CONFERENCE PROCEEDINGS

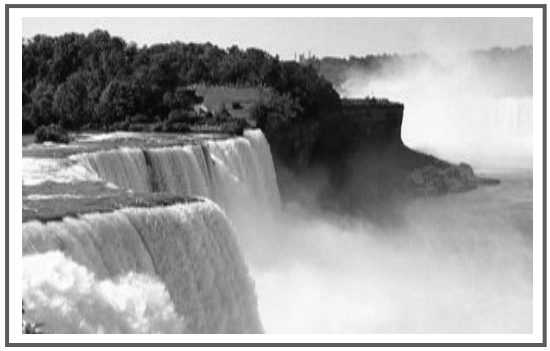
ICBO

International Conference on Biomedical Ontology

July 28-30, 2011
Buffalo, New York, USA

SPONSORED BY
University at Buffalo College of Arts and Sciences
National Center for Ontological Research
National Center for Biomedical Ontology
with support from
National Human Genome Research Institute

CONFERENCE PROCEEDINGS



ICBO

International Conference on Biomedical Ontology

July 28-30, 2011
Buffalo, New York, USA

Sponsored by:

University at Buffalo The State University of New York

ncor national center for ontological research

THE NATIONAL CENTER FOR BIOMEDICAL ONTOLOGY

University at Buffalo College of Arts and Sciences, National Center for Ontological Research, National Center for Biomedical Ontology, Computer Task Group, Cognigen Corporation, IMIA WG6 and the Protein Ontology

We would also like to acknowledge generous support from the National Human Genome Research Institute (NHGRI)

Funding for this conference was made possible in part by the United States National Institutes of Health (NIH) through the NIH Roadmap for Medical Research, Grant 1 U54 HG004028, and by 1 R13 HG006231-01 from the National Human Genome Research Institute (NHGRI). The views expressed in written conference materials or publications and by speakers and moderators do not necessarily reflect the official policies of the Department of Health and Human Services or the National Institutes of Health; nor does mention by trade names, commercial practices, or organizations imply endorsement by the U.S. Government. Information on the National Centers for Biomedical Computing can be found at http://nihroadmap.nih.gov/bioinformatics.

ICBO ORGANIZING COMMITTEE

Conference Chair: Barry Smith, University at Buffalo, Buffalo, NY, USA

Judith A. Blake, The Jackson Laboratory, Bar Harbor, ME, USA
Suzanna E. Lewis, Lawrence Berkeley National Laboratory, Berkeley, CA, USA
Mark A. Musen, Stanford University, Stanford, CA, USA
Alan Ruttenberg, University at Buffalo, Buffalo, NY, USA
Susanna-Assunta Sansone, Oxford University, Oxford, UK
Dagobert Soergel, University at Buffalo, Buffalo, NY, USA
Christian J. Stoeckert Jr., University of Pennsylvania, Philadelphia, PA USA

ICBO PROGRAM COMMITTEE

Program Committee Chair: Alan Ruttenberg, University at Buffalo, Buffalo, NY USA
Co-Chair: Olivier Bodenreider, NIH National Library of Medicine, Bethesda, MD, USA
Co-Chair: Maryann E. Martone, University of California at San Diego, San Diego, CA, USA
Workshops and Tutorials Chair: Stefan Schulz, Medical University of Graz, Austria
Doctoral and Postdoctoral Consortium Chair: Albert Goldfain, University at Buffalo, Buffalo, NY, USA
Software Demonstrations Chair: Patricia L. Whetzel, Stanford University, Stanford, CA, USA

