1. Content Issue Summary
   a. Summary of issue

There are currently approximately 770 (20180131 release) concepts in the Clinical findings and Disease hierarchy that contain the word "acquired" in the FSN. These are used clinically to differentiate clinical conditions that may be either congenital or acquired later in life. Of these, 445 are primitive and 275 are sufficiently defined. There are a number of issues related to the presence of these primitive concepts:

- Many of the existing terms have been modeled using "Acquired X (morphologic abnormality)" concepts, which allows them to be sufficiently defined, but results in creation of additional morphology concepts that only differ in the period of life in which they appear. This is analogous to the "Congenital X" morphologies that are being inactivated through remodeling of the Congenital disease subhierarchy.
- "Acquired" and "Congenital" are not morphologies, but timeframes. We currently do not have a way of denoting "All periods of life after birth" like we do for "Congenital".
- The ability to define acquired disorders might also open the door that all disorders that are normally acquired after birth have an OCCURRENCE relationship added, which seems to be "overmodeling". The "Acquired deformity" morphology concepts are incomplete and a suboptimal way to address temporality.
- Some diseases, especially those appearing in the neonatal period, may actually be later manifestations of congenital conditions.
- The application of the proposed modeling pattern would only be applied in situations where there is a specific need to differentiate between congenital and acquired conditions of the same type.
b. Related content projects and requests

<table>
<thead>
<tr>
<th>Related Issues</th>
<th>Comment</th>
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<tbody>
<tr>
<td>PCP-201 &lt;disorder&gt; acquired in &lt;environment or geographical location&gt;</td>
<td>Out of scope for this project as it adds another dimension, i.e. the location of the acquisition.</td>
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<tr>
<td>IHTSDO-1047 healthcare associated infection vs nosocomial infection vs hospital acquired infection</td>
<td>Partially addressed by this project as these are all acquired conditions</td>
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<tr>
<td>IHTSDO-300 art6288-definition and clarification of familial, genetic, and inherited, plus congenital / acquired and related</td>
<td>Partially addressed by this project, hereditary and genetic diseases out of scope</td>
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c. Scope of project

The scope of this project is the approximately 770 existing concepts that contain the string “acquired” in the FSN. There is no intent to apply the congenital or acquired occurrences to any (set) of concepts where the distinction is not explicitly needed (i.e. parsimonious application of the OCCURRENCE attribute). Conversely, the addition of the OCCURRENCE attribute will only be added in situations where it is needed to make explicit distinction between congenital and acquired (or age specific) forms for clinical reasons.

2. Analysis of Issue

Acquired conditions are those which originate and manifest after birth. Since this relates to a period of life as opposed to a specific process or structure, all diseases that occur after birth may be considered to be acquired. According to the American medical association, the periods of life that are included under the “postnatal period” include all periods after birth including the neonatal or immediate postpartum period. Diseases that manifest during the neonatal period of life are challenging since the inciting cause may be present at birth, the clinical manifestation may take a day or two to appear (e.g. Neonatal isoimmune neutropenia).

Three alternatives have been considered:

- Create an intermediate primitive grouper - “Acquired disorder” that would allow subtypes to be fully defined under. These would also classify under the appropriate parent term related to the disorder.
- Create a “period of life” subtype that included all periods except “fetal, congenital, and neonatal(?)” (substantial testing of impacts would be needed)
- Create a set of “Acquired X (morphologic abnormalities)” to support definition of acquired disorders.

There are challenges with using period of life as a way to classify these as age ranges differ among jurisdictions. There is also the issue that the designation of a period of life in an FSN does not always reflect the period of life in which the origin and manifestation are identified. For example, hereditary diseases often manifest later in life. Would these be considered “Congenital”, “Acquired”, both? What do we mean when we say “acquired”?

