



**Disease ontologies and information:  
its linkage to genes, gene ontologies and phenotypic information.**



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EMBL-EBI



EBI is an Outstation of the European Molecular Biology Laboratory.

## Areas of Interest



- Where we can find good information about diseases, especially diseases with known or suspected linkage to genes, and integrate this with information available in biomedical informatics data resources?
- How can we best link this information to disease models in animal and in particular mouse phenotypes, where such models are used as a proxy for disease information in humans?
- What approaches are currently being made in the community to address disease ontologies orchestration?
- What is the linkage between bioinformatics resources and disease/medical information?
- Which diseases share common symptoms or are otherwise related?
- How can animal model phenotypes be related to human disease information to make them more useful and amenable to automatic interrogation?
- Most generally, how do we overcome the historical poor linkage between bioinformatics data and disease and medical information?



## Agenda



10:15	Wellcome and Background to Meeting (Prof. Janet Thornton, & Dr. Dominic Clark, EMBL-EBI)
10.30	“Ontologies for Biologists – The Promise and the Reality” (Prof. Michael Ashburner, U. Cambridge).
10.50	The OBO (Open Biomedical Ontologies) Foundry and PATO: Phenotype And Trait Ontology (Prof. Michael Ashburner, U. Cambridge.)
11.10	“Towards an Open Source Disease Ontology” Prof. Warren Kibbe (Northwestern University)
11.50	Tea/Coffee
12.20	<a href="#">Mouse Genome Informatics Database</a> : Phenotypes and Phenotype Ontology (Prof. Janan Eppig, Jackson Lab)
12.50	Discussion

## Agenda 19<sup>th</sup> June pm



14.20	Presentations by Industry Partners, Dr. Bertram Weiss, Bayer Schering Pharma, Dr Ian Harrow Pfizer, Dr. Andrew Garrow, Unilever
15.00	Prof. Giorgio Valle (University of Padua): Gene Ontology of CNS and Disease Ontology.
15.20	The GEN2PHEN Project – Harnessing the Web for Genomics (Prof Anthony Brookes, U. Leicester).
15:40	Discussion
16:00	Tea/Coffee
16:20	Healthgrid, the EU SHARE project and the EMBRACE project (Dr. Vincent Breton, CNRS, Clermont-Ferrand)
16:40	Disease Ontologies and their Relation to Genes (Dr Anita Burgun, University of Rennes)
17:00	Developing an Application Ontology for Annotation of Experimental variables across HTP data sets (Helen Parkinson and James Malone, EMBL-EBI)
17:20	Summary and discussion
17.45	Bus transfer to Alimentum Restaurant, Hills Road, Cambridge



## Agenda 20<sup>th</sup> June am



09.00	Transport from Homerton College to WT Genome Campus
09.30	Tea/Coffee
09.45	The Influenza Infectious Disease Ontology (I-IDO) (Dr Joanne Luciano, Mitre Corporation)
10.05	UniMed: Mapping Protein Information to Disease Terminologies. (Dr. Anne-Lise Veuthey, Swiss Institute of Bioinformatics)
10.25	Text Mining and Ontologies: an analysis of all gene-disease associations over Medline based on GO terms (Dr. Dietrich Rebholz-Schuhmann, EMBL-EBI)
10.45	Observations on presentations and themes by invited discussants.
11.20	Tea/Coffee
11.45	Discussion led by industry partners and documentation of Actions/Next Steps (Dr. Dominic Clark)



## Agenda 20<sup>th</sup> June pm



13:45	Introduction to ELIXIR Project WP7 (Dr. Andrew Lyall & Prof. Amos Bairoch)
14.10	Facilitated Discussion (Dr. Dominic Clark) with observations on presentations and themes by invited discussants.
15.30	Documentation of input from workshop into ELIXIR WP7 Roadmap document (Prof. Amos Bairoch & Prof. Michael Ashburner)
15:45	Tea/Coffee and Departures



## Acknowledgements



- The Scientific Coordinator for this workshop is Prof. Michael Ashburner (University of Cambridge and former joint head of EMBL-EBI).
- The programme advisors are Dr. Ian Harrow (Pfizer), Ms. Wendy Filsell (Unilever), Dr. Matt Hall (GSK) and Dr. Bertram Weiss (Bayer Schering Pharma).
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