Summary:
High-throughput sequencing (HTS) now means that scientists and clinicians are now regularly faced with an often bewildering set of genetic variants that might relate to a disease or phenomenon of interest. We have developed numerous tools over the past decade for interrogating genetic variants for their possible impact on protein function that can help to rank/prioritize variants for further study or clinical applications. In this presentation, I will describe some of these tools, and discuss our recent applications to understanding biological mechanism and disease. These will include an analysis of cancer variants impacting GPCR/G-protein coupling with implications for off-label drug repurposing, and a summary of how our tools and expertise are increasingly used to aid clinical diagnosis and treatment decisions in genetic diseases and cancer.

Example references:

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