Disease as an example of non-Aristotelian categories

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Abstract This paper discusses the problem of definition of disease in formal ontologies. We argue that different conditions usually regarded as disease have no common exclusive intrinsic property, due to their ontologically heterogeneous nature. We maintain that being a disease depends on social circumstances. From a purely ontological view it is hard to accept. Still our proposed model can be used in formal systems and has the benefit to point to some aspects that are absolutely relevant for disease control and prevention.

What is non-Aristotelian?

The principles of defining categories (perhaps even the word 'category' as well) was first described by Aristotle. His way of definition was based on the following principles of logic:

- All statements must be either false or true
- No statement can be false and true at the same time and under the same circumstances.
- There is nothing in between true and false.

An Aristotelian category is defined by two statements: one specifies the 'genus proximum' (an IsA statement) and the other a 'differentia specifica'. This way of definition is still valid today and widely used in ontology engineering. It is often referred to as intension. Another accepted way of definition is the extension, when we give an exhaustive list of things (or subcategories) that constitute a category. We have to stress, that via extension it is possible to define any set (in a mathematical sense) but such an exhaustive list forms a category or entity only if the listed things have a common exclusive property (a proposition that is true for all and only for the listed things). This common exclusive property corresponds to the 'differentia specifica', so extensions and intensions are alternative ways to achieve the same.

Contemporary literature mentions fuzzy logic and fuzzy categories as non-Aristotelian [1]. Indeed in fuzzy logic there is a continuum of truth values between 0 and 1, so fuzzy logic violates the third law listed above. For instance the statement "I am a talented man" might have a truth value of something like 0.2 or 0.87, depending on how talented I am. "Talented man" is therefore a fuzzy concept. But in case this value is 0.2 for me than the question can be reformulated: is it true that my membership function value in the set "talented man" is 0.2? This question can be answered by 'Yes' or 'No'. Doing so, we can still satisfy all criteria of Aristotelian logic. So what I mean by 'non-Aristotelian' is something different.

We demonstrated in a previous paper [2] that there are many biomedical categories that neither theoretically nor practically can be defined in terms of an Aristotelian definition. This is not because of their fuzzy nature, but due to the fact that things constituting the category apparently lack any common exclusive property. Notion of 'Disease' seems to be a typical example. E.g. some diseases are painful, but not all of them, while some painful conditions are not diseases, etc. Such categories can not be defined in Aristotelian sense either by using the two-valued or fuzzy logic. Defining 'disease' by extension would mean to create an exhaustive list of diseases, including all extinct ones and those that will occur in the future only. It is unnecessary to emphasise the hopelessness of such an approach. We may wonder, how humans are still can agree for the most part what constitutes a disease and what not.

What is a 'Disease'?

The main point in understanding what is a 'disease' lies in the sentence above: "for the most part". This means that we do not agree always. So there are some conditions considered as disease by certain people but not by others. The level of agreement depends on the similarity of the social/cultural background of people. L King stressed that classifying something as disease depends on social situations [3]. Homosexuality is a good example: depending on the culture of the society it can be regarded as sin, disease, or normal variation. This corresponds how a society reacts to homosexuality: by punishment, by treatment or by tolerance (no reaction). Whenever a condition is regarded as disease in a given society it provokes some specific (health) reaction against itself. The actual reaction can be different depending again

on the culture and the development of the given society, but basically it can have three forms: 1) avoid situations where the disease may occur ('prevention'); 2) help somehow the patient to get rid of the disease ('treatment') or at least to learn how to live with it ('rehabilitation'); 3) exclude the patient from the community in order to avoid making more harm to population ('isolation'). The latter happened e.g. with people suffering from leprosy in ancient time, or with many psychiatric patients up to the modern age. Obviously some conditions are regarded as disease in every known and conceivable society. E.g. appendicitis, nose-bleeding, fracture of extremities, and cholera probably were considered as diseases in every culture, even in lack of recognition of their pathological nature or cause. But the definition of disease can not be restricted to those conditions. Serious diseases, e.g. epilepsy, chondrodystrophy would be excluded. Epilepsy, the 'morbus sacer' (sacred disease) could be seen as a transcendental phenomenon in certain cultures, while condrodystophic patients due to their short extremities and relatively oversized head were beloved court clowns of the middle age.

We do not state however that all instances of all diseases necessarily provoke a health activity actually. Many of disease occurrences might remain unrecognised or ignorantly tolerated. What we state is that any disease has this potential or function. As all screw drivers have the ability to fix or loose screws even if some of them may be never used for that. So we rephrase our definition as follows:

Anything is a disease that in a given society has the potential to provoke some specific (health) action against itself.

