BIO

Shu-Jui Hsu, Ph.D.

As a bioinformatician and human geneticist, I am interested in utilizing computational approaches to tackle unresolved questions in biological mechanisms, especially in the field of human genomics. In this overall context, the application of next-generation sequencing (NGS) technology for the delineation of disease etiology is the major focus of my research. As a huge amount of data is generated from NGS platforms, data analysis is a crucial step in precision medicine. To this end, we have been involved in the development of bioinformatic algorithms for the efficient integration of NGS data with the phenotypic characterization of clinical patients. We collaborate extensively with clinicians on implementing these algorithms for the analysis of whole genome sequence (WGS) and whole exome sequence (WES) data obtained from clinical patients suffering from diseases, such as neurodegenerative disorders (Parkinsonism, ALS, Dementia), cancer (Lung cancer, Leukemia), and Mendelian disorders (Deafness, Jaundice, and other Developmental Disorders).

In general, there are three specific research interests in this interdisciplinary laboratory:

- Clinical NGS study design, data analysis, and molecular diagnostics
- Development of bioinformatic algorithms for pathogenicity prioritization in human WGS/WES data based on genetic variations
- Validation of genetic/bioinformatic algorithms by real-world NGS data

Currently, we are using internal cohort data and Taiwan Biobank data to evaluate major DNA-seq analysis pipelines. By quantifying the robustness of each bioinformatic pipeline, we can standardize our analysis specifically for the Taiwanese population. In addition, we are examining the genetic profiles of Taiwanese individuals for the genes listed in the American College of Medical Genetics (ACMG) guidelines, which are considered clinically actionable. On the other hand, advances in the molecular diagnostics pipeline are envisioned to help us improve both bioinformatic algorithms in clinical settings and genetic diagnostics in the hospital.