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A pipeline for the design and analysis of molecular diagnostics

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Biomarker panels are becoming increasingly important clinical tools. In the research lab they also have a wide array of applications, including the identification and characterization of unknown and experimentally manipulated samples. In order to keep cost down and facilitate high sample throughput, biomarker panels are often limited to a relatively small number of markers. Typically these are handpicked genes deemed important or informative by the researcher. However, without statistical support that the most informative biomarkers have been selected, biomarker panels can be subject to extensive sampling bias, resulting in wasted resources and misclassification. Moreover, even in the presence of proper biomarker selection, the accurate interpretation of marker profiles is often difficult and frequently undertaken without statistical rigor. Here we present a pipeline for the rational, easy, and statistically-robust design and interpretation of biomarker panels. The first half of the pipeline consists of an algorithm designed to identify the minimum unique biomarker profile (MUMP) necessary to accurately and efficiently classify sample types within a set of given experimental constraints. The second part of the pipeline provides statistically robust matching of biomarker profiles to reference samples, providing a statistically defensible interpretation of the biomarker assay. Single-blind testing on samples of differentiating stem cells shows that the MUMPs pipeline can accurately identify cells in the lab. The MUMPs pipeline is reliable, easy, and cost-effective tool that will allow bench scientists to readily implement computationally supported rational design of biomarker assays.

Biography

James Lindsay is a Ph.D. student at the University of Connecticut in the Computer Science and Engineering department. James is also the CTO of Smpl Bio, a bioinformatics company commercializing software to simplify the design and analysis of genetic tests.

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