

RESEARCH METHODS TRIAL

Paper 2: June 2017

Methods Trial Papers

Yael Garten



Methods Trial Papers:

, *Papers on Sociological Methods* Francis Stuart Chapin, 1924 **MEDINFO 2007** K.A. Kuhn, J.R. Warren, T.-Y. Leong, 2007-08-02 Contains papers which reflect the breadth and depth of the field of biomedical and health informatics covering topics such as health information systems education standards consumer health and human factors emerging technologies sustainability organizational and economic issues genomics and image and signal processing **Text Mining of the Scientific Literature to Identify Pharmacogenomic Interactions** Yael Garten, 2010 Pharmacogenomics is the study of how variation in the human genome impacts drug response in patients It is a major driving force of personalized medicine in which drug choice and dosing decisions are informed by individual information such as DNA genotype The field of pharmacogenomics is in an era of explosive growth massive amounts of data are being collected and knowledge discovered which promises to push forward the reality of individualized clinical care However this large amount of data is dispersed in many journals in the scientific literature and pharmacogenomic findings are discussed in a variety of non standardized ways It is thus challenging to identify important associations between drugs and molecular entities particularly genes and gene variants Thus these critical connections are not easily available to investigators or clinicians who wish to survey the state of knowledge for any particular gene drug disease or variant Manual efforts have attempted to catalog this information however the rapid expansion of pharmacogenomic literature has made this approach infeasible Natural Language Processing and text mining techniques allow us to convert free style text to a computable searchable format in which pharmacogenomic concepts such as genes drugs polymorphisms and diseases are identified and important links between these concepts are recorded My dissertation describes novel computational methods to extract and predict pharmacogenomic relationships from text In one project we extract pharmacogenomic relationships from the primary literature using text mining We process information at the fine grained sentence level using full text when available In a second project we investigate the use of these extracted relationships in place of manually curated relationships as input into an algorithm that predicts pharmacogenes for a drug of interest We show that for this application we can perform as well with text mined relationships as with manually curated information This approach holds great promise as it is cheaper faster and more scalable than manual curation Our method provides us with interesting drug gene relationship predictions that warrant further experimental investigation In the third project we describe knowledge inference in the context of pharmacogenomic relationships Using cutting edge natural language processing tools and automated reasoning we create a rich semantic network of 40 000 pharmacogenomic relationships distilled from 17 million Medline abstracts This network connects over 200 entity types with clear semantics using more than 70 unique types of relationships We use this network to create collections of precise and specific types of knowledge and infer relationships not stated explicitly in the text but rather inferred from the large number of related sentences found in the literature This is exciting because it demonstrates that we are able to overcome the heterogeneity of

written language and infer the correct semantics of the relationship described by authors Finally we can use this network to identify conflicting facts described in the literature to study change in language use over time and to predict drug drug interactions These achievements provide us with new ways of interacting with the literature and the knowledge embedded within it and help ensure that we do not bury the knowledge embodied in the publications but rather connect the often fragmented and disconnected pieces of knowledge spread across millions of articles in hundreds of journals We are thereby brought one step closer to the realization of personalized medicine and ensure that as scientists we continue to build on the knowledge discovered by past generations and truly to stand on the shoulders of giants

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