# A METAONTOLOGY FOR MEDICAL DIAGNOSTICS OF ACUTE DISEASES. PART 1. AN INFORMAL DESCRIPTION AND DEFINITIONS OF BASIC TERMS

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*Abstract: The aim of this article is to describe formally a metaontology for medical diagnostics of acute diseases in the language of applied logic. The article includes an informal description of the metaontology, and the part of its model which contains the definitions for basic terms of knowledge and situations and also their integrity restrictions in the form of ontological agreements.*

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## Introduction

Any expert system is based on a conceptualization of the domain. An explicit representation of the conceptualization is usually called ontology [1]. Publications of domain ontologies and their models representing the conceptualizations which are near to the ones used in science, education and practical activities are of particular interest. There are three reasons for this. First, such publications extend our view of complex ontology construction. Second, they bring out in which direction the languages for representation of complex ontology models should be developed. Third, the published ontologies and their simplifications can be used in expert systems development.

These ideas relate to expert systems for medical diagnostics in full measure. The ontologies which are the basis for some expert systems of medical diagnostics are described in sufficient detail [2], sometimes in a formal manner [3]. But more often it is rather difficult to extract the ontology that is the basis from publications on expert systems [4, 5]. The ontologies which are the basis for many expert systems of medical diagnostics are significantly simplified ones in comparison with the real conceptualizations of this domain. Such properties as development of pathological processes in time, interaction of various types of cause-and-effect relations, and also combined and complicated pathologies are usually not considered in these ontologies.

Studying real ontologies of medical diagnostics (which are different in details for medicine of different countries, as it is fairly noted in [1]) goes back to [6, 7]. At present time various means for ontology description in form of computer languages [8, 9] as well as mathematical ones [10] have been developed. The development of means for ontology description led to the publication of a formal model of medical diagnostics ontology that was close to real conceptions in medicine [11].

Medical diagnostics is a wide domain. It means that ontologies of many its divisions are special cases of the same metaontology of this domain. The aim of this article is an explicit and formal description of this metaontology in the language of applied logic [10].

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## 1. An informal description of the metaontology

In this article a metaontology of medical diagnostics is the object of modeling. The metaontology is a set of conceptual ideas about the processes in a patient's body and in its environment which are essential for solving the task of medical diagnostics. These ideas are based on the results of the works [7, 11].

The reality of the domain is a set of situations [12]. Every situation contains information about patient's body during a time interval. The beginning of this interval is the earliest moment to which the information about this situation is related. Time moments are measured in hours from this beginning using the scale of nonnegative integers.

The processes which proceed in a patient's body can be arbitrary divided into external (observable) and internal ones. The latter processes are the object of diagnostics. The observable processes are called signs, and the internal ones are called diseases. The signs have values which can be obtained during their examination and vary in time. They are considered as qualitative (scalar)[1]. The signs are a subclass of the observations class. Another subclass of the observations class is the anatomical-and-physiological features of a patient's body. They also have scalar values. In this article it is assumed that these values cannot vary in time. The last subclass of the observations class is the events which happened to a patient. They also have scalar values. The events can happen at individual time moments and the values of an event can be different at different time moments.

In this article only the acute diseases are considered. A patient can be healthy or have one or more diseases. Every disease proceeds in time and can sequentially pass through several stages in its development. They are called development periods. The diagnosis is a set of the diseases with which a patient is ill in the situation. Every disease from the diagnosis can have a single cause.

The basic type of the association between the processes which proceed in a patient's body is the class of cause-and-effect relations. This class includes complications, etiologies, clinical manifestations, clinical manifestations modified by event's influence, responses to event's influence and normal reactions. A cause-and-effect relation develops in time according to one of the possible variants of its development which is determined by the values of acting factors (anatomical-and-physiological features) and perhaps by the cause.

A complication associates a (primary) disease from the diagnosis (the cause) with another (secondary) disease from the diagnosis (the effect). The secondary disease arises as a complication of the primary disease in a time lapse after its beginning. A variant of a complication depends on the values of the acting factors only.

