A Consideration on Identity of Diseases

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ABSTRACT. We discuss an consideration on identity of diseases based on an ontological model of diseases as causal chains. Because we define an individual disease is constituted by a causal chain, changes of the causal chain implies changes of the disease. That is, in order to consider an identity of diseases, we have to investigate how an identity of a causal chain changes. For this purpose, we introduce a classification of causal inks and causal chains. Then, we propose an ontological model to deal with identity of diseases as causal chains. It would contribute to the ontological treatment of identity of a disease in medical information systems.

1 Introduction

Recently, many medical ontologies are developed for realizations of sophisticated medical information systems such as OGMS [Scheuermann et al. 2009], DOID[Osborne et al. 2009], IDO[Cowellet al. 2010]. We also proposed a definition of a disease by capturing a disease as a causal chain of clinical disorders [Mizoguchi et al. 2011].

This extended abstract discusses identity of individual diseases while the main concern in our previous work is capturing static characteristics of diseases. An individual causal chain changes through its processes such as extending, branching, and fading etc. When we suppose an individual disease is constituted by such a causal chain, changes of the causal chain implies changes of the disease. Then, how the identity of the disease should be dealt with? To answer this issue, we consider identities of a disease as causal chain based on some ontological theories.

2 A model of disease

In [Mizoguchi et al. 2011], we defined a disease as:

Definition 1: Disease

A disease is a dependent continuant constituted of one or more causal chains of clinical disorders appearing in a human body and initiated by at least one disorder.

It is based on observations that a typical disease as a dependent continuant enacts extending, branching, and fading processes before it disappears. Such an entity (a disease) can change according to its phase while keeping its identity.

When we collect individual causal chains belonging to a disease, we are able to find a common causal chain (partial chain) which appears in all the instance chains.

By generalizing such a partial chain, we can get the notion of Core causal chain of a disease as follows:

Definition 2: Core causal chain of a disease[1]

A sub-chain of the causal chain of a disease whose instances are included in all the individual chains of all the diseases. It corresponds to essential property of a disease.

Definition 2 provides a necessary and sufficient condition for determining the type to which a given causal chain of clinical disorders belong. That is, any concrete causal chain of clinical disorders which includes instances of the core causal chain of a particular disease type belongs to the disease type. We thus can define such a type of disease that allows all possible variations of physical chains of clinical disorders observed for patients who contract the disease. We can introduce an *is-a* relation between diseases using chain-inclusion relationship between causal chains as follows:

Definition 3: *Is-a* relation between diseases [Mizoguchi et al. 2011]

Disease A is a super type of disease B if the core causal chain of disease A is included in that of disease B. The inclusion of nodes (clinical disorders) is judged by taking an *is-a* relation between the nodes into account, as well as sameness of the nodes.

Assume, for example, that core chains of (non-latent) diabetes, type-I diabetes, and steroid diabetes are defined as follows;

(Non-latent) diabetes: <deficiency of insulin → elevated level of glucose in the blood>

type-I diabetes: <destruction of pancreatic beta cells → lack of insulin I in the blood → deficiency of insulin → elevated level of glucose in the blood>

Steroid diabetes: <long term steroid treatment → … → deficiency of insulin → elevated level of glucose in the blood>

Then we get <*type-I diabetes is-a (non-latent) diabetes*> and <*steroid diabetes is-a (non-latent) diabetes*>according to Definition 3. .

3 Identity of Diseases

3.1 Classification of causal chains

In order to discuss identity of disease, we need to consider identity of a causal chain of clinical disorders. Because it is a continuant, it can change keeping its identity. Before discussing identity of causal chains, we introduce a classification of them to capture how they change.

Our theory of processes and events exploited in this paper is found in [Galton & Mizoguchi 2009] in which the two key notions are (1) processes are intrinsically ongoing/in-progress and events are not and (2) an event is constituted of processes. Based on this theory, we introduced two kinds of processes as follows [Mizoguchi et al. 2011]:

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[1] This is a revised version of Definition 3 in [Mizoguchi et al. 2011].

(1) **Cumulative continuous process:** a process that proceeds without completing the current process at every instant in time.

(2) **Non-cumulative process:** a process that proceeds by completing the current process at every instant in time.

