Parkinson’s Disease Research Initiative Case Study

A collaborative initiative by The Parkinson’s Institute and Clinical Center, The Michael J. Fox Foundation for Parkinson’s Research and 23andMe, Inc.

The Challenge:
To significantly accelerate the pace of Parkinson’s research, at a lower cost

The Solution:
The 23andMe Parkinson’s Disease Research Initiative

The Results:
- One of the world’s largest databases of Parkinson’s patients, in less than a year
- 4,250 participants enrolled in first year (2,000 in first month)
- Over 30,000 questionnaires completed by Parkinson’s patients
- Over 80% of the participants are engaged in our online research
- Significantly lower cost than traditional studies
- 8,000 person control group with minimal effort and cost
- One of the world’s largest cohorts of early on-set Parkinson’s patients
- One of the world’s largest databases of people with rare G2019S mutation in LRRK2 gene
- Replicated several well-established Parkinson’s gene associations
- Identified five novel Parkinson’s gene associations in the first year
"We still do clinical research the way we’ve been doing it for decades. Using the Internet to connect with more people at a lower cost—that just makes sense to us."

**Katie Hood**  
Chief Executive Officer  
The Michael J. Fox Foundation for Parkinson’s Research

"In less than a year, 23andMe has identified 600 patients with early-onset Parkinson’s, one of the largest cohorts in the world."

**Dr. J. William Langston**  
Chief Executive Officer and Scientific Director  
The Parkinson’s Institute and Clinical Center

"It could take years to get 2,000 participants using traditional methods. 23andMe enrolled that many during the first month."

**Dr. Todd Sherer**  
Vice President of Research Programs  
The Michael J. Fox Foundation for Parkinson’s Research

Internet-Based Research: Faster, Cheaper—and Built for Engagement

Since inception, 23andMe had been discussing a vision for a new kind of online research that would give individuals the opportunity to actively participate in research that was meaningful to them. In January 2009, 23andMe and The Michael J. Fox Foundation for Parkinson’s Research (MJFF) decided to form an online Parkinson’s community that would make participating in research its primary cause. The decision came after months of discussion with partners, researchers and patients.

Katie Hood, CEO of MJFF, constantly takes the pulse of the patient community and shares their impatience: “Traditionally, patients have been on the sidelines cheering for the scientists and clinicians to deliver new treatments. Now they want to be involved.”

Having funded other genome-wide association studies (GWAS), MJFF understood the potential for genetics in Parkinson’s research. Hood was also intrigued by the opportunity to accelerate the process: “We still do clinical research the way we’ve been doing it for decades. Using the Internet to connect with more people at a lower cost—that just makes sense to us.” Hood enthusiastically agreed to the 23andMe online community proposal.

**The Numbers Game**

To do meaningful research, you need a significant sample size. It takes lots of study participants to do good science. Each must be qualified, enrolled, surveyed, and tracked. You have to keep them engaged for the length of the study, often many years.

Dr. Todd Sherer, Vice President of Research Programs at MJFF, emphasizes the need for large-scale studies: “Parkinson’s has a high rate of variability in its symptoms, progression, and response to therapeutic interventions. Often, we just don’t have enough data to answer the tough questions. Sample size is very important.”
For people diagnosed with Parkinson’s Disease, it’s natural to hope for a cure. For isn’t enough—you want to do something.

A year later, 23andMe has recruited over 4,250 Parkinson’s patients from 49 US states and 17 countries—and the number grows every week. These patients have submitted saliva samples for analysis and completed more than 30,000 online questionnaires describing their health, activities, and environment. In addition, 8,000 people without Parkinson’s have also completed the same questionnaires, forming a very large control group.

Of particular interest to scientists are subgroups that represent especially promising avenues for research. One subgroup in the 23andMe database consists of people diagnosed with early-onset Parkinson’s (diagnosed before age 50). This is an extremely rare condition that could shed light on the triggers for the disease. “In less than a year, 23andMe has identified 600 patients with early-onset Parkinson’s, one of the largest cohorts in the world,” Dr. Langston reports.

Another key subgroup involves people with the G2019S mutation in the LRRK2 gene. Researchers have found that this mutation dramatically increases the likelihood of developing Parkinson’s. One recent study puts the chances as high as 59% or higher over a person’s lifetime. Many researchers believe this particular mutation will yield breakthrough discoveries. With one of the world’s largest databases of patients with this LRRK2 mutation, 23andMe is uniquely positioned to support research into this key mutation.