An etiology associates a value of an event (the cause-event) with a disease from the diagnosis (the effect). The disease arises as an effect of this event in a time lapse after the event happened. A variant of an etiology depends on the value of the cause-effect and on the values of the acting factors.

A clinical manifestation associates a disease from the diagnosis (the cause) with a sign (the effect). A variant of the clinical manifestation development depends on a development period of the disease and on the values of the acting factors. The values of the sign can be the effect of the disease on the time interval which corresponds to a diseases' development period. This development period in its turn can be divided into several dynamics periods which are determined by a variant of the clinical manifestation's development.

A clinical manifestation modified by an event's influence has the cause that is a disease from the diagnosis and the cause-event with a value. Its effect is a sign. A variant of a clinical manifestation's development modified by an event's influence depends on the value of the cause-event and on the values of the acting factors. The values of the sign can be the effect of the combined action of the cause and cause-event on a time interval which begins at the moment when the event happens. This time interval in its turn can be divided into several dynamics periods which are determined by a variant of the clinical manifestation's development modified by the event's influence.

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[1] In medical diagnostics the values of signs (and also of events and anatomical-and-physiological features) can also be quantitative (dimensional) and be represented by integer or rational numbers. But in this article only scalar values will be considered in order to shorten the description of the metaontology.

A response to an event's influence associates a value of an event (the cause-event) with a sign (the effect). A variant of the development of a response to an event's influence depends on the value of the cause-event and on the values of the acting factors. The values of the sign can be the effect of the cause-event on a time interval which begins at the moment when the event happens. This time interval in its turn can be divided into several dynamics periods which are determined by a variant of the development of the response to the event's influence.

The cause of a normal reaction is not identified and its effect is a sign. A variant of a normal reaction's development depends on the values of the acting factors only. During the time intervals when the values of the sign do not have other causes they are the effect of the normal reaction.

The values of a sign can vary as a result of the simultaneous influence of several cause-and-effect relations. The whole time interval of each sign's examination can be divided into such periods that during each period the values of the sign are the effect of a single cause-and-effect relation (from all the possible ones). As this takes place, the beginning of the time interval during which a cause-and-effect relation acts can be only the beginning of such a period, and the end of the time interval during which the cause-and-effect relation acts can be only the end of such a period. Among different types of cause-and-effect relations a partial order is established which is determined by the modality[1] of cause-and-effect relations and by the moments of the cause's initiation.

The domain knowledge consists of knowledge about all the observations, diseases and cause-and-effect relations. Knowledge about an observation includes the range of its possible values. Knowledge about a disease includes a sequence of its development periods. Every period contains an interval of the durations of this period. In reality the duration of every development period belongs to the interval of possible durations which corresponds to this period.

In addition, knowledge about cause-and-effect relations, signs and diseases can contain necessary conditions. The fulfillment of such a condition is necessary, so that the associated sign, disease or cause-and-effect relation can take place for a patient. A necessary condition is a conjunction of components. Every component is a reference to an anatomical-and-physiological feature, and also to a subset of possible values of this feature. A component of a condition is considered to be fulfilled if the examined value of this feature belongs to the subset of possible values of this feature. If the necessary condition is absent in the description of a concept then it is considered to be always fulfilled.

Knowledge about any cause-and-effect relation includes the causal regularity that is a disjunction of variants. If a cause-and-effect relation takes place in reality then one of these variants is fulfilled. In general case a variant is an implication. Its antecedent can contain a condition on the cause, a condition on the cause-event, and a condition on the acting factors[2], and its consequent can contain either an interval of possible durations for the time lapses between the moment when the cause took place and the beginning of the disease (of the effect) or a sequence of the dynamics periods[3].

