

Guidance on writing clinical description for Condition Phenotypes

Gowtham A. Rao

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Contents

1 Condition: [Name of condition mentioned above.]	1
1.1 Overview	2
1.2 Synonyms	2
1.3 Presentation	2
1.4 Diagnostic Evaluation	2
1.5 Differential Diagnoses	2
1.6 Treatment Plan	2
1.7 Prognosis	2
1.8 Exclusions	2
2 Ambiguity	2
2.1 Subtypes	2
2.2 Patient Profile	3
2.3 References	3

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Note: This document is a DRAFT guidance.

We ask you to write a brief clinical description. This description is aimed at professionals with a medical terminology, informatics, or clinical background, so as to facilitate shared understanding of the target clinical idea being modeled in the phenotype. It is common to use medical technical terms related to general pathophysiology in the clinical description, but we try to avoid delving into molecular biology. The reader is not expected to have a background in research methods and can be a clinician. We are not rewriting a medical textbook or chapter. This document may also be an input to 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 and provide justifications for the concept set expression, and the rules for entry event criteria, inclusion rules, and exit criteria. Please use the following structure.

1 Condition: [Name of condition mentioned above.]

This will serve as the title for the clinical idea.

1.1 Overview

Write a concise description of the condition. Keep it to about 3 sentences.

1.2 Synonyms

List other names for the condition. This is to help build the concept set expression.

1.3 Presentation

Detail the signs and symptoms of the condition.

1.4 Diagnostic Evaluation

Describe the assessment criteria for Condition. Include suitable tests and potential results where relevant.

1.5 Differential Diagnoses

Provide a list of conditions that could potentially be mistaken for the condition.

1.6 Treatment Plan

Outline the standard treatment plan for the condition, using bullet points. If multiple treatment options exist, treat each as a separate bullet point. Indicate whether the treatment plan is the current or a previous standard that has since evolved.

1.7 Prognosis

Write a brief paragraph on the prognosis for a patient diagnosed with Condition, distinguishing between short-term (within the first 3 months after diagnosis) and long-term (1 year or more after diagnosis) prognoses.

1.8 Exclusions

List any conditions or treatments that must be ruled out at the time of diagnosis.

2 Ambiguity

Identify any conditions with similar names to the condition but that represent different clinical ideas. Do not include subtypes of the condition.

2.1 Subtypes

Identify any subtypes of the condition.

2.2 Patient Profile

Provide one or more short case narratives of a typical patient with the clinical concept.

2.3 References

Include at least 10 references that were used in creating this report, ensuring they are indexed on PubMed and compatible with a reference manager.