CURRENT PROGRESS IN IDENTIFYING DISEASE GENES: INSIGHTS FROM MULTIPLE SCLEROSIS

Identifying disease genes is a crucial step towards understanding the underlying mechanisms of disorders, including Multiple Sclerosis (MS). This talk will provide an overview of the current status and advancements in the search for disease genes, focusing on the context of MS. Multiple sclerosis (MS) is a chronic, neuroinflammatory, neurodegenerative disease of the central nervous system with both genetic and environmental risk factors.
Over the past decades, significant progress has been made in uncovering the genetic basis of MS, aided by advancements in high-throughput sequencing technologies and large-scale collaborative efforts including Genome-wide association studies (GWAS) initiated by International Multiple Sclerosis Genetics Consortium (IMSGC). These variants are in genes involved in immune response, inflammation, and myelin formation, shedding light on the key biological processes underlying MS pathogenesis. Furthermore, rare variants with large effect sizes have been discovered through exome and whole-genome sequencing, providing insights into rare and familial forms of MS. However, the genetic architecture of complex diseases like MS is not solely determined by individual variants. Polygenic score analysis has emerged as a powerful tool to assess the cumulative effect of multiple genetic variants across the genome. By calculating a polygenic score based on the collective contribution of many variants, we can estimate an individual's genetic risk for a particular disease. These scores can be utilized for risk stratification, facilitating early interventions and personalized treatment approaches.