Guidelines on writing clinical description for Condition Phenotypes

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Contents

PhenotypeLibrary is part of HADES

This document is a DRAFT guidance.

Note (caution): We are not re-writing a medical textbook or chapter. The content is designed to help the phenotyper, i.e. the person who develops and evaluates the cohort definition. If the phenotyper is building cohorts using Atlas - the clinical description is expected to guide the entry event criteria, inclusion rules and exit criteria.

0.1 Start with one or more Authoritative source(s):

- We ask that you start by referencing (e.g., by providing an internet hyperlink to) an authoritative source that guided your understanding of the phenotype's clinical description. This is expected to be rewritten by clinical subject matter experts in the field of your phenotype. It almost uses medical Jargon like 'Etiology', 'Pathogenesis', 'Evaluation', 'Treatment', 'Prognosis'.
- Because OHDSI subscribes to Open Science and international collaboration - we ask that you try to prioritize authorititaive sources that are publicly and openly available. Example of such authoritative sources include NIH National Library of Medicine Stat Pearls [StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2022 Jan-. Available from: https://www.ncbi.nlm.nih.gov/books/NBK430685/] (https://www.ncbi.nlm.nih.gov/books/NBK430685/)
- We then expect a short/succinct abstraction of the referenced authoritative source, but written in the context of helping a phenotyper. It is a reasonable expectation for the phenotyper to review the authoritative source referenced - however, by abstracting out the key ideas, we have observed, it helps make phenotype development more efficient and less likely to miss key ideas.

0.2 Summary of Authoritative source:

We ask that you summarize using the following headings/components:

- **Overview:** Key summary of the clinical concept.
- **Presentation:** observations from a clinician who sees persons with the clinical concept, including signs and symptoms.

- **Diagnostics Evaluation:** clinical workup done to confirm or refute the presence of the clinical concept in a person, including diagnostic procedures and measurements. (Formerly we used to call this Assessment.), These are the things a clinician would like to do to determine etiology, disease state / extent, and disease complications.
- **Therapy Plan:** treatment performed to manage the risks to a person from the clinical concept, including therapeutic procedures, surgeries, and drug exposures (where applicable). Pharmacologic, non-pharmacologic clinical actions.
- **Prognosis:** statements that indicate the duration the clinical concept is expected to persist in a person. Three questions are expected to be addressed:
- Can the person fully recover marking the end of an episode i.e., is full recovery possible, or is it considered life long? Yes/No
- If it's not lifelong and recovery/end of condition is possible – on **median**
  - What is the minimum (median) duration that persons are expected to have the disease? Example: neutropenia is observed on a median for a minimum of 7 days.
  - What is the maximum (median) duration that persons are expected to have the condition? Example: neutropenia is observed on a median for a maximum of 365 days.
- If end of condition is possible, can a new episode of the condition independently re-occur in the same person after full recovery from prior independent episode?
- **Disqualifiers:**
- A dis-qualifier is a mutually exclusive idea i.e. if this construct is present in the person then they cannot, at the same time, have the phenotype of interest. A mere presence of a clinical construct that can predispose/put a person at higher/lower risk for the phenotype is not a dis-qualifier.
- Occasionally, we may call something that is known to cause the outcome a dis-qualifier e.g., genetic conditions or malignancies. For this, we need a strong a priori knowledge with high certainty, e.g. 70 to 80% of ppl with this cystic fibrosis will develop the chronic lung disease - then cystic fibrosis may be a dis-qualifier for chronic lung disease. To be a dis-qualifier, it has to be stated explicitly as a dis-qualifier in the clinical description (consider adding a reference). A good justification/rationale must be given to allow this construct as a dis-qualifier.
- All other ideas that don't meet the two guidelines above, but are of interest to the phenotyper to be a dis-qualifier - have to be empirically evaluated and numerically shown to have an impact along with clinical description describing why it should be a dis-qualifier + what would happen to the phenotype of interest if it is not a dis-qualifier.
- In general we expect not more than a few dis-qualifiers. If we have more than 4 dis-qualifiers, we need to think hard as to why we have many dis-qualifiers. Dis-qualifiers are usually observed in some temporal relationship (i.e., an event about a person that should or should not occur during a specified temporal period with respect to cohort start date), e.g. should not have a diagnosis of liver cirrhosis at any time prior to acute hepatic failure.
- **Strengtheners:**
- conditions, when present, increase the likelihood that the person has the clinical idea. The observation of established risk factors in the baseline period, or treatments for the condition after index date are considered strengtheners. A positive re-challenge, where the phenotype is observed to start again after exposure to the risk factor.