

## Modelling functions in bio-medicine

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**Background** A central notion in biomedical research is the concept "function". Uses of this concept include the Gene Ontology [1], which includes a "Molecular Functions" taxonomy. The field of functional genomics is dedicated to finding the function of genes and their products. However, the question how something obtains a function or even what a function is, is not precisely defined. Such a model is, however, needed in order to find common errors in the conceptual modelling of function and the relation of functions to other entities in the world, such as processes or physical structures.

We introduce an ontological model of functions, based on the work of [2], and show how it can be applied to biology, medicine and in biomedical ontologies.

**Methods** In an adaption of [4, 5], we consider functions as the abstraction of biological processes or other entities towards a goal: when X has the function Y with the goal Z, then X is supposed to cause or otherwise bring about the state of the world Z, thus realizing Y.

Three major issues concerning functions are addressed:

1. *function structure*: how to represent and determine functions independently of their realizations
2. *realization*: the conditions under which a given entity realizes a function
3. *has-function relation*: the determination of the notion of an entity having a function

Functions can be represented independently of their realizations. The structure of a function consists of a set of *labels*, a set of *requirements*, a *goal*, and a *functional item*. *Labels* are natural language expressions which name the function. The *requirements* of the function are the preconditions for the function realization. Functions are teleological – *goal-oriented* – entities. A goal is a part of the world which is intended to be brought about by a realization of the function. The goal may specify only a small, incomprehensible part of reality. Therefore, a *final state* situates the goal in a wider context, a situation. A situation is a structured, comprehensive part of reality. Functions are always the function of some other entity. The *functional item* is a role [3] played by this entity in any realization of the function.

Let us now focus on the second issue addressed, the realization. A realization of a function is a transition of one state of the world, satisfying the requirements, to another state of the world, satisfying the goal. Functions and their realizations must not be confused. The specification of a function answers the question "what" is done, the specification of a realization answers the question "how".

As the third issue, the has-function relation is a ternary relation between a function, an individual and a context. The relation between the first two arguments is, that the individual is involved in a realization of the function as its functional item. The third argument captures the notion that a function is always ascribed in some context, where a context is considered as a situation (e.g. a structured part of the world).

**Results** We show how to use an elaborated notion of "function" to expand on currently unanalyzed occurrences of "function" in biomedical ontologies. An analysis of the use of "function" in biomedical ontologies reveals new relationships between the currently disjoint graph structures representing the ontologies, and new entities which are not covered by these ontologies. For example, the "is-realization" relation can now be modelled, as a new relation between a function and a process. Furthermore, physical entities such as cells may have a function, modelled by the has-function relation. Therefore, use of our model serves to discover new relations and entities, and thus complete biomedical ontologies. The model provided can also be used to analyze biomedical ontologies and search for missing or malformed categories: assuming that each function must be realized by some process, we can discover new processes, or, if there already is a process we believe to be realizing a function, verify whether the definition and description of the process satisfy this belief.

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