Rethinking drug treatment rationale with biomedical mechanism-based phenotype descriptions

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ABSTRACT:
Developing analysis pipelines that integrate complex biomedical mechanisms with quantitative population data to inform disease stratification and treatment rationale, provides a timely research area with unexplored, well-motivated research directions in translational bioinformatics. With the growth of semantic Web technologies that enable people to create data stores on the Web, build vocabularies, and write rules for handling data, it is only recently that it has become feasible to gather large data sets needed for the research in this area. By considering clinical signatures and possible disease and drug mechanisms, we are establishing mechanism-based descriptions of patient phenotypes and exploring potential for those descriptions to inform treatment choice and patient outcomes. Two examples from our work will be presented: one project that combines existing genetic and pharmacologic knowledge with patient phenotype data to make personalized drug exposure predictions in breast cancer patients; and another project that uses an ontological approach to mine clinical signature in order to assist the diagnosis of drug-induced liver injury.