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Health informatics — Conceptual framework for patient findings and problems in terminologies

Informatique de santé — Cadre conceptuel pour les constats des patients et les problèmes de terminologies

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Foreword

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In other circumstances, particularly when there is an urgent market requirement for such documents, a technical committee may decide to publish other types of document:

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Attention is drawn to the possibility that some of the elements of this document may be the subject of patent rights. ISO shall not be held responsible for identifying any or all such patent rights.

ISO/TS 22789 was prepared by Technical Committee ISO/TC 215, *Health informatics*.

Introduction

Enormous investment has occurred over the last two decades in creating health care terminological systems. Further expansion or additions are likely. Many developments overtly or covertly share features of a common model of clinical findings.

The objective of this Technical Specification is to express a core **categorial structure** of clinical findings based upon existing schemes including the World Health Organization *ICD-10*, NHS *Clinical Terms Version 3*, International Health Terminology Standards Development Organisation's *SNOMED Clinical Terms* and the WONCA 2001 *International Classification of Primary Care*. The development of a Technical Specification will help existing users to develop a **mapping** or cross-walks between one scheme and another and provide a robust logical framework for construction of new areas or consistency for updated versions of existing terminological systems. The model describes the underlying principles of clinical findings and important **semantic link**s referencing a set or sets of **characterizing concept**s such as anatomy and causative organisms. It utilizes ISO 17115, which has been designed to describe such patterns for **concept** representation in a particular domain.

Within this Technical Specification, the following notation is used:

A **bold** notation has been used where references are made to terms defined in Clause 2.

An item enclosed by single angle brackets < > refers to a category that can be specialized to various **concept**s, as required.

An item enclosed within the text by single accolades { } identifies a **semantic link**.

Health informatics — Conceptual framework for patient findings and problems in terminologies

1 Scope

1.1 Main purpose

The purpose of this Technical Specification is to specify a **categorial structure**, within the subject field of patient findings and problems, by defining a set of common **domain constraint**s for use within terminological systems including a classification, coding scheme, coding system, reference terminology and clinical terminology.

Clinical findings are **concept**s that are recorded in clinical records and can describe any state observed directly or indirectly concerning a patient and their relationship with the environment. This Technical Specification is focused on a sub-population of these findings concerning *in vivo* descriptions of state (structure and function) directly related to the patient. This class of **concept**s includes:

— diseases, which may be defined as a state caused by a known or assumed pathological process impairing the normal physiological function and/or anatomical structure affecting all or part of a patient, where a specific pathological change is caused by a defined mechanism;

— findings of state or function (including normal findings) observed directly relating to a patient.

This Technical Specification describes a concept system detailing a domain constraint of sanctioned characteristics each composed of a semantic link and an applicable characterizing category.

The potential uses for this **concept**ual framework are to:

— support developers of new terminology systems concerning patient findings and problems;

— support developers of new detailed content areas of existing terminology systems concerning patient findings and problems to ensure conformance;

— facilitate the representation of patient findings and problems using a standard core model in a manner suitable for computer processing;

— provide a **concept**ual framework for the generation of **compositional concept representation** of patient findings and problems;

— facilitate the **mapping** and improved **semantic correspondence** between different terminologies by proposing a core specification for patient findings and problems;

— provide a core model to describe the structure of patient findings and problems, and facilitate improved **semantic correspondence** with information models.

1.2 Target groups

The target groups for this Technical Specification are:

— developers of terminology systems concerning patient findings and problems;

— developers of information systems that require a structured framework of **concept**s to facilitate implementation;

— IT specialists, analysts and epidemiologists who require common models of knowledge to facilitate analysis of current and legacy data from one or more information systems;

— clinicians and coders to provide greater consistency in structure and organization when entering and retrieving data using one or more terminology systems;

— managers and administrative personnel in providing a benchmark by which to judge terminology solutions: as to whether the potential options will deliver compatibility with legacy data and future proofing to emerging terminology products.

1.3 Topics considered outside the scope

Topics considered outside the scope of this Technical Specification include:

— a comprehensive **categorial structure** for clinical findings;

— laboratory findings (including biochemical and histological results);

— signal findings (including the output from imaging and electrophysiological tests);

— social findings;

— the absence of findings, e.g. absent bowel sounds, the absence of a knee reflex, are not included within this Technical Specification as it might prejudice subsequent attempts at standardizing the modelling of such instances;

— an exhaustive list of all possible **characterizing concept**s that could be used to describe clinical findings.