The description of a complication includes the references to the cause (a primary disease), the effect (a secondary disease), the acting factors, the modality and the causal regularity. A variant of the causal regularity is an implication. Its antecedent can contain a condition on the acting factors and its consequent contains an interval of possible durations for the time lapses between the beginning of the primary disease and the beginning of the secondary disease. The antecedent of such implication is fulfilled if the condition on the acting factors is

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[1] The modality can take one of two values: *necessity* or *possibility*. The value of *necessity* means that if there is a cause or cause-event for a patient then the cause-and-effect relation necessarily takes place. The value of *possibility* means that cause (or cause-event) does not need to rise to the cause-and-effect relation.

[2] A condition on the acting factors has the same structure as a necessary condition. A condition on the acting factors is fulfilled if each its component is fulfilled (if the condition is absent then it is considered to be always fulfilled).

[3] A dynamics period contains an interval of possible durations for this dynamics period and a set of values of the sign which are possible in this period.

fulfilled. The consequent of the implication is fulfilled if the difference between the time moments when the primary disease and the secondary disease began belongs to the interval of possible durations of this time lapse.

The description of an etiology includes the references to the cause-event, the effect (a disease), the acting factors, the modality and the causal regularity. Its variant is an implication. Its antecedent contains a condition on the cause-event and can contain a condition on the acting factors. Its consequent contains an interval of possible durations for time lapses between the moment when the cause took place and the beginning of the disease (of the effect). A condition on a cause-event is a subset of the range of the possible values for the event. A condition on a cause-event is fulfilled if the value of the cause-event belongs to this subset. The antecedent of an implication is fulfilled if both the condition on the cause-event and the condition on the acting factors are fulfilled. The consequent of an implication is fulfilled if the difference between the time moments when the cause-event happened and the disease began belongs to the interval of possible durations of this time lapse.

The description of a clinical manifestation includes the references to the cause (a disease), to the effect (a sign) to the acting factors, and also the modality and the causal regularity for every development period of the disease. A variant of the causal regularity is an implication. The antecedent of the implication can contain a condition on the acting factors, and its consequent contains a sequence of dynamics periods. The antecedent of the implication is fulfilled if the condition on the acting factors is fulfilled. The consequent of the implication is fulfilled if there is such a partition of the time interval of the development period for the disease into dynamics periods that the duration of every dynamics period belongs to the interval of possible durations for this period, and all the values of the sign which were examined at the moments from this period belong to the set of values which are possible in this period.

The description of a clinical manifestation modified by an event's influence includes the references to the cause-event, to the cause (a disease), to the effect (a sign), to the acting factors, and also the modality and the causal regularity. Its variant is an implication. The antecedent of the implication contains a condition on the cause-event, and can contain a condition on the acting factors. The consequent of the implication contains a sequence of dynamics periods. The antecedent of the implication is fulfilled if both the condition on the cause-event and the condition on the acting factors are fulfilled. The consequent of the implication is fulfilled if there is such a partition of a time interval, that began at the moment when the event happened and the disease was proceeding, into dynamics periods that the duration of every dynamics period belongs to the interval of possible durations of this period, and the values of the sign which were examined at the time moments from this period belong to the set of values which are possible in this period.

The description of a response to an event's influence includes the references to the cause-event, to the effect (a sign), to the acting factors, and also the modality, and the causal regularity. Its variant is an implication. The antecedent of the implication contains a condition on the cause-event, and can contain a condition on the acting factors. The consequent of the implication contains a sequence of dynamics periods. The antecedent of the implication is fulfilled if both the condition on the cause-event and the condition on the acting factors are fulfilled. The consequent of the implication is fulfilled if there is such a partition of a time interval, that began at the moment when the event happened, into dynamics periods that the duration of every dynamics period belongs to the interval of possible durations for this period, and the values of the sign which were examined at the moments from this period belong to the set of values which are possible in this period.