A causal chain is composed of one or more pairs of entities such as a causal event/process and an effect event/process, in which the latter has been caused by the former. The effect becomes another cause that causes another effect in the case of multiple-pair chains. What makes causal chains of clinical disorder special is that causal entities are usually still active when the effect entity has been caused. Therefore, the two entities overlap in temporal space. This shows that clinical causal chains belong to cumulative continuous process (Fig.1). On the other hand, in causal chains of non-cumulative process, each event is caused by a completed previous event (Fig.2).

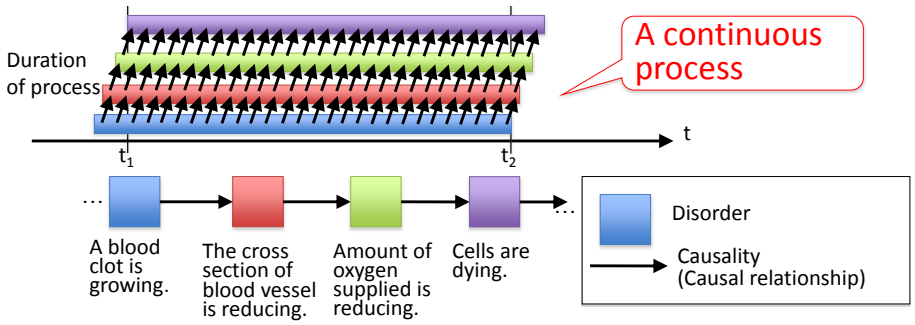

Fig.1 An example of cumulative continuous process.

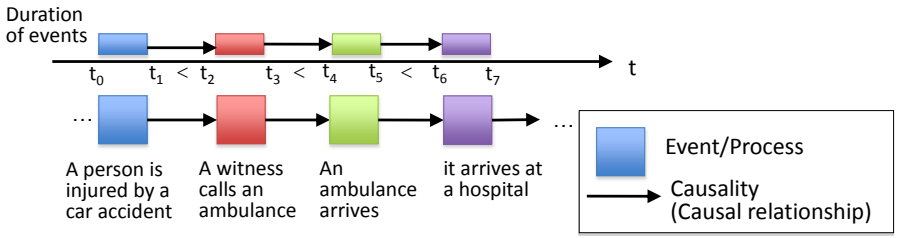

Fig.2 An example of non-cumulative process.

In order to discuss kinds of causal chains, we define three kinds of causal links from the perspective of "ongoingness".

Definition 4: Ongoing causal link

This is a link which shows that the propagation of causality from the cause to the effect is ongoing.

Definition 5: Historical causal link

This is a link which shows that the propagation of causality from the cause to the effect had been terminated.

Definition 6: Primitive causal link

This is a link which shows only primitive relation between cause and effect excluding the temporal information. It is used mainly for causal events which can be viewed as that they happened at the same time.

Continuous propagation of causal effect requires the fact that both causal and effect processes are ongoing. Therefore, ongoing links can appear only between ongoing processes, and they correspond to causal relations found in Cumulative continuous processes.

Transition from an ongoing process to a completed event can be explained in terms of the change from ongoing causal link to historical causal link as follows: When the causing process has been completed, the process constitutes a new event as a past event of the causation together with a change of the ongoing causal link to a historical causal link.

Primitive causal link can be interpreted as a degenerated causal link which includes the change of ongoing causal link to historical causal link in it and is useful for representing causal events composed of non-cumulative processes. It does not include non-zero temporal interval but deals with the two events as if they happen at the same time with keeping the causal order.

Now, we introduce three kinds of causal chains to capture the change of diseases according to the causal links defined above.

Definition 7: Strongly-connected causal chain

Any causal chain all of whose causal links are ongoing causal links. That is, it is composed of only ongoing processes. A causal chain composed of one ongoing process is included as a special case.

Definition 8: Weakly-connected causal chain:

Any causal chain which includes at least one historical or primitive causal link and at least one ongoing process.

Definition 9: Historical causal chain:

Any causal chain all of whose causal links are historical or primitive causal links. That is, it is a causal chain composed only of completed events.

Definition 10: Ongoing causal chain:

Any causal chain which has at least one ongoing process. It subsumes Strongly-connected and Weakly-connected causal chains.

When an ongoing process of a strongly-connected causal chain has been completed, the whole chain turns to a weakly-connected causal chain. If all the ongoing processes have been terminated, then the chain turns to a historical causal chain. That is, a causal chain born as a strongly-connected causal chain and grows such as extending, branching etc., then it turns to a weakly-connected causal chain, and finally it turns a historical causal chain.