Colin Batchelor, Royal Society of Chemistry, Cambridge, UK
Thomas Beale, Ocean Informatics
Judith A. Blake, The Jackson Laboratory, Bar Harbor, ME, USA
Sebastian Brandt, University of Manchester, Manchester, UK
Werner Ceusters, University at Buffalo, Buffalo, NY, USA
Rex Chisholm, Northwestern University, Chicago, IL, USA
Jim Cimino, National Institutes of Health, Bethesda, MD, USA
Mélanie Courtot, British Columbia Cancer Research Centre, Vancouver, BC, Canada
Lindsay G. Cowell, University of Texas Southwestern Medical Center at Dallas, Dallas, TX, USA
Alexander D. Diehl, University at Buffalo, Buffalo, NY, USA
Michel Dumontier, Carleton University, Ottawa, ON, Canada
Louis J. Goldberg, University at Buffalo, Buffalo, NY, USA
Janna Hastings, European Bioinformatics Institute, Hinxton, UK
Pascal Hitzler, Wright State University, Dayton, OH, USA
Robert Hoehndorf, University of Cambridge, Cambridge, UK
Larry Hunter, University of Colorado, Denver, CO, USA
Marijke Keet, School of Computer Science, University of KwaZulu-Natal, Durban, South Africa
Jobst Landgrebe, Cognotekt UG, Cologne, Germany
Suzanna E. Lewis, Lawrence Berkeley National Laboratory, Berkeley, CA, USA
Philip Lord, Newcastle University, Newcastle upon Tyne, UK
Jose L. V. Mejino, Jr., University of Washington, Seattle, WA, USA
Alexa McCray, Harvard Medical School, Boston, MA, USA
Christopher J. Mungall, Lawrence Berkeley National Laboratory, Berkeley, CA, USA
Mark A. Musen, Stanford University, Stanford, CA, USA
Darren Natale, Georgetown University, Washington, DC, USA
Chimezie Ogbuji, Case Western Reserve University, Cleveland, OH, USA
Helen Parkinson, European Bioinformatics Institute, Hinxton, UK
Bjoern Peters, La Jolla Institute for Allergy and Immunology, San Diego, CA, USA
Daniel Rubin, Stanford University, Stanford, CA, USA
Peter Robinson, Charité Hospital, Berlin
Susanna-Assunta Sansone, Oxford University, Oxford, UK
Ulrike Sattler, University of Manchester, Manchester, UK
Richard Scheuermann, University of Texas Southwestern Medical Center at Dallas, Dallas, TX, USA
Barry Smith, University at Buffalo, Buffalo, NY, USA
Dagobert Soergel, University at Buffalo, Buffalo, NY, USA
Robert Stevens, University of Manchester, Manchester, UK
Ida Sim, University of California at San Francisco, San Francisco, CA, USA
Harold Solbrig, Mayo Clinic, Rochester, MN, USA
Kent Spackman, International Health Terminology Standards Development Organization
Christian J. Stoeckert Jr., University of Pennsylvania, Philadelphia, PA USA
Cathy Wu, Georgetown University, Washington, DC, USA; University of Delaware, Newark, DE, USA

TABLE OF CONTENTS

Preface iii

ICBO Committees v

ICBO Presentations 1

Where GO is Going and What it Means for Ontology Extension 3
Catia Pesquita, Francisco M. Couto

Revising the Cell Ontology 11
Terrence F. Meehan, Christopher J. Mungall, Alexander D. Diehl

Rapid Development of an Ontology of Coriell Cell Lines 19
Chao Pang, Tomasz Adamusiak, Helen Parkinson, James Malone

Cell Line Ontology: Redesigning the Cell Line Knowledgebase to Aid Integrative Translational Informatics 25
Sirarat Sarntivijai, Zuoshuang Xiang, Terrence F. Meehan, Alexander D. Diehl, Uma Vempati, Stephan Schurer, Chao Pang, James Malone, Helen Parkinson, Brian D. Athey, Yongqun He

Towards a Semantic Web Application: Ontology-Driven Ortholog Clustering Analysis 33
Yu Lin, Zuoshuang Xiang, Yongqun He

HeartCyc: A Cardiac Cycle Process Ontology Based in the Ontology of Physics for Biology 41
Daniel L. Cook, Michal Galdzicki, Maxwell L. Neal, Jose L.V. Mejino, John H. Gennari

Composite Annotation for Heart Development 47
Tariq Abdulla, Ryan Imms, Jean-Marc Schleich, Ron Summers

Ontology-Based Analysis of Event-Related Potentials 55
Gwen Frishkoff, Robert Frank, Paea LePendu

River Flow Model of Diseases 63
Riichiro Mizoguchi, Kouji Kozaki, Hiroko Kou, Yuki Yamagata, Takeshi Imai, Kayo Waki, Kazuhiko Ohe

Dispositions and Processes in the Emotion Ontology 71
Janna Hastings, Werner Ceusters, Barry Smith, Kevin Mulligan

An Advanced Strategy for Integration of Biological Measurement Data 79
Hiroshi Masuya, Georgios V. Gkoutos, Nobuhiko Tanaka, Kazunori Waki, Yoshihiro Okuda, Tatsuya Kushida, Norio Kobayashi, Koji Doi, Kouji Kozaki, Robert Hoehndorf, Shigeharu Wakana, Tetsuro Toyoda, Riichiro Mizoguchi

Annotating Experimental Records Using Ontologies 87
Alexander García, Olga Giraldo, Jael García

Ontology Driven Data Collection for EuPathDB 95
Jie Zheng, Omar S. Harb, Christian J. Stoeckert Jr.

