MedicalXpress

23andMe identifies two novel genetic associations and substantial genetic component for Parkinson's

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Today 23andMe, an industry leader in personal genetics, announced the discovery of two significant, novel genetic associations with Parkinson's disease (PD) and provided new evidence that there is a substantial genetic component remaining to be discovered for Parkinson's. "Web-based genome-wide association study identifies two novel loci and a substantial genetic component for Parkinson's disease" was published online today in *PLoS Genetics*.

The 23andMe study discovered two novel associations with Parkinson's disease. The first lies near the gene SCARB2, which is involved with known Parkinson's disease pathways. The second lies near the genes SREBF1 and RAI1 and is of unknown function. The 23andMe research team also replicated twenty previously discovered genetic associations, providing support for the novel web-based design used in this study.

While the evidence of a genetic basis for early-onset Parkinson's is well-known, the importance of genetics for the late-onset form of the disease has been less clear to date. The 23andMe study, however, provides strong evidence that at least one-quarter of the variation in susceptibility to late-onset Parkinson's disease is due to genetic factors. This finding shows that a considerable proportion of the genetics of Parkinson's has yet to be explained while also confirming the importance of environmental factors.

According to lead author Chuong B. Do. "Not only are these new genetic findings significant, but we've also shown that the data collected by 23andMe support discovery of new associations as well as replication of previously known associations. This study is a rigorous "proof of principle" and clearly demonstrates that web-based phenotyping works for a disease of real public health significance."

23andMe conducted its research using a study cohort of 3,426 PD cases assembled in collaboration with The Parkinson's Institute and Clinical Center ("PI") and The Michael J. Fox Foundation. Patients were recruited through a targeted email campaign by The Parkinson's Institute and The Michael J. Fox Foundation, as well as other Parkinson's patient groups and clinics. Some patients were also recruited in person at workshops and conferences. The cohort was assembled over an 18-month period with more than half of the participants registering in the first month.

This research utilized 23andMe's web-based platform in a novel approach to study complex diseases. Using the Internet to query and interact with people in the research cohort significantly increases efficiency and reduces the cost of recruiting participants and conducting research. The broad reach of the web allows individuals who are not near research centers to contribute and participate in the program. Utilizing 23andMe customers, who are not Parkinson's patients as study controls also lowers the overall cost.

"We believe this is just the beginning of the tremendous research potential that will evolve from the work being done at 23andMe," said J.William Langston, M.D., Parkinson's Institute CEO, Scientific Director, and Founder.

Todd Sherer, Ph.D, CEO of The Michael J. Fox Foundation said, "23andMe's Parkinson's initiative has proven the tremendous potential in

leveraging DNA technology, the Internet, and patient participation to accelerate discoveries that enhance our understanding of Parkinson's disease. As more individuals with Parkinson's continue to join 23andMe's community, patient-driven research will accelerate and support development of breakthrough therapeutics."

The 29,624 study controls consisted of users of 23andMe's Personal Genome Service® who consented to participate in IRB-approved studies and answered online questionnaires. Since completing this specific research effort, the 23andMe PD research cohort has grown to more than 5,000 enrolled PD patients and the 23andMe database contains more than 100,000 users of whom more than 76,000 have consented to participate in ongoing research efforts.

"We believe this paper proves the potential of our approach of combining genetic information with web-based data about specific conditions to make novel research discoveries," said 23andMe President and CEO Anne Wojcicki. "It's extremely rewarding that 23andMe has made a significant contribution to Parkinson's research through our novel web-based approach to research. This approach has the potential to be used in many other conditions."

In addition to the PD community, the 23andMe sarcoma community of more than 500 individuals diagnosed with sarcoma is one of the world's largest sarcoma research cohorts and studies are underway. Additional disease-specific communities and research efforts are planned.

Provided by 23andMe Inc.

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