2 Terms and definitions

For the purposes of this document, the following terms and definitions apply.

2.1
associative relation
relation between two **concept**s (2.9) having a non-hierarchical thematic connection by virtue of experience

[ISO 1087-1:2000, definition 3.2.23]

EXAMPLE An **associative relation** (2.1) exists between the **concept**s (2.9) “education” and “teaching”, “baking” and “oven”.

NOTE Other relations between **concepts** (2.9) are: hierarchical, generic, partitive, sequential, temporal and causal.

2.2
categorial structure
minimal set of **domain constraint**s (2.11) for representing **concept system**s (2.10) in a subject field

[ISO 17115:2007, definition 2.4.5]

2.3
category
formal category
concept (2.9) represented by a **formal definition** (2.12)

NOTE Adapted from ISO 17115:2007, definition 2.5.3.

2.4
characterizing category
formal **category** (2.3) whose specialization by a **domain constraint** (2.11) is allowed to be used as **characterizing concept** (2.5) in a particular context

[ISO 17115:2007, definition 2.3.3]

EXAMPLE <INFECTIOUS_ORGANISM>={bacterium, virus, parasite}, in the context of "Infection that hasCause INFECTIOUS_ORGANISM".

2.5
characterizing concept
concept (2.9) that is referenced by a **semantic link** (2.17) in a **composite characteristic** (2.7)

[ISO 17115:2007, definition 2.2.2]

EXAMPLE "Bacterium" in the construct "Disease that hasCause Bacterium" and "Yellow" in the construct "SkinLesion that hasColor Yellow".

2.6
characteristic
abstraction of a property of an object or of a set of objects

[ISO 1087-1:2000, definition 3.2.4]

NOTE **Characteristic**s are used for describing **concept**s (2.9).

2.7
composite characteristic
qualifier
formal representation of a **characteristic** (2.6)

EXAMPLE hasCause Bacteria; Location = LeftUpperLobeOfLung.

NOTE 1 Adapted from ISO 17115:2007, definition 2.2.1.

NOTE 2 Can be compared to an attribute-value pair.

2.8
compositional concept representation
intensional definition of a **concept** (2.9) using as delimiting characteristics one or more **composite characteristic**s (2.7)

[ISO 17115:2007, definition 2.4.1]

NOTE Allows inference and subsumption within a compositional system. Usually expressed in a formalism, such as description logic.

2.9
concept
unit of knowledge created by a unique combination of characteristics

[ISO 1087-1:2000, definition 3.2.1]

2.10
concept system
system of concepts
set of **concept**s (2.9) structured according to the relations among them

[ISO 1087-1:2000, definition 3.2.11]

2.11
domain constraint
sanction
rule prescribing the set of **sanctioned characteristic**s (2.15) that are valid to specialize a concept representation in a certain subject field

EXAMPLE "Infection possibly hasLocation SkeletalStructure" explains that an infection in a certain context can be located in a structure that is a kind of skeletal structure.

NOTE 1 Adapted from ISO 17115:2007, definition 2.3.2.

NOTE 2 The rule describes the set of **sanctioned characteristic**s (2.15) by combining the **semantic link** (2.17) and the **characterizing category** (2.4) it links to, possibly by enumeration of the concepts in the **characterizing category** (2.4).

NOTE 3 Different levels of sanctioning are possible, e.g. conceivable, sensible, normal, usuallyInTheContextOf, necessary.

2.12
formal definition
definition within a **formal [concept representation] system** (2.13)

NOTE Adapted from ISO 17115:2007, definition 2.4.3.

2.13
formal [concept representation] system
set of machine processable definitions in a subject field

[ISO 17115:2007, definition 2.5.1]

2.14
mapping
assigning an element in one set to an element in another set through **semantic correspondence** (2.16)

[ISO 17115:2007, definition 2.6.1]

2.15
sanctioned characteristic
formal representation of a type of **characteristic** (2.6)

[ISO 17115:2007, definition 2.3.1]

EXAMPLE 1 performedUsing <INSTRUMENT>; hasLocation <BodyPartOrImplantedDevice>.

EXAMPLE 2 "CauseOfInflammation canBe set{bacteria, virus, parasite, autoimmune, chemical, physical}", where "canBe" is the **semantic link** (2.17), and "set{bacteria, virus, parasite, autoimmune, chemical, physical}" is the **characterizing category** (2.4).