Disease as hybrid class

But what kinds of things have the ability to provoke a health action? Let us investigate the ontological nature of those things. Here another problem arises, that explains why many attempts failed to formally define disease. Top level ontologies distinguish endurants and perdurants (often the names occurrent/continuant or snap/span entities are used respectively). Whatever names are used, it concerns disjoint categories: Endurants exist entirely at any time they exist; while perdurants have temporal parts. It is very clear that nothing can be both endurant and perdurant. However, taking the definition of disease presented above, we realise that nothing forbids both endurants and perdurants to have the ability to provoke health actions. We do not say that a certain given disease (or any actual instance of it) could be endurant and perdurant as well. What we state is that disease is a category that may comprise both endurants and perdurants. That is why we call this category heterogeneous: it is a union of mutually disjoint subcategories. If we consider different diseases in detail we might see, that in most of the cases there is some pathological process (or there is a pathological alteration of or lack of some physiological process) that leads to some morphological alterations. Processes are perdurants, whereas morphological phenomena are endurants. Again looking at the details we may realise that a pathological process is a consequence of a structural state that is again a consequence of another process. So, most of the diseases are rather complex compositions of processes and morphological alterations at different organisational levels (molecular, cellular, organ level, etc.). E.g. a cancer (as a pathological mass in the body) is an endurant resulting from some pathological cell division process that is a perdurant. But this process probably is a result of some abnormal macromolecular structures that are endurants, and so on. Even if we do not go into the details we realize, that in case of many diseases the presence of a structural condition dominates the clinical picture (e.g. arterial aneurisms, fractures and ruptures, various cysts etc.) while in other cases (e.g. consumptive coagulopathy, autoimmune disorders) are process-dominated. Even if we know that all these conditions are complexes of processes and structural states, it is worthwhile to set up two categories for morphology and process dominated diseases. The former is subsumed by endurant the latter by perdurant. Then 'Disease' (as such) could be defined as the union of these two, and the ability to provoke health action should be added as a necessary condition for disease.

Alignment to top level ontologies

In order to maintain consistency and compatibility, it is often recommended that domain ontologies should be aligned to some top level ontology. Testing two frequently referenced top level ontologies, DOLCE (Descriptive Ontology for Linguistic and Cognitive Engineering created by Laboratory of Applied Ontology, CNR Rome, see http://www.loa-cnr.it/DOLCE.html) and BFO (Basic Formal Ontology created by IFOMIS see http://www.ifomis.uni-saarland.de/bfo/) we found some conflicts between them. In both top level ontologies processes are classified as perdurants (named occurents in BFO) Structural conditions in BFO are considered as "quality" (actually 'shape of a nose' is a given example of quality in BFO; think of nasal deformity as a good example of structurally defined diseases). In case of DOLCE quality is one of the top level categories, neither endurant nor perdurant. Structural conditions can be classified in DOCLE also as 'features' that are dependent essential wholes, classified as physical endurants, e.g. holes in a cheese (or similarly a cyst in the kidney), etc. Consequently, our notion of disease should be represented in BFO as a union of some processes (perdurants) and some qualities (endurants), while in case of DOLCE as union of some processes (perdurants) some features (endurants) and some qualities that are neither endurants nor perdurants.

Not questioning their theoretical merits, we have to say that at the present state of the art philosophically strong top-level ontologies are sometimes conflicting and sometimes offer little help in domain ontology development.

Another important issue here is whether the proposed notion of disease can be represented in description logic or first order logic. If not, automated reasoning is hardly possible. There is no space here to discuss this question; I just want to mention that according to our initial investigations first order logic can be sufficient to represent our disease definition; however work around solutions can be necessary to avoid second order logic statements. Gangemi et al represent inflammation as 'descriptions' that are social objects in DOLCE [4]. This is probably also due to avoid second order logic, but the author is not in favour of such a solution, since the commonsense notion of disease tells us that they are real world thing rather that merely human inventions. Of course, certain real world things can satisfy a description but a disease can exist without any appropriate description.

Conclusion

A unique totally satisfying formal definition of disease is hard to give, because conditions used to be considered as diseases have usually no common exclusive intrinsic property, and because those conditions are ontologically heterogeneous. The criterion of disease we propose is not an intrinsic (ontological) property of disease, but a socially determined feature. Purely philosophically it is hard to accept that the definition of disease depends on social conditions, but this opens the way toward practically useful formal systems. E.g. for disease control agencies like CDC and ECDC it is absolutely important to consider that certain conditions may provoke health action in one country but may not in another. This can be especially important in case of transmittable diseases.

Acknowledgements

The author expresses his special thanks to Arie Hasman and to members of the Health Ontology Forum (László Balkányi Gergely Héja and Péter Varga in particular) for commenting and advising this manuscript. The work was partly supported by Semantic Mining Project, EU IST FW6 507-505

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