

Call for Papers: Deep Phenotyping for Precision Medicine

Due date for submission: March 1, 2019

We invite submissions for a special issue of the *Journal of Biomedical Informatics* focused on Deep Phenotyping to enable Precision Medicine. This special issue aims to provide a collection of emerging theories, cutting-edge methodologies, and novel technologies that enable scalable human phenotype discovery and application in clinical data for continuous health learning.

An important goal of Precision Medicine is to develop a framework for creating a new taxonomy of human diseases based on molecular biology and then to create it [1]. Towards this goal, clinical data have been recognized as the basic staple of health learning [2]. The rapidly growing interoperable clinical datasets, including electronic health records (EHR), patient-generated self-tracking data, administrative and claims records, and clinical research results data, have presented unprecedented opportunities for developing high-throughput methods for deep phenotyping.

In the context of this special issue, phenotype is “*the trait or observable characteristic of a human being representing his or her unique morphological, biochemical, physiological, or behavioral property*” [3]. Related, phenotyping refers to the process of characterization or classification of a patient’s phenotype. Deep phenotyping further emphasizes the precision and comprehensiveness of the characterized phenotype [4]. Fundamental to studying disease similarities to assist in the development of a precise disease taxonomy, deep phenotyping can shed light on gene functions and enable precise diagnoses, subtyping, and treatments. Software or algorithms leveraging deep phenotyping for gene prioritization have evolved to the point of demonstrating their usefulness in genomic diagnostic decision support [5-13].

Possible topics include, but are not limited to:

- Computational phenotype analysis (e.g., causal [14] or probabilistic phenotyping [15])
- Temporal phenotyping

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