NOTE A **sanctioned characteristic** (2.15) is typically made up of a combination of a **semantic link** (2.17) and a **characterizing category** (2.4), and can be used in **domain constraint**s (2.11).

2.16
semantic correspondence
measure of similarity between two concepts

[ISO 17115:2007, definition 2.6.2]

2.17
semantic link
formal representation of a directed **associative relation** (2.1) or partitive relation between two **concept**s (2.9)

[ISO 17115:2007, definition 2.2.3]

EXAMPLE hasLocation (with inverse isLocationOf) and isCauseOf (with inverse hasCause).

NOTE 1 This includes all relations except the generic relation.

NOTE 2 A **semantic link** (2.17) always has an inverse, i.e. another **semantic link** (2.17) with the opposite direction.

NOTE 3 A **semantic link** (2.17) can be part of a **composite characteristic** (2.7) where it defines the role of the **characterizing concept** (2.5). Similarly, it defines the role of a **characterizing category** (2.4) in a **sanctioned characteristic** (2.15).

3 Conceptual framework for patient findings and problems

3.1 Overview

This Technical Specification describes a **concept system** detailing a **domain constraint** of **sanctioned characteristic**s, each composed of a **semantic link** and an applicable **characterizing category**. An item enclosed by single angle brackets < > refers to a **characterizing category** that can be specialized to various **concept**s as required. An item enclosed within the text by single accolades { } identifies a **semantic link.** Subclause 3.2 lists a set of potential **sanctioned characteristic**s that are illustrated in a **concept** diagram in Figure 1.

3.2 Sanctioned characteristics

3.2.1 Anatomical site

Formal representation of a **sanctioned characteristic** composed of the **semantic link** {HasAnatomicalSite} with an **associative relation** to the **characterizing category** including, but not limited to <Anatomical structure>.

EXAMPLE "Pleuritic pain" has a **composite characteristic** expressed by a **semantic link** {HasAnatomicalSite} and a **characterizing concept** <Pleura structure>.

3.2.2 Morphology

Formal representation of a **sanctioned characteristic** composed of the **semantic link** {HasMorphology} with an **associative relation** to the **characterizing category** including, but not limited to, <Morphological structure>.

EXAMPLE "Adenocarcinoma of rectum" has a **composite characteristic** expressed by a **semantic link** {HasMorphology} and a **characterizing concept** <Adenocarinoma - morphology>.

