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Our exome experience: Informatics, technology and discoveries

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Whole exome sequencing (WES) has emerged as a powerful technique to identify disease genes for Mendelian disorders. Both informatics and technical challenges exist and have limited the WES success rate. We have established a highly modular and primarily database driven exome variant analysis pipeline to address the informatics challenges by incorporating our ever-increasing knowledge of human genetic variations in both the general populations and the specific disease cohorts. Using this pipeline, we have analyzed over 400 exomes from multiple cohorts of rare genetic disorders, including especially inherited retinal degenerations and mitochondrial diseases. At the same time, we have developed and validated a custom exome design to address some coverage deficiencies with current commercial exome capture kits. Together, these informatics and technical advancements have allowed us to identify a number of novel disease genes for each of the disorders studied.

Biography

Xiaowu Gai is a human geneticist and a bioinformatician. He received his Ph.D. degree in genetics from Iowa State University in 1999. Currently, he is an Assistant Professor in the Department of Molecular Pharmacology and Therapeutics at the Loyola University Stritch School of Medicine. He is also the Director of Biomedical Informatics for the Stritch School of Medicine. He has published 26 papers and book chapters and developed a number of bioinformatics software packages for genomic data analysis. He has long-term research interest in understanding human genetic variations and the development of individualized medical treatments.

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