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## Appendix: Content Summaries of Selected Best Papers for the 2017 IMIA Yearbook, Section Knowledge Representation and Management

**Banda JM, Evans L, Vanguri RS, Tatonetti NP, Ryan PB, Shah NH**

**A curated and standardized adverse drug event resource to accelerate drug safety research**

*Sci Data* 2016;3:160026

This open science paper introduces a large, curated, and publicly available resource for adverse drug event (ADE) research: it includes an ADE dataset with the source code and documentation to be used by the research community. This resource (AEO-LUS) derives from the publicly available US Food and Drug Administration (FDA) Adverse Event Reporting System (FAERS) dataset, covering spontaneous reports of adverse drug events since 2012. The resource also integrates a legacy dataset covering 2004-2012. The final dataset results from a pipeline consolidating all relevant data for ADE research, normalizing different term usage, de-duplicating cases, mapping drugs to RxNorm, mapping drug indications

and reactions to MedDRA, and generating drug-outcome pairs with associated statistics. The provided documentation and code is expected to support the evolution of this resource when updates of the FAERS dataset are released.

**Bauer CR, Ganslandt T, Baum B, Christoph J, Engel I, Lobe M, Mate S, Staubert S, Drepper J, Prokosch HU, Winter A, Sax U**  
**Integrated Data Repository Toolkit (IDRT). A Suite of Programs to Facilitate Health Analytics on Heterogeneous Medical Data**  
*Methods Inf Med* 2016;55(2):125-35

This paper presents a set of tools developed within the Integrated Data Repository Toolkit (IDRT) German project in order to facilitate heterogeneous data integration in the i2b2 framework. Among various applications, this toolset efficiently allows researchers to design their own analyses. For example, the Mapping Editor of the IRDT Import and Mapping Tool helps to import different formats of data into the current i2b2 terminology. The originality at this step is to allow users to integrate data described in different terminologies (e.g., ICD-10-GM, MedDRA, LOINC, ICP-O, and others) and to build a new repository, with a unique model able to support the representation of these data. The new target ontology is a dedicated model of the data of the project which can be saved for new analyses – with a prior translation of the data from the original terminological model to a new target hierarchy. As a result, IDRT appears as a step forward to the semantization of i2b2.

**Greene D, NIHR BioResource, Richardson S, Turro E**

**Phenotype Similarity Regression for Identifying the Genetic Determinants of Rare Diseases**

*Am J Hum Genet* 2016;98(3):490-9

This paper is dedicated to model the association between multiple phenotypic traits described with the Human Phenotype Ontology (HPO) and the corresponding genotype, in the specific context of rare disease (rare variants). HPO allows composite phenotypes to be represented systematically

but association methods accounting for the ontological relationship between HPO terms do not exist. The authors propose a Bayesian method to model the association between HPO-coded phenotypes and genotypes. The method uncovers associations between rare genotypes and the similarities between patients' phenotypes and a latent characteristic phenotype. The effectiveness of the approach is demonstrated on a simulation study and on a real dataset from the BRIDGE project.

**Sarntivijai S, Vasant D, Jupp S, Saunders G, Bento AP, Gonzalez D, Betts J, Hasan S, Koscielny G, Dunham I, Parkinson H, Malone J**

**Linking rare and common disease: mapping clinical disease-phenotypes to ontologies in therapeutic target validation**

*J Biomed Semantics* 2016;7-8

This paper proposes solutions to annotation-ontology mapping in genome-scale data. Of particular interest in this work is the Experimental Factor Ontology (EFO) and its generic association model, the Ontology of Biomedical Association (OBAN). EFO is a well-founded ontology, reusing ontologies from the Open Biomedical Ontologies (OBO) community (and other necessary models) for a comprehensive description of the domain, with the Minimum Information to Reference an External Ontology Term (MIREOT) strategy: Chemical Entities of Biological Interest Ontology (ChEBI), the Phenotypic And Trait Ontology (PATO), the Orphanet Rare Disease Ontology (ORDO), the BRENDA Tissue Ontology (BTO), the Uber Anatomy Ontology (Uberon), and the Gene Ontology (GO). OBAN is a means to represent diseases and phenotypes associations and the source of evidence for these associations. This was applied to the use case of linking rare to common diseases at the Centre for Therapeutic Target Validation. Based on these models, this work demonstrates the feasibility of rare and common diseases integration, using shared phenotypes. This paper offers a convincing example of the industrialization of integration. The EFO ontology is updated monthly and allows to propose regularly new associations.