Expansion of PD GENEration Return of Results Structure into Latin America through LARGE-PD

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PD GENEration

INTRODUCTION

Genetic testing is becoming standard of care and is increasingly required as inclusion criteria in clinical trials. However, a shortage of genetic counselors limit patient access to services. Many physicians, though they lack formal genetic counseling training, are interested in returning genetic results to patients.

In 2024, the PD GENEration study, in collaboration with the LARGE-PD consortium, expanded its initiative to return genetic results to Latin America.

To support capacity building, tailored training modules were developed to equip local providers with the knowledge required to return genetic results for seven Parkinson's disease-associated genes.

OBJECTIVES

Overview of the Expansion: Present the scale and impact of the initiative.

Genetic Counseling Capacity Building: Detail efforts to train and certify local providers in genetic counseling.

Progress Update: Provide an update on pilot sites, participant recruitment, and training status.

METHODS

Feasibility Assessment
- Evaluated the feasibility and logistics of LARGE-PD sites through a survey.
- Integrated a refined protocol and results-return pipeline into the existing framework of the LARGE-PD study.

Training Assessment
- Conducted a needs-based assessment to identify genetic counseling training requirements for consortium sites.

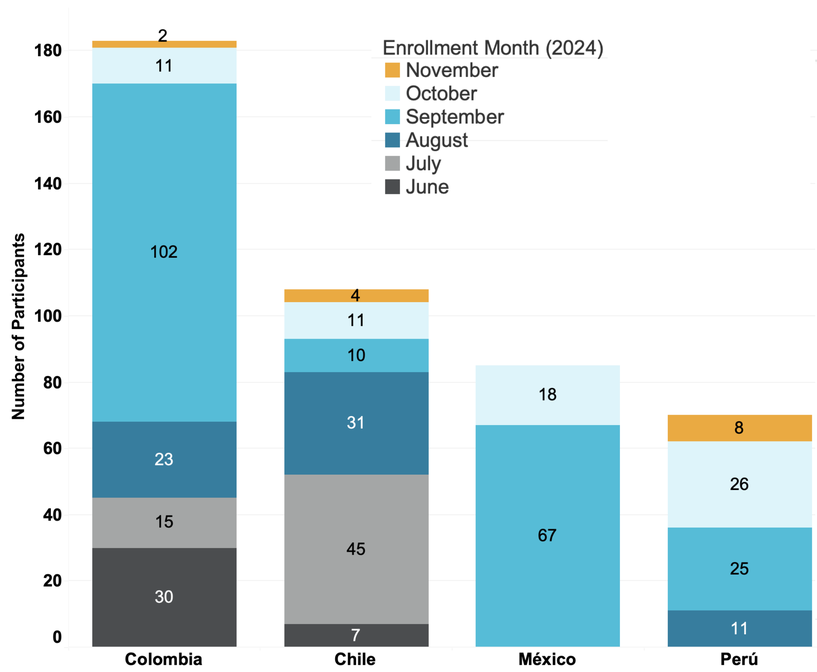
RESULTS

Pilot Sites
- Six pilot sites were selected after evaluating their infrastructure and regulatory alignment.
- Five sites are actively recruiting participants, while one site is awaiting final approval.

Participant Recruitment
- 446 participants recruited across active sites to date.

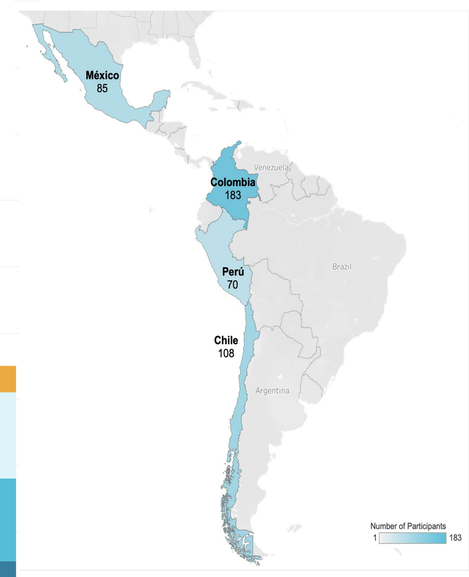
Genetic Counseling Training
- Each site has at least one trained provider who has completed the tailored genetic counseling training.
- 26 providers participated; 16 certified to return genetic results to participants

Figure 1. Monthly recruitment per country



*El Salvador has not started recruitment under merged protocol

Figure 2. Total recruitment numbers



CONCLUSION

The overarching objective of this collaboration is to improve access to genetic testing and counseling services for individuals with Parkinson's disease across Latin America.
This framework holds potential as a scalable model for implementing the study in other regions and delivering language-sensitive results to patients directly from their trusted physicians to the community.