The description of a normal reaction consists of the references to the effect (a sign), to the acting factors, and also of the causal regularity. Its variant is an implication. Its antecedent can contain a condition on the acting factors, and its consequent contains the set of the normal values of the sign (of the effect). The antecedent of the implication is fulfilled if the condition on the acting factors is fulfilled. The consequent of the implication is fulfilled if all the values of the sign which were examined at the moments from the interval when the normal reaction acted belong to the set of the normal values.

## 2. An extension of the language of applied logic and an applied logic theory which are used in this article

In the article the model of the metaontology of medical diagnostics is represented in the language of applied logic [10] with the use of extensions which were described in the same article. In addition, another specialized extension of the language that is called *categories* is introduced below. Also at this point a modernized variant of the applied logic theory that is called *a definition of partitions* is presented. The original variant of this theory was given in [10].

### 2.1. The extension "Categories"

2.1.1. The construction $(s_1 \to t_1, s_2 \to t_2, \ldots, s_m \to t_m)$ is a term of this extension. Here $s_1, s_2, \ldots, s_m$ are names, and $t_1, t_2, \ldots, t_m$ are terms. The values of these terms are sets. The value of this term is the set of structural values that is the domain of all the possible mappings with names $s_1, s_2, \ldots, s_m$. The ranges of these mappings are the values of terms $t_1, t_2, \ldots, t_m$ respectively.

2.1.2. The mappings with names $s_1, s_2, \ldots, s_m$ are called *attributes*, and the values of these mappings for a concrete structural value are called *the values of the attributes* for this structural value.

2.1.3. If x is a structural value that belongs to the value of term $(s_1 \to t_1, \ldots, s_i \to t_i, \ldots, s_m \to t_m)$ then any $s_i$ which is a part of terms $t_1, \ldots, t_m$ is considered as a term. The value of this term is the same as the value of term $s_i(x)$.

### 2.2. The applied logical theory "Definition of partitions"

The applied logical theory *Definition of partitions*(ST, *Intervals*, *Mathematical quantifiers*) contains only the descriptions of name's values:

2.2.1. *"Partitions"* is the set of all possible partitions for the set of nonnegative integers. Every partition is a finite strictly increasing sequence.

$$partitions \equiv (\cup \text{ (length: I[0, } \infty)) \{(\text{sequence: I} \Uparrow \text{ (length+1)) (\&(element : I[1, length])}$$
$$\pi(\text{element, sequence}) < \pi(\text{element+1, sequence}))\})$$

2.2.2. *"Element"* is a function; its arguments are a partition and an integer in the range from 0 to the number of elements in the partition; its result is the element of this partition with the number which is equal to the second argument.

$$element \equiv (\lambda(\text{partition: } partitions) \text{ (element: I[0, } length(\text{partition})-1)]) \ \pi(\text{element+1, partition}))$$

2.2.3. *"Interval"* is a function; its arguments are a partition and a positive integer which is less than the number of elements in this partition; its result is the interval of nonnegative integers between the element of this partition with the number which is equal to the second argument and the element with the previous number.

$$interval \equiv (\lambda(\text{partition: } partitions) \text{ (element: I[1, } length(\text{partition})-1)])$$
$$\text{I}[element(\text{partition, element}-1), element(\text{partition, element})])$$

## 3. The basic concepts and ontological agreements which define knowledge and reality of the domain

In this section all the classes of observations and diseases, and also the concepts of knowledge and reality which are related to them are described.

### 3.1. The basic concepts and ontological agreements which define knowledge of the domain (the parameters of the metaontology model)

In this section the basic terms which are used for description of the domain knowledge, and also the restrictions on their values are introduced. These restrictions do not depend on the values of the terms for defining reality.

