Guest Editorial

Introduction: International Medical Informatics Association Working Group 6 and the 2005 Rome Conference

This issue of the *Journal of Biomedical Informatics* is devoted to the topic of biomedical ontologies. It begins with a survey of the field, followed by a collection of papers growing out of an international conference on this topic, which was held in Rome from 29 April to 2 May 2005. The conference was organized by Working Group 6 of the International Medical Informatics Association (IMIA) and co-sponsored by the European Federation for Medical Informatics, the Institute for Formal Ontology and Medical Information Science, and the European Union Network of Excellence on Semantic Interoperability and Data Mining in Biomedicine.

IMIA's Working Group 6 was formed in 1981, with the task of reviewing the health data nomenclature and classification needs of the international community, evaluating information processing technology in meeting these defined needs, and recommending methods for future classification and nomenclature systems. Roger A. Côté, the founding editor of SNOMED, was founder and first chair of WG6, and the proceedings of the first WG6 conference, which took place under Côté's direction in Ottawa, Canada in 1984, served to define the field [1]. The second WG6 conference, held in Geneva in 1988, was directed by Jean-Raoul Scherrer under the sponsorship of the World Health Organization [2]. The next conference, also under Scherrer's direction, was held in Vevey, Switzerland in 1994 [3]. Conference number four took place in Jacksonville, Florida in 1997 under the directorship of Christopher Chute, with support from the Jacksonville Mayo Clinic [4]. The fifth conference occurred in Phoenix, Arizona in 1999, again under Chute's direction, and it continued a successful tradition, which has featured the most influential and creative scientists in the field of medical concept representation.

The Rome conference, which was organized under the scientific direction of Barry Smith, one of the two co-editors of this issue of JBI, sought to continue this tradition, but in a way that reflects the new integrating role played by ontology in biomedical informatics (Fig. 1). The meeting was designed to foster closer integration of thinking on biomedical ontologies at an international level and to explore fundamental issues at the cutting edge of biomedical ontology research, focusing on theoretical developments rather than operational questions. Participants were encouraged to adopt a longer-term (and even what some might call a "philosophical") perspective on questions pertaining to the development and use of biomedical ontologies in the future.

In what seems to have proved a successful experiment (inspired by practices more common at philosophical conferences), presentations were kept deliberately short, and the number of papers kept small. Each main paper was followed by a presentation by a 'respondent,' who had access to the paper prior to the conference and provided an oral response to the positions taken by its author. Respondents were deliberately chosen to represent different schools of thought, which might be roughly divided into 'philosophicals' on the one hand and 'practicals' on the other (or alternatively into: those who think that progress in biomedical informatics can benefit from—and may even require—the help of philosophers, and those who are still skeptical about such potential benefits). Following each such paired presentation, a large amount of time was set aside for the discussion of the theses and counter-theses advanced by the presenters and respondents.

Following the conference, both primary presenters and respondents were invited to submit their papers, modified on the basis of discussions at the meeting, to the JBI for peer evaluation. In several cases, presenters and respondents coordinated their views into a single paper, sometimes involving also other conference participants. This special issue presents those papers.

The paper by Alexander Yu [5] provides a survey of the field, drawing particular attention to the degree to which, with the growth in importance of ontology, there has arisen also an increasing realization of the difficulties involved in creating ontologies which are at one and the same time of high quality and of practical usefulness in the biomedical domain.

Alexa McCray sketches the historical background of the rise of ontology, pointing to the role of philosophers

Fig. 1. Group photograph of the 2005 IMIA Working Group 6 Conference attendees; conference host, Domenico Pisanelli, is shown in the inset at lower left.

in this history, and also making clear the degree to which ontologies and taxonomies have differed widely over time as a reflection of the different world views of their designers [6]. Where McCray draws pessimistic conclusions from this seemingly unconstrained diversity, arguing that ontologies must of necessity manifest one or other type of bias, the philosopher Ingvar Johansson, McCray's respondent in Rome, insists in his contribution that ontologies can comprehend knowledge about a mind-independent biological reality, and that ontology developers should always keep in mind the question of whether the terms they use in their ontologies correspond to referents in reality [7].

In Barry Smith's paper [8], this need to establish a relationship between terms in an ontology and corresponding universals or types in reality is transformed into a tool for creating and maintaining ontologies and terminologies themselves. Smith's idea is that reality itself should serve as a benchmark for the correctness of biomedical terminologies. He attacks along the way the so-called "concept orientation" in terminology development in the biomedical and other domains, arguing that it creates a redundant intermediary layer between terms and reality in a way which often brings confusion. James Cimino, in contrast [9], defends the concept orientation, arguing that both concepts and universals are needed and that they can coexist. This leads him to define additional desiderata which must be satisfied by controlled terminologies if they are to realize the variety of practical purposes for which they are designed.

Anita Burgun's paper [10] approaches the issue of desiderata for biomedical ontologies from the perspective of how such ontologies should be designed if they are to be of maximum value for the purposes of scientific research.

She considers two biomedical reference ontologies, in anatomy and chemistry, in light of the qualities they must have if they are to be used as a basis for drawing new inferences about more complex entities.

Judith Blake and Carol J. Bult [11] contribute a response from the side of the Gene Ontology to the problems posed by the massive expansion of genome-scale data. They show how bio-ontologies have an important role to play in supporting effective exploitation of such data; at the same time they show that the rapidly changing nature of biology has created obstacles which must be surmounted if such ontologies are to be used for purposes of dynamic reasoning.