Developing an Application Ontology for Biomedical Resource Annotation and Retrieval: Challenges and Lessons Learned 101
Carlo Torniai, Matt Brush, Nicole Vasilevsky, Erik Segerdell, Melanie Wilson, Tenille Johnson, Karen Corday, Chris Shaffer, Melissa Haendel,

Mapping Composition for Matching Large Life Science Ontologies 109
Anika Gross, Michael Hartung, Toralf Kirsten, Erhard Rahm

Generic Semantic Relatedness Measure for Biomedical Ontologies 117
João D. Ferreira, Francisco M. Couto

Aligning the Parasite Experiment Ontology and the Ontology for Biomedical Investigations Using AgreementMaker 125
Valerie Cross, Cosmin Stroe, Xueheng Hu, Pramit Silwal, Maryam Panahiazar, Isabel F. Cruz, Priti Parikh, Amit Sheth

A Case Study of ICD-11 Anatomy Value Set Extraction from SNOMED CT 133
Guoqian Jiang, Harold R. Solbrig, Robert J.G. Chalmers, Kent Spackman, Alan L. Rector, Christopher G. Chute

The HL7 Approach to Semantic Interoperability 139
Jobst Landgrebe, Barry Smith

Representing the Reality Underlying Demographic Data 147
William R. Hogan, Swetha Garimalla, Shariq A. Tariq

Information Models and Ontologies for Representing the Electronic Health Record 153
Daniel Karlsson, Martin Berzell, Stefan Schulz

Ontology-Based Mammography Annotation and Similar Mass Retrieval with SQWRL 159
Hakan Bulu, Adil Alpkocak, Pinar Balci

The CHRONIOUS Ontology Suite: Methodology and Design Principles 167
Luc Schneider, Mathias Brochhausen

Applying Rigidity to Standardizing OBO Foundry Candidate Ontologies 175
Patrice Seyed, Stuart C. Shapiro

Higgs Bosons, Mars Missions, and Unicorn Delusions: How to Deal with Terms of Dubious Reference in Scientific Ontologies 183
Stefan Schulz, Mathias Brochhausen, Robert Hoehndorf

Towards an Ontology for Conceptual Modeling 191
James P. McCusker, Joanne Luciano, Deborah L. McGuinness

What's in an 'is about' Link? Chemical Diagrams and the Information Artifact Ontology 201
Janna Hastings, Colin Batchelor, Fabian Neuhaus, Christoph Steinbeck

Bioassay Ontology to Describe High-Throughput Screening Assays and their Results 209
Uma Vempati, Ubbo Visser, Saminda Abeyruwan, Kunie Sakurai, Magdalena Przydzial, Caty Chung, Robin Smith, Amar Koleti, Christopher Mader, Vance Lemmon, Stephan Schürer

A Framework Ontology for Computer-Based Patient Record Systems 217
Chimezie Ogbuji

ICBO Posters 225

The Blood Ontology: An Ontology in the Domain of Hematology 227
Mauricio Barcellos Almeida, Anna Barbara de Freitas Carneiro Proietti, Jiye Ai, Barry Smith

Employing Reasoning within the Phenoscape Knowledgebase 230
James P. Balhoff, Wasila M. Dahdul, Hilmar Lapp, Peter E. Midford, Todd J. Vision, Monte Westerfield, Paula M. Mabee

Waiting for a Robust Disease Ontology: A Merger of OMIM and MeSH as a Practical Interim Solution 231
Susan M. Bello, Allan Peter Davis, Thomas C. Wiegers, Mary E. Dolan, Cynthia Smith, Joel Richardson, Judith Blake, Carolyn Mattingly, Janan T. Eppig

Developing a Reagent Application Ontology within the OBO Foundry Framework 234
Matthew H. Brush, Nicole Vasilevsky, Carlo Torniai, Tenille Johnson, Chris Shaffer, Melissa A. Haendel