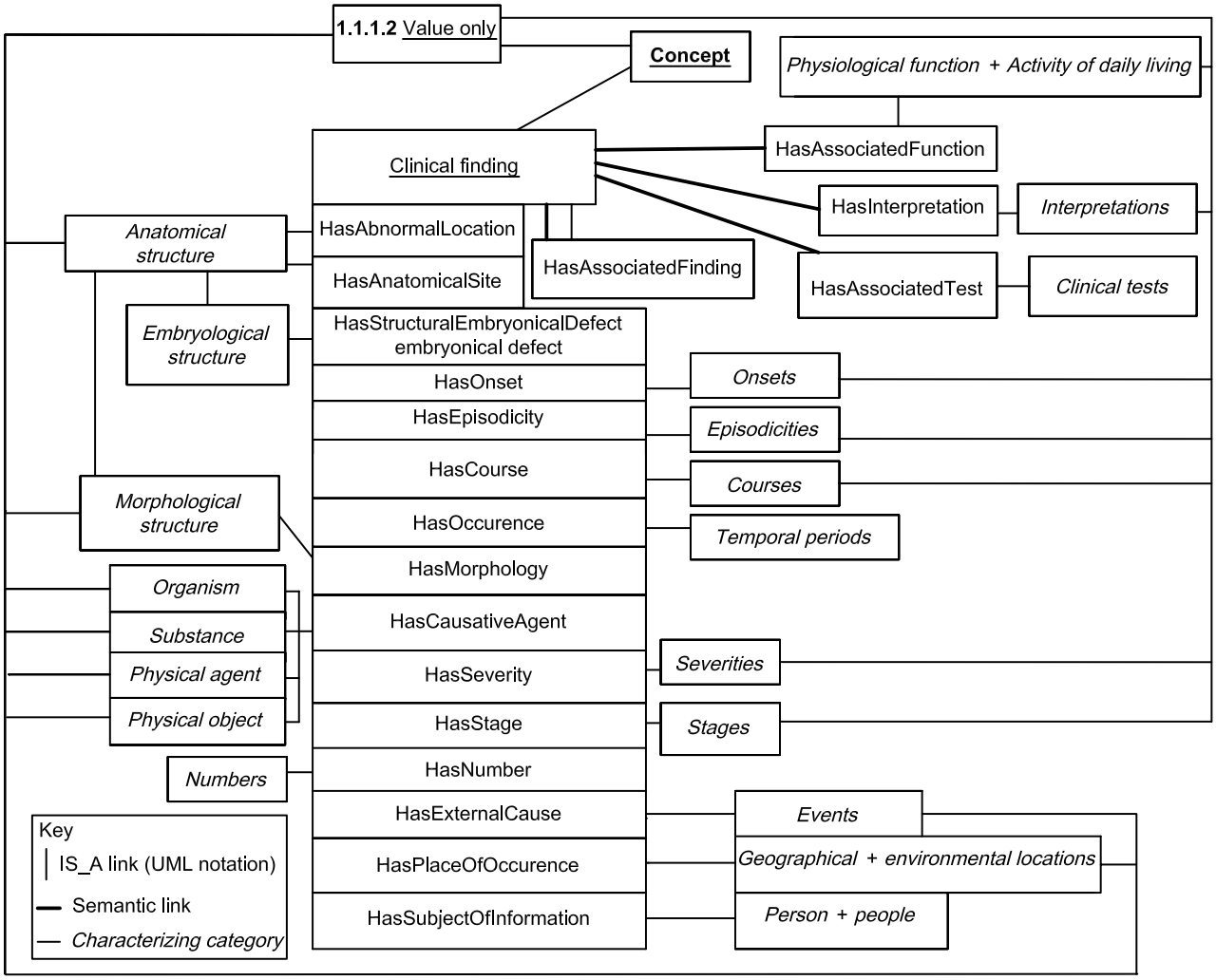

Figure 1 — Conceptual framework for patient findings

3.2.3 Abnormal location

Formal representation of a **sanctioned characteristic** composed of the **semantic link** {HasAbnormalLocation} with an **associative relation** to the **characterizing category** including, but not limited to, <Anatomical structure>.

EXAMPLE "Pelvic kidney" has a **composite characteristic** expressed by a **semantic link** {HasAbnormalLocation} and a **characterizing concept** <Pelvic anatomical region>.

3.2.4 Structural embryological defect

Formal representation of a **sanctioned characteristic** composed of the **semantic link** {HasStructuralEmbryologicalDefect} with an **associative relation** to the **characterizing category** including, but not limited to, <Embryological structure>.

EXAMPLE "Persistent vitelline duct" has a **composite characteristic** expressed by a **semantic link** {HasStructuralEmbryologicalDefect} and a **characterizing concept** <Vitelline duct>.

3.2.5 Causative agent

Formal representation of a **sanctioned characteristic** composed of the **semantic link** {HasCausativeAgent} with an **associative relation** to the **characterizing category** including, but not limited to, <Organism>, <Substance>, <Physical agent> and <Physical object>.

EXAMPLE 1 "Coxsackie pericarditis" has a **composite characteristic** expressed by a **semantic link** {HasCausativeAgent} and a **characterizing concept** <Coxsackie virus>.

EXAMPLE 2 "Alcoholic hepatitis" has a **composite characteristic** expressed by a **semantic link** {HasCausativeAgent} and a **characterizing concept** <Ethanol>.

EXAMPLE 3 "Burn to foot" has a **composite characteristic** expressed by a **semantic link** {HasCausativeAgent} and a **characterizing concept** <Heat>.

EXAMPLE 4 "Plant thorn synovitis" has a **composite characteristic** expressed by a **semantic link** {HasCausativeAgent} and a **characterizing concept** <Plant thorn>.

3.2.6 Onset

Formal representation of a **sanctioned characteristic** composed of the **semantic link** {HasOnset} with an **associative relation** to the **characterizing category** including, but not limited to, <Periods of onset>.

EXAMPLE "Sudden stoppage of urine flow" has a **composite characteristic** expressed by a **semantic link** {HasOnset} and a **characterizing concept** <Sudden>.

NOTE The use of the **semantic link** {HasOnset} may relate to a **characterizing concept** whose meaning may be weakly distinct, e.g. acute onset (as compared to an acute course).

3.2.7 Course

Formal representation of a **sanctioned characteristic** composed of the **semantic link** {HasCourse} with an **associative relation** to the **characterizing category** including, but not limited to, <Courses>.

