Abstract Title: Unravelling the genetics of young onset Parkinson disease

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Abstract: Parkinson's disease (PD) stands out as a genetically diverse neurodegenerative disorder influenced by poorly understood environmental factors. Recent genomic analyses of PD patients have unveiled pivotal monogenic genes, noteworthy, rare variants, and polygenic risk-associated variations. Our multi-centric study on young-onset Parkinson's disease (YOPD) encompassed a cohort of 674 patients and 1363 control samples. The investigation involved diagnostic testing to identify causal mutations, exploring genetic associations, and validating polygenic risk scores. This research represents the most extensive genetic exploration of PD, marking the initial revelation of the SNCA association with YOPD in the Indian population. We combined rare and common variants analysis together, to showcase a comprehensive genetic landscape of YOPD in Indian. To the best of our knowledge, this is the largest study in YOPD in India.

Area of expertise:

• Human Genetics, Diagnostics, Next Generation Sequencing (NGS)

• Operationalized South Asia’s largest genomics lab and business

• Built & launched diverse genetic testing portfolios at affordable prices (NIPT, Carrier screening, liquid biopsy, etc.)