ACKNOWLEDGMENT

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The authors sincerely thank all members of the Latin American Research Consortium on the Genetics of Parkinson's Disease (LARGE-PD) for their invaluable contributions, including their time, dedication, and steadfast commitment to every aspect of this study. We extend our deepest gratitude to all Parkinson's disease patients and their families from Latin America, whose participation and support have been integral to the success of this project.

Recruiting Underrepresented Populations in the Hawaiian Islands into PD GENEration: A Genetic Testing and Counseling Study for People with Parkinson's

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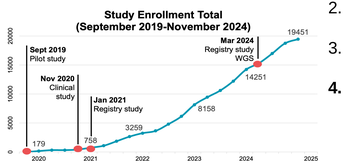
PD GENEration

Background

Four Phases of PD GENEration



1. Pilot study—launched in Sep. 2019 and aimed at feasibility.
2. Clinical study—launched in Nov. 2020; a continuation of the Pilot study at a larger scale, aimed at frequency and characterization of clinical phenotypes.
3. Registry study—launched in Jan. 2021 and aimed to make genetic counseling and testing accessible to 15,000 participants.
4. **Registry study WGS** (current)—launched in Mar. 2024 and aims to offer whole genome sequencing (WGS) genetic testing and counseling to 8,400 participants. While past enrollment has focused on the general Parkinson's population, this year the study focused on engaging and enrolling underrepresented populations (URPs) in partnership with Aligning Science Across Parkinson's (ASAP) and the Global Parkinson's Genetic Program (GP2).

Parkinson's Foundation in the Hawaiian Islands

The Parkinson's Foundation has had an ongoing partnership with the Hawai'i Parkinson's Association (HPA) since 2018, leading the foundation to many future partnerships, including but not limited to our current PD GENEration site, the Queen's Medical Center. Queen's Health first joined the Parkinson's Foundation's Comprehensive Care Center (CCC) in our Global Care Network (GCN) in July 2022. In 2023, the Parkinson's Foundation executed their first educational event in Hawaii, working alongside Queen's Health and the Hawaii Neurological Society.

PD GENEration Enrollment History in the Hawaiian Islands

- In August 2022, the first PD GENEration participant from Hawaii was enrolled.
- On average, 0-4 participants were typically enrolled per month in PD GENEration via the remote enrollment pipeline.
- In July 2023, Queen's Medical Center was onboarded to PD GENEration, increasing in-person enrollment to an average of 19 participants per month.
- Overall, Foundation partnerships with the Hawai'i Parkinson Association (HPA), Queen's Health Medical Center, and Jerry Boster (Former Board President of HPA and Parkinson's Foundation Hospital Safety Ambassador) were instrumental in bringing the PD GENEration study to Hawaii.

PD GENE Hawaiian Island Enrollment Prior to Event

Hawaii Enrollment Aug22-Sep24

62% Male

38% Female

236 Participants

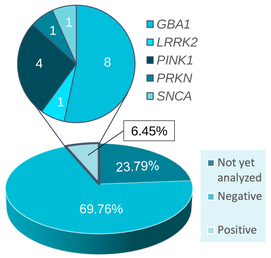
Average age of diagnosis
63.1 years old

Average age of onset
60.6 years old

Demographics

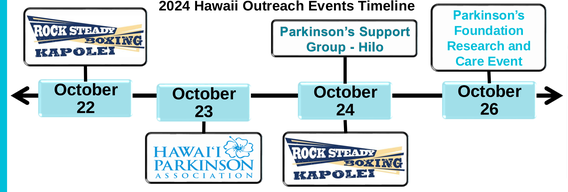
Race	
American Indian/Alaskan Native	2
Asian	111
Black/African American	1
Native Hawaiian/Pacific Islander	10
White	87
Other	1
More than one	33
Did not answer	3
Ethnicity	
Hispanic or Latino	13
Not Hispanic or Latino	230
Did not answer	5

Positivity Rate in Hawaii Cohort

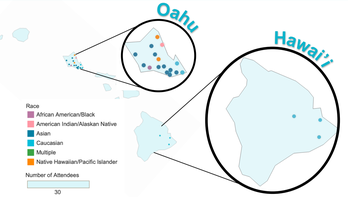


Methods and Results

Participation



Through outreach at local support groups, community opportunities, and healthcare professionals, the PD GENEration study was mobilized to bring information, resources, and live testing to members of the local Hawaii community. This was done by meeting with individuals at support groups, exercise classes, and at the HPA before the main Research and Care event. The event, the Hawaii Research and Care Event, was held on October 26, 2024 at the Hawai'i Convention Center.