The Ontology of Microbial Phenotypes (OMP): A Precomposed Ontology Based on Cross Products from Multiple External Ontologies that is Used for Guiding Microbial Phenotype Annotation 237
Marcus Chibucos, Adrienne Zweifel, Deborah Siegele, Peter Uetz, Michelle Giglio, James Hu

Towards an Adverse Event Reporting Ontology 240
Mélanie Courtot, Ryan R. Brinkman

OntoOrpha : An Ontology to Support Edition and Audit of Knowledge of Rare Diseases in ORPHANET 241
Ferdinand Dhombres, Pierre-Yves Vandenbussche, Ana Rath, Marc Hanauer, Annie Olry, Bruno Urbero, Rémy Choquet, Jean Charlet

An Ontology for Gastrointestinal Endoscopy 244
Shahim Essaid

Enriching the Ontology for Biomedical Investigations (OBI) to Improve its Suitability for Web Service Annotations . . 246
Chaitanya Guttula, Alok Dhamanaskar, Rui Wang, John A. Miller, Jessica C. Kissinger, Jie Zheng, Christian J. Stoeckert, Jr.

Recent Developments in the ChEBI Ontology 249
Janna Hastings, Paula de Matos, Adriano Dekker, Marcus Ennis, Kenneth Haug, Zara Josephs, Gareth Owen, Steve Turner, Christoph Steinbeck

Representing Local Identifiers in a Referent-Tracking System 252
William R. Hogan, Swetha Garimalla, Shariq A. Tariq, Werner Ceusters

owl_cpp, a C++ Library for Working with OWL Ontologies 255
Mikhail K. Levin, Alan Ruttenberg, Anna Maria Masci, Lindsay G. Cowell

Towards Ontologies for 'Textbook' of the Future in Obstetrics and Gynecology (OB/Gyn) 258
Yu Lin, Chris Chapman, Erin D. Doelling, Maya Hammoud

Gene Ontology Signatures for Immune Cell-Types Inferred by Gene Expression Analysis 259
Terrence F. Meehan, Christopher J. Mungall, Judith A. Blake, Alexander D. Diehl

Aligning Research Resource and Researcher Representation: The eagle-i and VIVO Use Case. 260
Stella Mitchell, Carlo Torniai, Brian Lowe, Jon Corson-Rikert, Melanie Wilson, Mansoor Ahmed, Shanshan Chen, Ying Ding, Nicholas Rejack, Melissa Haendel, the eagle-i Consortium, the VIVO Collaboration

An Ontology-Based Approach to Linking Model Organisms and Resources to Human Diseases 263
Chris J. Mungall, David Anderson, Anita Bandrowski, Brian Canada, Andrew Chatr-Aryamontri, Keith Cheng, P. Michael Conn, Kara Dolinski, Mark Ellisman, Janan Eppig, Jeffrey S. Grethe, Joseph Kemnitz, Shawn Iadonato, Stephen D. Larson, Charles Magness, Maryann E. Martone, Mike Tyers, Carlo Torniai, Olga Troyanskaya, Judith Turner, Monte Westerfield, Melissa A. Haendel

Using RxNorm to Extract Medication Data from Electronic Health Records in the Rochester Epidemiology Project . 266
Jyotishman Pathak, Shawn P. Murphy, Brian N. Willaert, Hilal M. Kremers, Christopher G. Chute, Barbara P. Yawn, Walter A. Rocca

Towards Desiderata for Provenance Ontologies in Biomedicine 269
Satya S. Sahoo

Modeling Issues and Solutions: Building a Taxonomy from a Biology Textbook 273
Patrice Seyed, John Pacheco, Andrew Goldenkranz, Vinay Chaudhri

Biomedical Analyses: OWL Model Based Edition 276
Pierre-Yves Vandenbussche, Ferdinand Dhombres, Sylvie Cormont, Jean Charlet, Eric Lepage

Ontobee: A Linked Data Server and Browser for Ontology Terms 279
Zuoshuang Xiang, Chris Mungall, Alan Ruttenberg, Yongqun He

ICBO Software Demonstrations. 283

Protein-Centric Connection of Biomedical Knowledge: Protein Ontology (PRO) Research and Annotation Tools . . 285
Cecilia N. Arighi, Darren A. Natale, Judith A. Blake, Carol J. Bult, Michael Caudy, Alexander D. Diehl, Harold J. Drabkin, Peter D'Eustachio, Alexei Evsikov, Hongzhan Huang, Natalia V. Roberts, Alan Ruttenberg, Barry Smith, Jian Zhang, Cathy H. Wu