EXAMPLE "Benign paroxysmal positional vertigo" has a **composite characteristic** expressed by a **semantic link** {HasCourse} and a **characterizing concept** <Paroxysmal>.

NOTE The use of the **semantic link** {HasCourse} may relate to a **characterizing concept** whose meaning may be weakly distinct, e.g. acute course (as compared to an acute onset).

3.2.8 Episodicity

Formal representation of a **sanctioned characteristic** composed of the **semantic link** {HasEpisodicity} with an **associative relation** to the **characterizing category** including, but not limited to, <Episodicity>.

EXAMPLE "Benign paroxysmal positional vertigo" has a **composite characteristic** expressed by a **semantic link** {HasEpisodicity} and a **characterizing concept** <Old episode>.

3.2.9 Occurrence

Formal representation of a **sanctioned characteristic** composed of the **semantic link** {HasOccurrence} with an **associative relation** to the **characterizing category** including, but not limited to, <Temporal periods>.

EXAMPLE "Morning nausea" has a **composite characteristic** expressed by a **semantic link** {HasOccurrence} and a **characterizing concept** <Morning period>.

3.2.10 Severity

Formal representation of a **sanctioned characteristic** composed of the **semantic link** {HasSeverity} with an **associative relation** to the **characterizing category** including, but not limited to, <Severities>.

EXAMPLE "Mild pyrexia" has a **composite characteristic** expressed by a **semantic link** {HasSeverity} and a **characterizing concept** <Mild>.

3.2.11 Stage

Formal representation of a **sanctioned characteristic** composed of the **semantic link** {HasStage} with an **associative relation** to the **characterizing category** including, but not limited to, <Stages>.

EXAMPLE "Membranous glomerulonephritis — stage 2" has a **composite characteristic** expressed by a **semantic link** {HasStage} and a **characterizing concept** <WHO stage 2>.

3.2.12 Number

Formal representation of a **sanctioned characteristic** composed of the **semantic link** {HasNumber} with an **associative relation** to the **characterizing category** including, but not limited to, <Numbers>.

EXAMPLE "Fracture of three ribs" has a **composite characteristic** expressed by a **semantic link** {HasNumber} and a **characterizing concept** <Three>.

3.2.13 Associated function

Formal representation of a **sanctioned characteristic** composed of the **semantic link** {HasAssociatedFunction} with an **associative relation** to the **characterizing category** including, but not limited to, <Physiological function>, <Activity of daily living>.

EXAMPLE "Walking ability normal" has a **composite characteristic** expressed by a **semantic link** {HasAssociatedFunction} and a **characterizing concept** <Walking ability>.

3.2.14 Test

Formal representation of a **sanctioned characteristic** composed of the **semantic link** {HasAssociatedTest} with an **associative relation** to the **characterizing category** including, but not limited to, <Clinical tests>.

EXAMPLE "Tinnels test positive" has a **composite characteristic** expressed by a **semantic link** {HasAssociatedTest} and a **characterizing concept** <Tinnels test>.

3.2.15 Interpretation

Formal representation of a **sanctioned characteristic** composed of the **semantic link** {HasInterpretation} with an **associative relation** to the **characterizing category** including, but not limited to, <Interpretations>.

EXAMPLE 1 "Walking ability normal" has a **composite characteristic** expressed by a **semantic link** {HasInterpretation} and a **characterizing concept** <Normal interpretation>.

EXAMPLE 2 "Tinnels test positive" has a **composite characteristic** expressed by a **semantic link** {HasInterpretation} and a **characterizing concept** <Positive interpretation>.

NOTE The use of **semantic link** {HasInterpretation} is always in conjunction with either the **semantic link** {HasAssociatedFunction} or {HasAssociatedTest}.

3.2.16 External cause

Formal representation of a **sanctioned characteristic** composed of the **semantic link** {HasExternalCause} with an **associative relation** to the **characterizing category** including, but not limited to, <Events>.

EXAMPLE "Whiplash following road traffic accident" has a **composite characteristic** expressed by a **semantic link** {HasExternalCause} and a **characterizing concept** <Road traffic accident>.

3.2.17 Place of occurrence

Formal representation of a **sanctioned characteristic** composed of the **semantic link** {HasPlaceOfOccurrence} with an **associative relation** to the **characterizing category** including, but not limited to, <Geographical and environmental locations>.

