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Implementation of a hybrid informatics solution for a large personalized medicine program

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The bioinformatics needs a large personalized or precision medicine program which exemplify the 3Vs of big data challenges: volume, variety and velocity. In the center of personalized medicine at Children's Hospital Los Angeles, we offer a variety of genomics-based somatic and germline genetic tests (single-gene, gene-panel, whole-exome, whole-genome, chromosomal microarray) for a large number of pediatric patients with either cancers or constitutional disorders. To address the needs and challenges, we implemented a hybrid informatics solution in terms of both the IT infrastructure and the bioinformatics tools. First of all, we employed an on-premise High-Performance Computing Cluster (HPCC) to address the mission-critical computing needs, established the parallel IT infrastucture in AWS cloud for redundancy, bursting nature of our computing needs and cost-effectiveness. Similarly, while implementing a phenotype-driven analytic platform for these genomics-based tests, we used a combination of open-source and custom software tools, striving to provide the most cutting-edge bioinformatics solution at an academic medical center. Examples of our custom solution includes 1) a cloud- and web-based variant store using the OpenCGA package that we developed in collaboration with the Genomics England Project team for storing and analyzing somatic and germline variants from whole-exome sequencing and whole-genome sequencing of thoussands of patients, 2) an algorithm for computational phasing two candidate heterozygous variants in a single gene and for improved clinical diagnosis rate of recessive disorders therefore, and 3) an NGS coverage analysis algorithm that is 10 times more efficient than the most popular similar tools, namely SAMtools and Sambamba.

Biography

Xiaowu Gai serves as the Director of Bioinformatics in the Center for Personalized Medicine at Children's Hospital Los Angeles (CHLA). He is an Associate Professor of Clinical Pathology at the Keck School of Medicine, University of Southern California. Before joining CHLA, he was the Director of Bioinformatics and Associate Director of the Ocular Genomics Institute in the Department of Ophthalmology at Harvard Medical School & Massachusetts Eye and Ear Infirmary, Boston. Prior to that, he served as the Director of Biomedical Informatics at Loyola University Chicago Stritch School of Medicine.

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