Recruitment Event Details and Demographics

Rock Steady Boxing, Kapolei, 10/22

Rock Steady Boxing, Kapolei, 10/24

Research and Care Event, Honolulu, 10/26

30
Participants enrolled throughout the week

Male : Female
77% : 23%

Average age of diagnosis:
65.1 years

Average age of onset:
63.7 years

Race	
American Indian/Alaskan Native	1 (3.3%)
Asian	11 (36.7%)
Black/African American	1 (3.3%)
Native Hawaiian/Pacific Islander	4 (13.3%)
White	13 (43.3%)
Ethnicity	
Hispanic or Latino	1 (3.3%)
Not Hispanic or Latino	29 (96.7%)

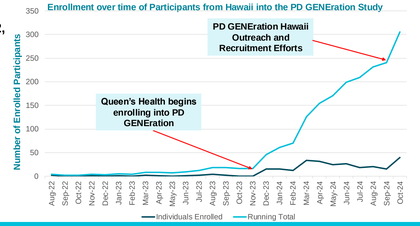
Increase in Enrollment from Hawaii from Recruitment Event

236 participants enrolled from Hawaii between August 8th, 2022, and October 21st, 2024.

30 participants enrolled through PD GENEration outreach and recruitment between October 24th, 2024, and October 26th, 2024

▲12.71%

Enrollment over time of Participants from Hawaii into the PD GENEration Study



▲40%
Increase in Native Hawaiian/Pacific Islander enrollment

Participants who had never participated in PD related research before
90%

56.7%
of enrollment was diverse populations

Study Impact and Local Community

Bringing the PD GENEration study to the Hawaiian Islands is vital for representing underrepresented communities and addressing their unique needs. By engaging with the community, understanding their history and values, and ensuring clear communication about the study's purpose and benefits, we aimed to build trust and encourage informed, voluntary participation. Collaborating with local leaders helped create an inclusive and culturally sensitive environment.

Impact

Lessons Learned

•**Community Engagement**: Through PD GENEration's outreach, we gained valuable insight into the population of Hawaii and their community's historical mistrust of the medical field, as well as potential hesitance towards sharing personal health information.

•**Historical Context**:
- Western explorers introduced diseases to the Hawaiian Islands and the native population had no immunity to these diseases.
- To prevent the spread of illness, individuals were often isolated, shunned, or sent away.
- These practices led to generations of secrecy and fear regarding personal health matters.

•**Impact on Health Participation**:
- This history has resulted in:
  - Hesitancy to participate in support groups, medical events, and research.
  - A deep-rooted mistrust of healthcare systems and sharing of health information.

- **Impact on Study Participation:**
  - After outlining the study's purpose, goals, personal connections to Parkinson's Disease, and the rationale for conducting the research in the Hawaiian Islands, potential participants expressed increased interest in learning more about the study and participating.

"Having PF come not once but twice to our State was **dramatic, necessary** and **generous.** What is important for our folks is **visibility** and **connection.** You can imagine how hard it is to be in the middle of the Pacific and not sure you are getting the proper care and have the necessary resources available. Bless you and the work you all do as it **truly does make a difference.**"

- Pat Bemis, Research and Care Event Attendee

Conclusion

Bringing PD GENEration recruitment events to the Hawaiian Islands not only allows for the increased participation of URPs in clinical research but ultimately facilitates the contribution of valuable and unique genetic data to aid the understanding of Parkinson's Disease in these populations. Physical presence in the community has facilitated greater engagement with the Hawaiian population, resulting in increased enrollment of local participants in the study.