EXAMPLE "Found lying on the floor" has a **composite characteristic** expressed by a **semantic link** {HasPlaceOfOccurrence} and a **characterizing concept** <Floor of room>.

3.2.18 Subject of information

Formal representation of a **sanctioned characteristic** composed of the **semantic link** {HasSubjectOfInformation} with an **associative relation** to the **characterizing category** including, but not limited to, <Person and people> describing the context of the relationship of the subject to the clinical finding.

EXAMPLE "Foetal bradycardia" has a **composite characteristic** expressed by a **semantic link** {HasSubjectOfInformation} and a **characterizing concept** <Foetus>.

NOTE There are several issues related to representation of subject of information for which there are several possible solutions. These include: subject "direction" of certain findings (for example, domestic abuse of partner/domestic abuse by partner) and representing the **concept** "foetal" in expressions such as "foetal bradycardia" (by: {Anatomical site} <foetal heart>; {Occurrence} <foetal period>; {Subject of information} <foetus>).

Until there is further experience of various solutions, users of the **concept**ual model should note that these issues exist and that whichever solution is adopted should be consistently applied and that users of the resultant terminology should be informed of the implications of the solution in their own applications and for interoperability with others. Future revisions of this Technical Specification will take account of the experience of terminology developers and users to extend the current specification in this area.

3.2.19 Associated finding

Formal representation of a **sanctioned characteristic** composed of the **semantic link** {HasAssociatedFinding} with an **associative relation** to the **characterizing category** including, but not limited to, <Clinical findings>.

EXAMPLE "Left cerebral infarction causing right hemiparesis" has a **composite characteristic** expressed by a **semantic link** {HasAssociatedFinding} and a **characterizing concept** <right hemiparesis>.

NOTE This representation of an **associative relation** between two or more clinical findings is acknowledged to be simplified and does not take into account partitive relation, sequential relation, temporal relation or causal relation.

4 Conformity to this Technical Specification

4.1 This is within the scope of this Technical Specification, as defined in 1.1.

4.2 A clinical finding claiming to conform with this Technical Specification shall describe **concept**s that are recorded in clinical records that describe any state observed concerning *in vivo* descriptions of structure and function (and not indirect observations, e.g. related to a sample from that patient measuring a biochemical level).

4.3 A clinical finding claiming to conform with this Technical Specification shall describe a state related to a known or assumed pathological process impairing the normal physiological function (3.2.13 {HasAssociatedFunction}) and/or anatomical structure (3.2.1 {HasAnatomicalSite}).

4.4 A clinical finding claiming to conform with this Technical Specification may describe a state affecting all or part of a patient (3.2.18 {HasSubjectOfInformation}) where a specific pathological change (3.2.2 {HasMorphology} and/or 3.2.3 {HasAbnormalLocation} and/or 3.2.4 {HasStructuralEmbryologicalDefect}) may be caused by a defined mechanism (3.2.5 {HasCausativeAgent} and/or 3.2.16 {HasExternalCause}).

4.5 A clinical finding claiming to conform with this Technical Specification may describe an interpretation (3.2.15 {HasInterpretation}) of a state or function (including normal findings) which has been observed directly by the application of a test (3.2.14 {HasAssociatedTest}) relating to a patient.

4.6 Furthermore, a clinical finding may also describe qualitative features 3.2.6 {HasOnset} and/or 3.2.7 {HasCourse} and/or 3.2.8 {HasEpisodicity} and/or 3.2.9 {HasOccurrence} and/or 3.2.10 {HasSeverity} and/or 3.2.11 {HasStage} and/or 3.2.12 {HasNumber}, and their relationship with the environment (3.2.17 {HasPlaceOfOccurrence}).

NOTE Attempting to represent the language of clinical practice gives rise to a number of challenges that have not yet been addressed through agreements on standard clinical expressions or agreed information models. Therefore, there can, at present, be more than one way of representing the same clinical expression – the “foetal bradycardia” example is a case in point: according to the conceptual model, the “foetal” element could be represented in at least three ways (see 3.2.18). This first edition of this Technical Specification does not inhibit existing mechanisms from handling this kind of problem. However, this means that interoperability cannot be guaranteed between different **categorial structures** each claiming conformance to the conceptual model with implications for accuracy of computer processing and electronic communications. Future revisions of this Technical Specification will take account of the experience of terminology developers and users to specify distinct domain constraints, where this is possible.

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