders that are considered a normal consequence of aging.¹

The problems described above, illustrate limited capacity for the *Understandable* and *Reproducible* principles to be applied.

¹ <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5540438/>

PROPOSED SOLUTION

The *Usefulness* of identifying certain concepts as “necessarily abnormal” within the terminology is of limited (if any) value. Any assertion as to the normality of a clinical finding should be at the discretion of authors and viewers of a particular clinical record, within the context of that record. For example, a finding or diagnosis of Azoospermia is always preceded by a semen analysis, and associated with infertility. Though would be a desired outcome following a vasectomy compared when investigating *difficulty conceiving*.

This proposal therefore recommends that the concept 64572001| Disease (disorder)| be inactivated without replacement.

SCOPE OF WORK

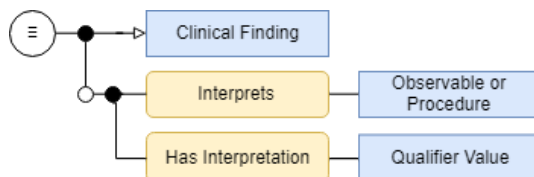
The immediate scope of work proposed here (concepts affected in parenthesis) is to:

1. Retire the concept 64572001| Disease (disorder)|; (1)
2. The FSN of all subtypes is updated to use the semantic tag “(finding)”; (75,363)
3. Create 404684003 | Clinical finding (finding) | subtype axioms for concepts with no other stated parent concept (16,768);
4. Finally, any duplicates revealed upon classification are to be resolved (TBD).

SNOMED International has already implemented such a change for the allergy content in January 2019 release.

IMPACT AND OTHER IMPLICATIONS

Rather than requiring a hierarchy to identify a subset of content that is “necessarily abnormal”, it may be more useful to consider a partition of findings that are the outcome of specific clinical investigation. Concepts of a pattern such as



The concept 441742003| Evaluation finding (finding)| (almost) provides this function. However, there numerous concepts that could adhere to this modelling pattern that are not currently subtypes.

Work in this area might also support better management of what does and doesn't go into the Situation with explicit context (SWEC) hierarchy. For example evaluation findings could be “present or absent” with assertions about the absence of clinical conditions (the other findings) being Findings with explicit context.

E.g. 299742001| Moro reflex absent (finding)| & 247079003| Red reflex absent (situation)|

However, the distinction between SWEC and Findings is difficult and beyond scope.

Administrative findings may also prove a useful partition.