

Formal and Computable Representations of Allergic Diseases in the Electronic Health Record: An Approach Based on the Ontology of General Medical Science

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Background

Increasing numbers of allergist-immunologists are using electronic health records (EHR), as they are thought to have applications in facilitating provider communication and interoperability of systems, compliance with reporting requirements, evaluating provider performance, decision support, and data-mining in research studies. Formal and computable representation of medical data and knowledge is fundamental in establishing a common vocabulary and semantics to support these applications. Unfortunately, many existing biomedical terminology standards rely on "incomplete, inconsistent or confused definitions of terms pertaining to diseases, diagnoses, and clinical phenotypes" (1). Further, standard coding systems change slowly in response to scientific advances, and so are inadequate when measured against the needs of clinical and translational science.

Methods

We present our results in the form of terms and definitions designed to provide a consistent starting point for an incrementally evolving representation of both allergic disease and diagnosis in EHRs. We created new definitions drawing on best practices in ontology development and consistent with the current definitions of disease, diagnosis, disorder, and syndrome as presented in the Ontology of General Medical Science (OGMS)(1,3) and definitions of more general entities in the Basic Formal Ontology (BFO) (2).

BFO top-level entities definitions²

Continuant =def. An entity [bfo:Entity] that exists in full at any time in which it exists at all, persists through time while maintaining its identity and has no temporal parts.

Occurrent =def. An entity [bfo:Entity] that has temporal parts and that happens, unfolds or develops through time. Sometimes also called perdurants.

OGMS definitions^{1,3}

Disease =def. A disposition to undergo pathological processes that exists in an organism because of one or more disorders in that organism. A *disposition* is an attribute of an organism in virtue of which it will initiate certain specific sorts of processes when certain conditions are satisfied.

Disorder =def. A causally relatively isolated combination of physical components that is (a) clinically abnormal and (b) maximal, in the sense that it is not a part of some larger such combination.

Diseases are related to disorders by the ogms:*has-material-basis* relation.

Results

Allergy Ontology definitions

Allergic disease =def. a disposition to realize allergic processes under certain conditions (e.g. exposure to triggering allergen), i.e. a symptomatic immune reaction is made to a normally innocuous environmental antigen (4). All instances of allergic disease [ao: allergic disease] are also instances of ogms:disease, and therefore are also dispositions.

IgE-mediated allergic disease =def. allergic diseases [ao:allergic disease] that are IgE-mediated, i.e. whose material basis is an IgE-mediated allergic disorder

IgE-mediated allergic disorder = def. a material entity (a disorder) in which allergen-specific IgE are present in an organism and are bound to plasma membrane-bound FcεRI receptors on mast cells and basophils.

Anaphylaxis =def. a severe, life threatening, generalized or systemic hypersensitivity reaction (5,6). A [bfo:Occurrent].

Allergic anaphylaxis (Immunologic anaphylaxis) =def. anaphylaxis that is mediated by an immunologic mechanism, e.g. IgE, IgG, and immune-complex complement-related.(5,6)

Pseudoallergic anaphylaxis (non-immunologic anaphylaxis) =def. anaphylaxis that is not mediated by an immunologic mechanism.(5,6)

Discussion

Our work describes an extension to the approach taken in the OGMS, which is designed to be a logically and biologically coherent framework for the representation of biomedical entities and relations between them. OGMS adopts a view of disease as a disposition toward the realization of pathological processes, and involving some physical basis within the organism that bears the disease (1). In this work, we focus on IgE-mediated allergic disease and its underlying material basis (the disorder). We also provide an account of the term allergic anaphylaxis as a severe pathological process. We were able to construct a basic ontological framework out of several basic terms in allergy that extend the approach taken in OGMS, including formal, logically coherent and computable definitions of IgE-mediated allergic disease, IgE-mediated allergic disorder and IgE-mediated allergic pathologic process.

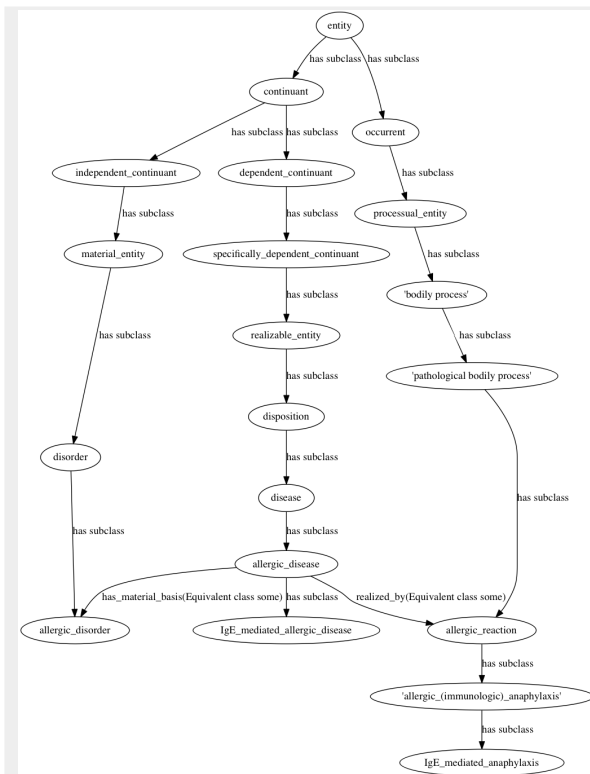


Fig 1. A partial graph of the Allergy Ontology.

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