



CPH Seminar in Precision Medicine

Using Artificial Intelligence to Plan and Conduct Bioinformatics Analyses

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Due to advancing technologies and plummeting costs, the use of genomic assays has rapidly become commonplace in biology research. Unfortunately, the collection of this data far exceeds the ability to analyze it, a situation that is acutely felt and widely documented. From our prior work, we found that the reason that it has been so difficult meet the need for bioinformatics is due to the expert knowledge required. Analyzing a data set is rarely a straightforward task that can be automated, but instead, must be specialized to address the biological question being asked. Thus, it is unlikely that the current approach of automating repetitive tasks using pipelines will significantly reduce the burden. There are no common projects in biological discovery.

To address the need for bioinformatics, we have developed an artificial intelligence that can perform bioinformatics analyses. The Bioinformatics Expert SYstem (BETSY) includes a knowledge base that explicitly and formally encodes the abilities of bioinformatics software to process biological data. BETSY also includes a backwards-chaining rule-based expert system comprised of a data model that can capture the richness of biological data, and an inference engine that reasons on the knowledge base to produce workflows. Currently, the knowledge base is populated with rules to analyze microarray and next generation sequencing data. We evaluated BETSY and found that it could generate workflows that reproduce and go beyond previously published bioinformatics results. In sum, we found that an expert system approach can facilitate exploratory bioinformatics analysis by automating the development of workflows, a task that formerly required significant human domain expertise.

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