SHIRAZ and CABERNET: Leveraging Automation, Crowdsourcing, and Ontologies to Improve the Accuracy and Throughput of Zebrafish Histological Phenotype Annotations. 288
Brian Canada, Georgia Thomas, John Schleicher, James Z. Wang, Keith C. Cheng

Biomedical Ontology Matching Using the AgreementMaker System 290
Isabel F. Cruz, Cosmin Stroe, Catia Pesquita, Francisco M. Couto, Valerie Cross

Bioportal: Ontologies and Integrated Data Resources at the Click of a Mouse. 292
Patricia L. Whetzel, Natasha Noy, Nigam Shah, Paul Alexander, Michael Dorf, Ray Fergerson, Margaret-Anne Storey, Barry Smith, Chris Chute, Mark Musen

Populous: A Tool for Populating an Ontology 294
Simon Jupp, Matthew Horridge, Luigi Iannone, Julie Klein, Stuart Owen, Joost Schanstra, Katy Wolstencroft, Robert Stevens

The Vitro Integrated Ontology Editor and Semantic Web Application 296
Brian Lowe, Brian Caruso, Nick Cappadona, Miles Worthington, Stella Mitchell, Jon Corson-Rikert, and VIVO Collaboration

The BioPortal Import Plugin for Protégé 298
Jithun Nair, Tania Tudorache, Trish Whetzel, Natalya Noy, Mark Musen

The Biomedical Ontology Applications (BOA) Framework 300
Bruno Tavares, Hugo P. Bastos, Daniel Faria, João D. Ferreira, Tiago Grego, Catia Pesquita, Francisco M. Couto

The NCBO Annotator: Ontology-Based Annotation as a Web Service 302
Patricia L. Whetzel, Clement Jonquet, Cherie Youn, Michael Dorf, Ray Fergerson, Mark Musen, Nigam Shah

OntoFox and its Application in Development of Brucellosis Ontology 304
Zuoshuang Xiang, Yu Lin, Yongqun He

Pre-ICBO Workshops

Tuesday, July 26, 2011

Representing Adverse Events. 307

AEO: A Realism-Based Biomedical Ontology for the Representation of Adverse Events 309
Yongqun He, Zuoshuang Xiang, Sirarat Sarntivijai, Luca Toldo, Werner Ceusters

Reporting Adverse Events: Basis for a Common Representation 316
Mélanie Courtot, Ryan R. Brinkman, Alan Ruttenberg

Adverse Events from Clinical Studies in Pharmaceutical Research and Development 324
Pia Emilsson, Kerstin Forsberg

Classifying Adverse Events from Clinical Trials 326
Bernard LaSalle, Richard Bradshaw

An Ontological Representation of Adverse Drug Events 329
Guoqian Jiang, Jon D. Duke, Jyotishman Pathak, Christopher G. Chute

Toward Answering Time-Related Questions from Adverse Event Reports Using Ontology-Based Approaches . . 332
Cui Tao, Guoqian Jiang, Kim Clark, Deepak Sharma, Christopher G. Chute

Supporting Medical Device Adverse Event Analysis in an Interoperable Clinical Environment: Design of a Data Logging and Playback System 335
David Arney, Sandy Weininger, Susan F. Whitehead, Julian M. Goldman

Using Failure Modes, Mechanisms, and Effects Analysis in Medical Device Adverse Event Investigations 340
Shunfeng Cheng, Diganta Das, Michael Pecht

Working with Multiple Biomedical Ontologies 347

NIFSTD and NeuroLex: A Comprehensive Neuroscience Ontology Development Based on Multiple Biomedical Ontologies and Community Involvement 349
Fahim T. Imam, Stephen D. Larson, Jeffrey S. Grethe, Amarnath Gupta, Anita Bandrowski, Maryann E. Martone

Use of Multiple Ontologies to Characterize the Bioactivity of Small Molecules 357
Ying Yan, Janna Hastings, Jee-Hyub Kim, Stefan Schulz, Christoph Steinbeck, Dietrich Rebholz-Schuhmann

A Meta-Data Approach to Querying Multiple Biomedical Ontologies 364
Ravi Palla, Dan Tecuci, Vinay Shet, Mathaeus Dejori