More about PD GENEration

Implementation and Use of the Tasso+ Device to Support Whole Genome Sequencing in PD GENEration

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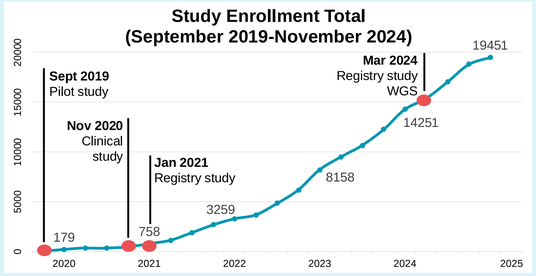
1. Parkinson's Foundation, New York, NY, USA; 2. Fulgent Genetics, El Monte, CA, USA; 3. Tasso Inc, Seattle, WA, USA; 4. Navitas Clinical Research, Rockville, MD; 5. National Institute of Health, Bethesda, MD, USA; 6. Tel Aviv Medical Center, Tel Aviv, Israel and Columbia University Irving Medical Center, New York, NY, USA

PD GENEration

Abstract

Objective: To describe the implementation and use of the Tasso+ device, a novel blood collection tool, in the PD GENEration study to support the transition to whole genome sequencing (WGS) in the US. **Background:** Since its launch in 2019, PD GENEration—a multi-center observational study that offers genetic counseling to people with Parkinson's disease (PWP)—has enrolled over 18,000 participants. A key factor in the success of enrollment has been the ability to conduct the entire study from home. In March 2024, the study transitioned from targeted exome sequencing to WGS, requiring a shift from buccal swab collection to whole blood sampling for DNA quality purposes. With the introduction of blood collection, the study needed a solution to maintain its level of accessibility. **Methods:** PD GENEration deployed the Tasso+ device in May 2024 in the US. The Tasso+ device is a sterile blood lancing device that can be used either in clinic or independently at home by PWP. To assist with its implementation, key resources, such as instructional videos specifically for PWP and device usage materials were created, and live participant support was made available with Tasso's proctoring service and the Parkinson's Foundation Helpline. **Results:** As of November 5, 2024, 1,853 participants have been successfully enrolled into the WGS phase of PD GENEration in the US, with 1,132 (61.1%) participants enrolled using the Tasso+ device. Of these participants, 857 (46.2%) were enrolled remotely at home and 996 (53.8%) were enrolled in the clinic setting. As of 11/5/24, 367 Tasso samples have been successfully extracted by the lab with a 2.9% failure rate. **Conclusion:** The implementation of the Tasso+ device has allowed for the successful transition to WGS and ensures the study remains accessible to all PWP regardless of where they receive care and how they enroll in the study.

Background



Four phases of PD GENEration:

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WGS Gene Panels

Primary Gene Panel	Secondary Gene Panels
GBA1 *LRRK2* *PRKN* *SNCA* *PARK7* *PINK1* *VPS35*	**21 PD related** *RAB39B, VPS13C, PTRHD1, SYNJ1, POLG, DNAJC6, ATP13A2, DCTN1, ATP1A3, SLC6A3, TH, GCH1, FBXO7, PLA2G6, ATP7B, MAPT, GRN, TBK1, VCP1, RAB32, CHCHD2* **10 Non-PD related (CDC Tier 1)** *BRCA1, BRCA2, MLH1, MSH2, MSH6, PMS2, EPCAM, LDLR, APOB, PCSK9* Hereditary Breast and Ovarian Cancer Syndrome (HBOC) Lynch syndrome (LS) Familial hypercholesterolemia (FH)

Transition to WGS and the Tasso+ Device

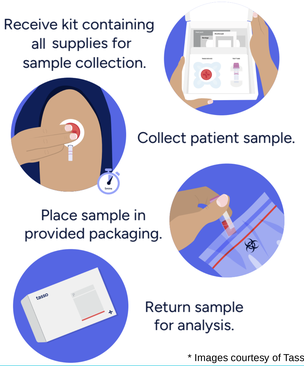
The launch of WGS required the study to change from buccal swab collection to whole blood sampling for DNA quality purposes.

Whole blood collection traditionally can be a barrier to study participation:

- Logistical issues with travel to a clinic
- Fear of needles, worries about pain or discomfort, and anxiety can deter enrollment

The Tasso+ device is a sterile, disposable blood lancing device that allows for self-collection of microliter capillary whole blood samples. Capillary blood collection is generally less invasive and less painful than venipuncture, and the device can be utilized either in a clinic or at home by participants who have enrolled remotely.