The contribution of Christiane Fellbaum, Udo Hahn, and Barry Smith [12] combines the presentation by Fellbaum and the response by Hahn to address the potential of WordNet, an influential digitalized lexicon, to support the creation of new kinds of information resources in the domain of consumer health. In particular the authors describe how an improved version of WordNet might be applied to the study of differences between expert and non-expert belief systems and of the communication problems which such differences are known to bring in their wake.

Alan Rector's presentation and Thomas Bittner's response are combined into the paper by Rector, Jeremy Rogers, and Bittner [13]. This addresses the issues surrounding the treatment of granularity and scale in biomedical informatics, combining this with a discussion of how relative and absolute size, and also phenomena such as aggregations, mixtures, density and connectivity, should be treated in structured representations of medical reality.

One important question in this connection concerns the relation of parthood between different kinds of collective

and non-collective entities, a relation which is at the center, too, of the paper by Stefan Schulz, Anand Kumar, and Thomas Bittner, which combines Schulz's presentation and Kumar's response [14]. This paper addresses parthood relations involving artifacts and biological entities (is your heart pacemaker a part of your body, or is it merely spatially located within it?). It addresses not only parthood relations involving continuant entities (for example, the molecules inside your body), but also the relations between biological processes and subprocesses in the realm of occurrents.

The issue concludes with an expanded statement of the theory of referent tracking in electronic health records expounded in the presentation by Werner Ceusters. Ceusters and Smith [15] show how such records have thus far done a poor job in identifying the concrete entities relevant to healthcare—which include not only particular patients but also their parts, diseases, therapies, and lesions. They describe a regime in which all such entities would be referred to directly by means of unique tracking numbers, and argue that this would bring a radically new perspective to the domain of clinical coding and terminology development.

In addition to the authors of the papers included in this issue, the speakers and respondents at the Rome meeting were: Robert Baud (Geneva), Olivier Bodenreider (NLM), Christopher G. Chute (Mayo Clinic), Stan Huff (Utah), Suzanna Lewis (Berkeley, California), Mark Musen (Stanford), Domenico Pisanelli (Rome), Jean-Marie Rodrigues (St. Etienne), Cornelius Rosse (Seattle), Robert Stevens (Manchester), György Surján (Budapest), and Pierre Zweigenbaum (Paris). Other participants included: Hans Ahlfeldt (Linköpping), Iulian Alecu (Paris), M. Juan Bonal (Thales IS), Jack Bowie (Apelon), Hans Gill (Linköpping), Louis Goldberg (Buffalo), Marie-Christine Jaulent (Paris), Marijke Keet (Bolzano), Rüdiger Klar (Freiburg), Kathy Lesh (Kevric), Jan-Eric Litton (Stockholm), Dirk Marwede (Leipzig), Kazuhiko Ohe (Tokyo), Matti Ojala (Helsinki), David Ouagne (Paris), Martin Romacker (Freiburg), Harold Solbrig (Mayo Clinic), Windy C. Thompson (GlaxoSmithKline), Anders Thurin (Göteborg), and Sumi Yoshikawa (Yokohama).

We extend our thanks to all these persons for their contributions to lively and fruitful discussions, and to all of those meeting attendees who served as reviewers for this special issue. Our thanks go also to the sponsoring organizations for their generous support.

References

[1] Côté RA, Protti DJ, Scherrer J-R, editors. The role of informatics in health data coding and classification systems. Amsterdam: Elsevier Science; 1985.

[2] Scherrer J-R, Côté RA, Mandil SH, editors. Computerized natural medical language processing for knowledge represe. Amsterdam: Elsevier Science; 1989.

[3] McCray AT, Safran C, Chute C, Scherrer J-R, editors. Natural language and medical concept representation, 34(1/2). Methods of Information in Medicine; 1995.

[4] Chute CG, Baud RH, Cimino JJ, Patel VL, Rector AL, editors. Coding and language processing, 37(4–5). Methods of Information in Medicine; 1998.

[5] Yu A. Methods in biomedical ontology. J Biomed Inform 2006;39:252–66.

[6] McCray AT. Conceptualizing the world: lessons from history. J Biomed Inform 2006;39:267–73.

[7] Johansson I. Bioinformatics and biological reality. J Biomed Inform 2006;39:274–87.

[8] Smith B. From concepts to clinical reality: an essay on the benchmarking of biomedical terminologies. J Biomed Inform 2006;39:288–98.

[9] Cimino JJ. In defense of the desiderata. J Biomed Inform 2006;39:299–306.

[10] Burgun A. Desiderata for domain reference ontologies in biomedicine. J Biomed Inform 2006;39:307–13.

[11] Blake J, Bult CJ. Beyond the data deluge: data integration and bio-ontologies. J Biomed Inform 2006;39:314–20.

[12] Fellbaum C, Hahn U, Smith B. Towards new information resources for public health-From WordNet to MedicalWordNet. J Biomed Inform 2006;39:321–32.

[13] Rector AL, Rogers J, Bittner T. Granularity, scale, and collectivity: when size does and doesn't matter. J Biomed Inform 2006;39:333–49.

[14] Schulz S, Kumar A, Bittner T. Biomedical ontologies: what part-of is and isn't. J Biomed Inform 2006;39:350–61.

[15] Ceusters W, Smith B. Strategies for referent tracking in electronic health records. J Biomed Inform 2006;39:362–78.

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