3.2 Identity of a causal chain

In order to talk about identity of a disease instance we need to establish identity of causal chain of clinical disorders. We discuss identity of a causal chain based on identity of entities [Kozaki et al. 2010]. There are two types of identity:

Instance identity: Identity for discussing the sameness of instances.

Class identity of a thing: Identity for discussing the sameness of the class the entity belongs to.

During a course of change such as growing, branching, etc., the causal chain of clinical disorders, keeps its identity. That is, we should be able to identify it as the same causal chain in a different state. In order to deal with such change properly, we employ Identity for replacement (*Ire*) among four kinds of identity discussed in [Kozaki et al. 2010]. *Ire* is defined as:

Identity for replacement (denoted as ***Ire***):

Identity which an instance of the whole continues to be itself without becoming another thing while whose parts are being replaced independently of their kinds and number of the replaced parts.

A typical example for the use of *Ire* is a bike whose parts are replaced one by one. Whatever parts are replaced by how many, the bike whose parts are replaced remains the same in terms of *Ire*. Exactly the same applies to causal chains of clinical disorders. In the same way, any individual causal chain, *Ci*, never changes its *Ire* identity by replacement of its parts. Neither growth nor branching can change its identity. When an intermediate disorder is terminated to become an event, it becomes a weakly-connected causal chain keeping its *Ire* identity. In other words, it does not change its *Ire* identity if it has at least one ongoing disorder.

When all the links have become historical causal links, the resulting historical causal chain must be a different instance (chain) from the ongoing causal chain because all of its nodes and links are different from the original chain (chain before the change). That is, when all disorders included in an ongoing causal chain has gone (been recovered), the chain loses its identity and turns to a historical causal chain of different identity. It corresponds to cure of the disease and the resulting historical causal chain represents anamnesis of the patient.

Class identity, on the other hand, is used to talk about to what type an individual causal chain belongs. As stated in Definitions 1 and 2, an individual causal chain of clinical disorders, *Ci*, is identified to belong to a disease type, D_A, when it includes an instance of the core causal chain of D_A as its part, and then, it gains class identity for D_A. Such *Ci* is said to be an ongoing disease D_A if it is an ongoing causal chain. *Ci* can get another identity for a difference disease when it also includes an individual core causal chain of another disease, say, D_B. Class identity is kept until the ongoing causal chain turns to a historical causal chain.

3.3 Identity of diseases

Considering various cases concerning ongoing/historical states of a causal chain, we can discuss identity of the individual disease which is constituted by the causal chain. We discuss examples of the life of an individual disease as a causal chain of clinical disorders. Note here that we are not trying to predict the future state of a particular disease of a particular patient, but to exemplify possible changes of diseases in the real world represented as a causal chain of clinical disorders in terms of its *Ire* identity and type membership.

(1) A causal chain of clinical disorders, *Ci*, is born as a strongly-connected causal chain. At the same time, it gains its instance identity ($\boldsymbol{I_{ins\_1}}$). *Ci* can extend, branch, etc. with keeping its identity.

(2) Before it establishes itself as an instance of a particular disease type, its class identity is kept undetermined. When *Ci* extends and includes an instance of the core causal chain of a disease D_A as its partial chain, it becomes an instance of D_A and gets class identity $\boldsymbol{C_{ins\_A}}$ for D_A. Once *Ci* gets class identity for a disease type, the class

identity is kept unchanged independently of its extension and/or branching because it still includes the instance of the core causal chain of the disease.

(3) After some changes of the disorders as well as its termination of ongoing process to become an event, related causal links of *Ci* change from ongoing links to historical links. Then, *Ci* changes from a strongly-connected causal chain to a weakly-connected causal chain. Nevertheless, *Ci* keeps its instance identity as an ongoing causal chain.

(4) When all the processes i*n Ci* have been completed and become events, and hence all the causal links turns to historical links, *Ci* becomes a historical causal chain as a new individual causal chain, say, $\boldsymbol{I_{ins\_2}}$ all whose nodes are events constituted of related past processes.

4 Concluding remark

We have discussed identity of a disease as a causal chain of clinical disorders. We believe that it could contribute to an ontological account of evolution of diseases. It also would be a first step toward a comprehensive ontological account of diseases covering its evolution.

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