### Strong Correlation with Existing Research

An important avenue of investigation is to determine how well online research stacks up against traditional methods. In a presentation to the National Institutes of Health (NIH) in December 2009, Nicholas Eriksson, Principal Scientist at 23andMe, reported that 23andMe had replicated well-established associations between Parkinson’s disease and specific genetic mutations (GBA, LRRK2, SNCA, MAPT), with good correlation of the magnitude of the effects. For example, 23andMe showed that a particular variation in the MAPT gene reduces the odds of Parkinson’s by 21%, very much in line with the published figure of 23%.
The data collected through online surveys is being validated in other ways. 23andMe has been able to segregate participants into the classically observed subtypes of Parkinson’s disease (tremor-dominant and postural instability gait difficulty) solely on the basis of online survey responses.

With funding from MJFF, 23andMe and The Parkinson’s Institute are working on a long-term project to formally compare online survey data to information collected in a clinical setting, and quantify the degree of correlation.

### Moving the Science Forward

Published genome-wide association studies (GWAS) of European populations have reported three associations of specific genes to Parkinson’s disease—work that 23andMe has replicated. But the company is also making its own breakthrough discoveries. Scientists at 23andMe have identified five new significant gene associations and are now seeking to replicate these findings in independent populations.1

### Why Should Researchers Partner with 23andMe?

Asked about the benefits of doing research with 23andMe, Dr. Langston highlights the company’s willingness to collaborate: “They are eager to get the input of researchers. You can proactively develop research protocols with 23andMe.”

He goes on to point out 23andMe’s ability to deliver a large number of study participants: “When I started in neurology, studies of 10 people were not uncommon. Now we can work with thousands, even tens of thousands, using 23andMe’s expertise in online research and patient recruitment. We can address research questions that were impenetrable with smaller sample sizes.”

### Why Should Foundations Partner with 23andMe?

“Why should foundations partner with us?” asks Hood. “We are results-oriented at MJFF, and so our risk appetite is different from other funding institutions,” Hood says. “We constantly ask ourselves, ‘Can we shorten the time from discovery to intervention?’ To that end, we’ll fund new approaches if they can uncover a way to do research better and faster.”

Hood points to the positive reaction of a MJFF’s patient community to their collaboration with 23andMe: “People are excited that we are experimenting with a research method that lets them get involved. 23andMe provides a platform for directly reaching people with a specific disease and collecting important information that doesn’t exist anywhere else.”

It’s also easier for patients to participate in studies. Many newly diagnosed Parkinson’s patients are still working and able to continue their normal activities. They often find it a hardship to join a traditional study: travel to a clinical site, undergo an exam and interview, and then return a number of times over the course of the study. The 23andMe approach removes those barriers.

Like any funding institution, MJFF wants to maximize the impact of its spending. Dr. Sherer points to economics as a benefit of working with 23andMe: “Using traditional methods, getting 4,000 study participants would cost millions of dollars. 23andMe has done it for a small fraction of what it would have cost otherwise.” (Published studies show recruiting costs of approximately $2,000 per enrolled subject for a typical clinical study, compared to a quarter of the cost using 23andMe for an entire study, from recruitment to analysis.)

### 23andMe: Committed to Furthering Disease Research

Success in collaborative research depends on the commitment that each party brings to the venture. “23andMe is more than just another company trying to make money,” asserts Dr. Langston. “I have a great deal of confidence that 23andMe has a genuine interest in furthering the science through effective research.”

Hood concurs: “It’s going to take the whole community—scientists, foundations, philanthropists, patients, and commercial enterprises—to find cures for complex diseases. 23andMe has the expertise and is without question committed to this kind of work.”

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1. As of June 2010, 23andMe’s scientific team has identified five PD gene associations that were previously unknown: MCCC1/LAMP3, RIT2, SYN3, LOC729882, and SCARB2. Published genome-wide association studies (GWAS) in European populations had previously reported three associations with PD that were significant genome-wide: MAPT, SNCA, and PARK16 (Simon-Sanchez et al. (2009) Nat Genet; Edwards et al (2010) Ann Hum Genet). While additional genes have been associated with PD (e.g., LRRK2, GBA and PARKIN), these genes are not typically typed in a GWAS and thus are not included in this total.