3.1.1. "*Signs*" is a class of concepts which correspond to observable processes. The values of signs are determined by one of four possible cause-and-effect relations. Knowledge must contain at least one sign.

sort *signs*: {}N \ {∅}

3.1.2. "*Events*" is a class of concepts which correspond to events that can happen to patients, and that should be taken into account during diagnosing.

sort *events*: {}N

3.1.3. "*Features*" is a class of concepts which correspond to anatomical-and-physiological features of patients that should be taken into account during diagnosing.

sort *features*: {}N

3.1.4. The names of all the signs, events and features are different.

signs ∩ events = ∅ & features ∩ events = ∅ & signs ∩ features = ∅

3.1.5. "*Observations*" is the set of all the signs, events and features.

observations ≡ signs ∪ events ∪ features

3.1.6. "*Sets of values*" is the set of all the admissible sets of scalar values.

sets of values ≡ {}N \ {∅}

3.1.7. The values do not coincide with the names of observations.

observations ∩ ( ∪ (set: sets of values) set) = ∅

3.1.8. "*Possible values*" is a function that takes an observation and returns its possible value range.

sort possible values: observation → sets of values

3.1.9. Every observation has no less than two values.

(observation: *observations*) μ(*possible values*(observation)) ≥ 2

3.1.10. "*Conditions*" is the set of all possible conditions. It is the set of the sets consisting of structural values. Every condition is a finite set of structural values. Every such a structural value has attributes which are called *feature* и *range of values*. The value of the first one is the name of a feature, and the value of the second one is a proper subset of the possible values of this feature. The empty set represents the identically true condition.

conditions ≡ {}{(condition: (feature → features, range of values → sets of values))

range of values(condition) ⊂ possible values(feature(condition))}

3.1.11. "*Necessary condition*" is a function that takes a sign and returns a condition that is necessary so that the sign can be examined in the situation.

sort necessary condition: signs → conditions

3.1.12. "*Diseases*" is a class of concepts corresponding to the diseases which have their descriptions in knowledge. Knowledge must contain the description of at least one disease.

sort *diseases*: {}N \ {∅}

3.1.13. Every term from the *diseases* is a structural value with three attributes. They are *number of development periods*, *development periods* and *necessary condition*. The value of the first one is a positive integer, the value of the second one is a function that takes the number of a development period and returns an *interval*, and the value of the third attribute is a condition that is necessary so that this disease took place in the situation (if the value of this attribute is the empty set then the condition is considered to be true).

(disease: *diseases*) sort disease:

(*necessary condition* → *conditions*, *number of development periods* → I[1, ∞),

*development periods* → (I[1, *number of development periods*] → interval) )

3.1.14. "*Dynamics periods*" is a set of structural values with two attributes. They are *duration* and *range of effect*. The value of the first one is an interval, and the value of the second one is a set of values.

*dynamics periods* ≡ (*duration* → interval, *range of effect* → sets of values)

3.1.15. "*Interval*" is a set of structural values with two attributes. They are *lower bound* and *upper bound*. Their values are positive integers which are minimal and maximal durations of the interval. The duration is measured in positive integers, and the lower bound is less than the upper bound.

*interval* ≡ (*lower bound* → I[1, ∞), *upper bound* → I[*lower bound* + 1, ∞))

### 3.2. The basic concepts and ontological agreements which define the reality of the domain (the unknowns of the metaontology model)

Reality in medical diagnostics is considered as the set of the situations corresponding to diagnostic cases (patients). In this section the basic concepts for situation's descriptions and the restrictions on these values are introduced.

3.2.1. "*Moments*" is a function that takes a sign or an event and returns a set of nonnegative integers which are time moments in a situation when the sign was examined or the event happened. Every number means the amount of hours from the beginning of the examination to the moment when the sign was examined or the event happened. If for a sign the value of this function is the empty set then the sign was not examined for the patient. If the same takes place for an event then the event did not happen in the situation.

sort *moments*: *signs* ∪ *events* → {}I [0, ∞)

3.2.2. Every term from the class of *signs* is a function that takes a time moment of examining this sign and returns the value of this sign at this moment in the situation. Every term from the class of *events* is a function that takes a time moment when the event happened and returns the value of this event at this moment in the situation.