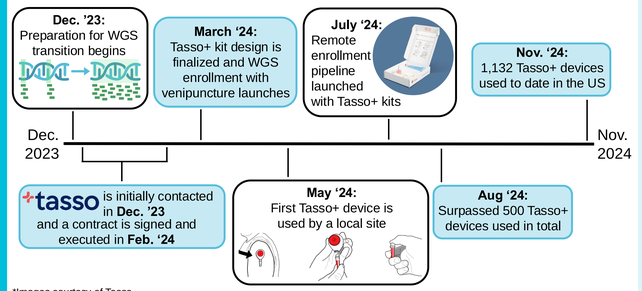
Sample Collection Process



* Images courtesy of Tasso

Methods

Timeline of Tasso+ Device Implementation



*Images courtesy of Tasso

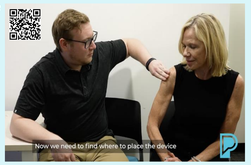
Key Resources for Tasso+ Device Implementation and Use

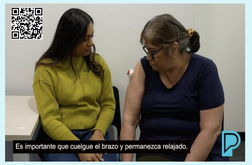
Available on our Study Resources Webpage:

- Instructional videos of at-home Tasso+ device usage by PWP (alone and with Care Partners) available in English and Spanish
- Written step-by-step instructions that mirror the video with pictures
- An FAQ document with commonly asked questions

Available to Local Sites:

- Training on Tasso+ device use and PD GENEration
- Study resources related to Tasso+ devices





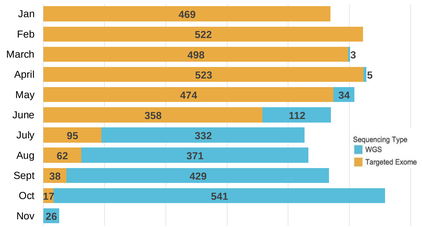
eMed — Live 1-1 video or phone instruction for Tasso+ use in English or Spanish for remote participation

tasso SUPPORT — Live team available for participants to call with questions related to their Tasso+ kit and shipping

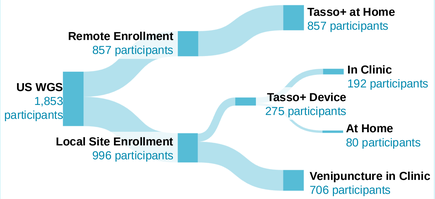
Parkinson's Foundation Helpline — Live team of PD information specialists who can answer participant questions

Results

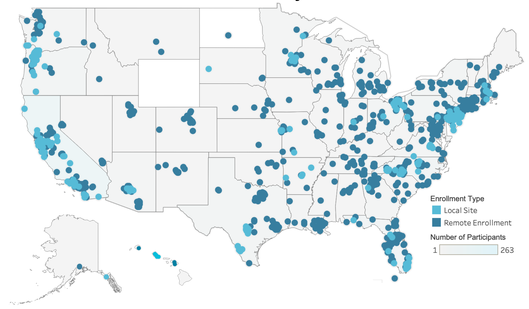
US Targeted Exome vs WGS Monthly Enrollment in 2024



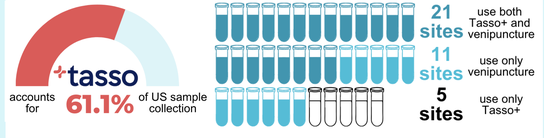
Participant Flow through PD GENEration



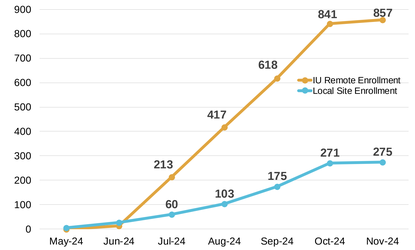
Distribution of Tasso+ Use by Enrollment Method



Tasso+ Kit Usage in the US



Total Tasso+ Device Usage in 2024



Lab Analysis of Tasso+ Device Samples

Of **672** samples received by the lab:

- **367** have been successfully analyzed
- **11** have failed quality control checks
- **305** are in progress

Average Concentration 48.12 ng/µl

Median Concentration 42.05 ng/µl

With a failure rate of only **2.9%**, utilization of the Tasso+ device proves to be an effective method of whole blood sampling for WGS, while simultaneously reducing common barriers to participation in research.

Conclusion

The implementation of the Tasso+ device has enabled a successful transition to WGS and ensures the study remains accessible to all PWP, regardless of where they receive care and how they enroll in the study. Key resources, such as access to the Parkinson's Foundation Helpline and tailored instructional materials, remain essential to remote usage and will continue to be evaluated by the study team to ensure successful implementation continues.

More about PD GENEration