Diagnoses, Syndromes, and Diseases: A Knowledge Representation Problem

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Abstract

Despite their widespread use, the terms "syndrome", "disease" and "diagnosis" are sometimes utilized improperly and ambiguously, compounding the complexities of medical knowledge representation. The definitions and illustrative examples provided here will be useful for developers of diagnostic expert systems.

Description of the Problem

Representing medical knowledge is a highly complex endeavor. The improper use of the terms "syndrome", "disease" and their relations to "diagnosis" is one of the difficulties with which medical informaticians must deal, especially when developing expert systems to support diagnoses. Although ubiquitous in medical and lay discourse, the term "disease" has no unambiguous, generally accepted definition. However, most of those using this term allow themselves the comfortable delusion that everyone knows what it means.

Only sparse and fragmented literature could be found regarding this issue.

Purpose

The different concepts about "what types of conditions may be said to constitute a disease" will be discussed. The "unorthodox" ways in which terms "disease" and "syndrome" are being used, as well as their different interrelationships, will be shown.

Illustrative cases will be used to present potentially ambiguous situations found when representing medical knowledge in expert systems.

The "Disease Entity" Assumption

Diagnostic categories (diseases and most syndromes) are simply concepts. They are justified only if they provide a useful framework for organizing and explaining the complexity of clinical experience in order to derive inferences about outcome and if they guide decisions about treatment.

A **syndrome** is a recognizable complex of symptoms and physical findings which indicate a specific condition for which a direct cause is not necessarily understood. Thus in practice doctors refer to the infamous "viral syndrome" as such because of the uncertainty regarding the legion of viral agents that is causing the illness. Once medical science identifies a causative agent or process with a fairly high degree of certainty, physicians may then refer to the process as a **disease**, not a syndrome. Mucocutaneous lymph node syndrome became Kawasaki syndrome which in turn metamorphosed into Kawasaki disease; the latter is properly a disease, no longer a syndrome, by virtue of its clearly identifiable diagnostic features and disease progression, and response to specific treatment.

Albert *et al.*[1] catalogued six general views or concepts about what types of conditions may be said to constitute a disease. These views range from nominalism and cultural-relativistic theories (i.e. some conditions become a disease when a profession or a society labels it as such) to a "disease realism" view (objectively demonstrable departure from adaptive biological functioning). The latter model is the one best suited to the present state of medicine; it emphasizes that the clinical signs and symptoms do not constitute the disease and that it is not until causal mechanisms are clearly identified that we can say we have "really" discovered the disease.

Medical literature, even that from governmental organizations and institutions authorized to implement standards, is plagued with misleading assertions such as "a syndrome is a disease …", "a syndrome indicates a particular disease…" and "Lyme disease syndrome" (It is inappropriate to apply "syndrome" to Lyme disease because its causative agent is known).

Some syndromes such as "heart failure" are useful medical concepts but are not diagnoses, whereas more specific syndromes such as "congestive heart failure" or "right heart failure" are diagnoses[2]. Due to the imprecision of natural language, some syndromes could also imply a simple pathological finding (vasculitis) or just a physical finding. Frequently, for example, arthritis syndromes are simply referred to as "arthritis".

References

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