Multiple Ontologies in Healthcare Information Technology: Motivations and Recommendation for Ontology Mapping and Alignment 367
Colin Puri, Karthik Gomadam, Prateek Jain, Peter Z. Yeh, Kunal Verma

Modularization for the Cell Ontology 370
Christopher J. Mungall, Melissa Haendel, Amelia Ireland, Shahid Manzoor, Terry Meehan, David Osumi-Sutherland, Carlo Torniai, Alexander D. Diehl

Building the OBO Foundry – One Policy at a Time 377
Mélanie Courtot, Chris Mungall, Ryan R. Brinkman, Alan Ruttenberg

Towards a Body Fluids Ontology: A Unified Application Ontology for Basic and Translational Science 381
Jiye Ai, Mauricio Barcellos Almeida, André Queiroz de Andrade, Alan Ruttenberg, David Tai Wai Wong, Barry Smith

Connecting Ontologies for the Representation of Biological Pathways 387
Anna Maria Masci, Mikhail Levin, Alan Ruttenberg, Lindsay G. Cowell

Bridging Multiple Ontologies: Representation of the Liver Immune Response 393
Anna Maria Masci, Jeffrey Roach, Bernard de Bono, Pierre Grenon, Lindsay Cowell

Hyperontology for the Biomedical Ontologist: A Sketch and Some Examples 399
Oliver Kutz, Till Mosskowski, Janna Hastings, Alexander Garcia Castro, Aleksandra Sojic

Wednesday, July 27, 2011

Facilitating Anatomy Ontology Interoperability 409

Mouse Anatomy Ontologies and GXD 411
Terry F. Hayamizu, Martin Ringwald

FUNCARO: A Functional Extension to CARO 412
David Osumi-Sutherland

The Vertebrate Bridging Ontology (VBO) 416
Ravensara Travillian, James Malone, Chao Pang, John Hancock, Peter W.H. Holland, Paul Schofield, Helen Parkinson

Multi-Species Anatomy Ontology Development Requires a Pluralist Approach to Label-Class Mapping 422
István Mikó, Matthew J. Yoder, Matthew A. Bertone, Katja C. Seltmann, Andrew R. Deans

CARO 2.0 425
David Osumi-Sutherland

Integrating Anatomy and Phenotype Ontologies with Taxonomic Hierarchies 426
James P. Balhoff, Peter E. Midford, Hilmar Lapp

Phenoscape: Use Cases and Anatomy Ontology Requirements for Linking Evolutionary and Model Organism Phenotypes 428
Wasila Dahdul, James Balhoff, Hilmar Lapp, Peter Midford, Todd Vision, Monte Westerfield, Paula Mabee

Ontologies in the Fish Tank: Using the Zebrafish Anatomy Ontology
with Other OBO Ontologies to Annotate Expression and Phenotype 430
Yvonne Bradford, Ceri Van Slyke

Doctoral and Post-Doctoral Consortium 431

One Empirical Study, Three Tests of Translation, Many Questions on Biomedical Ontologies:
Limited Contribution of MeSH Terms to Effective Literature Searches on 'Health-Related Values' 433
Mila Petrova

The Age of Data-Driven Medicine: Mining the Electronic Health Record 435
Paea LePendu, Mark A. Musen, Nigam H. Shah

Quality of Care Domain Modeling in Cancer: A Semantic Approach 436
Sina Madani, Dean F. Sittig, Parsa Mirhaji, Kim Dunn

Philosophy, Ontology, and Scientific Explanation 438
James A. Overton

Refining Ontology for Glucose Metabolism Disorders 440
Yu Lin

A Case Study in Using ZFA and PATO for Describing Histological Phenotypes in the Larval Zebrafish 441
Brian Canada, Georgia Thomas, Timothy Cooper, Keith Cheng

Epistemological Issues in Information Organization Instruments: Ontologies and Health Information Models 442
André Queiroz Andrade, Maurício Barcellos Almeida

The Foundational Model of Neuroanatomy Ontology:
An Ontology Framework to Support Neuroanatomical Data Integration 444
B. Nolan Nichols, Jose L.V. Mejino Jr., James F. Brinkley

Workflows and Framework for Nutritional/Metabolic Phenotype/Genotype
and Foods-for-Health Knowledge Integration 446
Matthew Lange, J. Bruce German, Jim Kaput

Applications for a Translational Biomedical Ontology Model 447
Robert Yao, Graciela Gonzalez

Author Index 449