International Consortium for Human Phenotype Terminologies (ICHPT)

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Research Areas

Data-Sharing Enabler

At a Glance

- Status: **Completed Consortium**
- Year Launched: **2012**
- Initiating Organization: **The American Society of Human Genetics**
- Initiator Type: **Third-party organization**
- **Rare** disease
- Location: **International**

Abstract

The International Consortium for Human Phenotype Terminologies (ICHPT) was created with input from several groups including primarily Orphanet, the Human Phenotype Ontology (HPO) team, and the Online Mendelian Inheritance in Man (OMIM) team. ICHPT has provided a mapping between the core set of terms in the HPO, PhenoDB, Orphanet, Elements of Morphology, POSSUM, SNOMED, MeSH, and MeDRA. Additionally, the HPO is currently being incorporated into the UMLS (United Medical Language System).

Mission

ICHPT is working to standardize a core set of terminology across the major different systems in use. At a workshop preceding the American Society of Human Genetics meeting in Boston in October 2013, representatives of the HPO team, PhenoDB, Orphanet, Elements of Morphology, and others came together to align more than 2,300 core terms across all phenotype ontologies. All tools using these ontologies will map to these terms to allow cross-compatibility between systems, and SNOMed-CT will

also be requested to implement the full set of aligned terms. Where ontologies contain more detailed terms at a finer level of granularity, these terms will map “up” to the broader aligned terms.

Consortium History

In 2012, Ségolène Ayme convened a panel of clinical geneticists and representatives from major databases such as the London Dysmorphology Database, the HPO, and Orphanet, to work toward standardization of lists of human phenotypes broken down by affected systems, associated disorders, and related genes and variants.

In 2012, ICHPT brought together every major organization doing research on rare genetic diseases for a 17-hour session to discuss preferred terms, working through a list of more than 2,700 phenotypes from six different databases.

Impact/Accomplishment

Phenotype terminologies in use for genotype-phenotype databases: A common core for standardization and interoperability. P. N. Robinson, S. Aymé, L. Chanas, A. Hamosh, A. Rath, International Consortium for Human Phenotype Terminologies, Institute for Medical Genetics, Charite-Universitätsmedizin, Berlin, Germany; INSERM, US14, Paris, France; and McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University School of Medicine, Baltimore, MD.

Links/Social Media Feed

Other website	http://www.bio-itworld.com/2013/10/30/how-accelerate-rare-disease-research-write-dictionary.html

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