It would be prudent to limit the use of the period of life grouper to necessity, i.e. for only those disease where the FSN specifies “acquired” or a need to specify a distinction between congenital and acquired forms of a disorder. What would be the advantage of a top level grouper “Acquired disorder”? The challenge is how to represent the acquisition/origin of the trait as opposed to the clinical manifestation of the trait, or if that distinction is needed. For non-semantically decomposable FSNs, it is more important to get the modeling correct than change the idiom.

3. Additional issues identified

This solution does not adequately address concepts containing the term “juvenile” as that term is used to represent the clinical manifestation of a disorder at a very broad range of life periods. There is an inactive concept 282035009 - Juvenile (qualifier value), which I suspect was inactivated due to the variability in determining which more specific life periods it encompasses.

Many of the affected concepts are not modeled according to the pattern shown in the solution section below. These will be edited to conform to the proposed pattern.

Concepts using the term “neonatal” must be individually and carefully curated to ensure that the correct origin of the disorder is assigned.

4. Solution

- A new qualifier value of “Period of life beginning after birth and ending before death (qualifier value)” will be created to aggregate the periods of life that would be used to define “Acquired” conditions.
- Move the following concepts under this new grouper:
  - Adolescence
  - Adulthood
  - Middle age
  - Childhood
  - Infancy
  - Old age
- All concepts that current explicitly state “acquired” in their FSN or in a “synonym” would have a new relationship "OCCURRENCE = Period of life beginning after birth and ending before death (qualifier value)" added to the defining relationship group(s), if an OCCURRENCE
relationship does not already exist. This will allow many of the currently primitive concepts to become sufficiently defined, resulting in a richer set of inferences.

- When revising modeling of acquired disorders, remove any “Acquired” morphologies and replace with general parent morphology. E.g.
  - Replace “acquired deformity” with ‘deformity’.
- As the use of the “Acquired X (morphologic abnormality)” concepts is eliminated, inactivate the morphology concepts.

The general pattern for revising the affected concepts is shown in the diagram below:

5. Construction

The attached file is a listing of the affected concepts present in the 20180131 release of SNOMED International:

https://docs.google.com/spreadsheets/d/1K1LhTklXJl8GhRGMN6G6j3xuX3HfGa7UoQS-0hiBRB8/edit?usp=sharing

Some identified issues:

- Most definitions include the neonatal period in definitions of acquired disorders. Given the variable use of the term to represent disorders occurring both antenatally as well as the clinical manifestation of genetic/hereditary diseases, these will need to be researched individually for the correct assignment.
- Some disorders that are developmental but manifest later in life are currently classified as acquired. AMA definition excludes genetic diseases even if manifesting later in life.
- Missing some “musculoskeletal structure of X” concepts for deformities
- Due to existence of primitive concepts, there is a need to inactivate duplicates that arise when the standard pattern is assigned. For example:
  - 367393006 - Acquired claw hand (finding) DUPLICATE TO 367393006 - Acquired claw hand (disorder)
  - Acquired forearm deformity, excluding fingers DUPLICATE to “Acquired forearm deformity” as the fingers are not anatomically part of the forearm.
  - Inactivate 45402003 – Juvenile disease (finding) as ambiguous.
- Periods of life assignment where the onset is at one stage of life but persists throughout the rest of life. How to assign OCCURRENCE?
- Some concepts contain the phase “congenital OR acquired”. Inactivate and replace?

6. Transition phase
## 7. Glossary of Terms

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<tr>
<th>Term</th>
<th>Definition</th>
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<tr>
<td>Acquired disease</td>
<td>Developed or originating after birth: not congenital or hereditary. Hyperoxaluria may be due to overproduction from hereditary disorders of metabolism or it may be an acquired disorder caused by intestinal disease or diet. Source JAMA 1988. Of or relating to a disease, condition, or characteristic that is not congenital but develops after birth.</td>
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<tr>
<td>Postnatal period</td>
<td>That interval of life after birth; in humans, usually divided into periods: neonatal, infancy, childhood, adolescence, and adulthood.</td>
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