(sign or event: *signs* ∪ *events*) sort sign or event: *moments*(sign or event) → *possible values*(sign or event)

3.2.3. "*Examined features*" is the set of features which were examined in the situation.

*sort examined features*: {}*features*

3.2.4. Every term from the class of *examined features* has the value that this feature has in the situation.

(feature: *examined features*) sort feature: *possible values*(feature)

3.2.5. "*Fulfilled*" is a predicate that takes an element of the set of *conditions* and returns *truth* if and only if for every component of this element which is an examined feature the value of the first attribute of the structural value (*feature*) belongs to the value of the second attribute (*range of values*) in the situation. The empty condition is identically *true*.

*fulfilled* ≡ (λ (condition: *conditions*) condition ≠ ∅ ⇒ (& (component: condition) *feature*(component) ∈

∈ *examined features* ⇒ j(*feature*(component)) ∈ *range of values*(component) ))

3.2.6. If in the situation a sign was examined at least once then the necessary condition of this sign must be fulfilled.

(sign: *signs*) *moments*(sign) ≠ ∅ ⇒ *fulfilled*(*necessary condition*(sign))

3.2.7. "*Diagnosis*" is the set of diseases with which a patient is ill. If a patient is healthy his or her diagnosis is the empty set.

*sort diagnosis*: {}*diseases*

3.2.8. If a disease belongs to the patient's diagnosis then the necessary condition for this disease must be fulfilled.

(disease: *diagnosis*) *fulfilled*(*necessary condition*(disease))

3.2.9. "*Development*" is a function that takes a disease from the diagnosis or an examined sign. In the first case this function returns a partition of the time axis. Every interval of this partition corresponds to a development period of this disease. In the second case the function also returns a partition of the time axis. During each interval of this partition the values of the sign are determined in the situation by a common cause, which is associated with this interval.

sort *development*: *diagnosis* $\cup$ {(sign: *signs*) *moments*(sign) $\neq \varnothing$} $\rightarrow$ *partitions*

3.2.10. The interval during which the development of a sign is observed covers all the examination moments of this sign.

(sign: *signs*) *moments*(sign) $\neq \varnothing \Rightarrow$ *element*(*development*(sign), 0) $\leq$ inf(*moments*(sign)) &

& *element*(*development*(sign),*length*(*development*(sign))) $\geq$ *sup*(*moments*(sign))

3.2.11. "*Development intervals of sign*" is a set of structural values that consist of two attributes. They are *sign* and *number of interval*. The value of the first attribute is the name of sign and the value of the second one is the number of its development interval.

*development intervals of sign* $\equiv$ (*sign* $\rightarrow$ *signs*, *number of interval* $\rightarrow$ I[1,*length*(*development*(*sign*))–1] )

3.2.12. If a disease belongs to the diagnosis then the number of development periods of this disease in the situation is the same as the number of its development periods in the knowledge base. The duration of each development period in the situation is between the lower and upper bounds for the duration of this development period.

(disease: *diagnosis*) *length*(*development*(disease)) = *number of development periods*(disease)+1 &

& (& (number of a development period: I[1, *length*(*development*(disease)) – 1])

*element*(*development*(disease), number of a development period) –

– *element*(*development*(disease), number of a development period – 1) $\in$

$\in$ I[*lower bound*(*development periods*(disease)(number of a development period),

*upper bound*(*development periods*(disease)(number of a development period)] )

## Conclusion

In this article an informal description of a metaontology of medical diagnostics of acute diseases has been presented. In this metaontology interaction of cause-and-effect relations of different types are taken into account. This metaontology is close to real ideas of the medical diagnostics in the Russian Federation. It defines combined and complicated pathology, the development of pathological processes in time and also the influence of treatment and other events on the manifestation of diseases. In addition, a part of the metaontology model is presented. This part includes the definitions terms of the knowledge model (parameters), the definitions of terms of the reality model (unknowns) and the unenriched logical relationships system consisting of the integrity restrictions for unknowns and parameters.

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