APGS
AUSTRALIAN PARKINSON'S GENETICS STUDY

News

Issue 3 Winter 2025

Thank You for Being Part of This Journey!

Since the beginning, our mission at APGS has been simple: to better understand Parkinson's disease by listening to the experiences of the people living with it—and learning from their DNA. We're now **20,000 participants** strong, and we couldn't have come this far without you. This newsletter is our way of keeping you in the loop, sharing what your contribution is helping to uncover, and what's coming next.

Associate Professor Miguel Renteria and Professor Nick Martin, on behalf of the APGS Research Team

What is **APGS** all about?

More than 150,000 Australians are living with Parkinson's disease. We know it affects everyone differently, and we still don't fully understand why. That's where you come in.

The Australian Parkinson's Genetics Study (APGS) is one of the largest efforts worldwide to study Parkinson's through both questionnaires and DNA. Our goal is to figure out how your genes, environment, and lifestyle all interact to shape Parkinson's risk, symptoms, and progression. We're doing this by bringing together data from thousands of people like you across the country.

We're also part of a global team, working with researchers in over 65 countries through the Global Parkinson's Genetics Program (GP2). By working together, we can move faster and go further in the search for answers and better treatments.

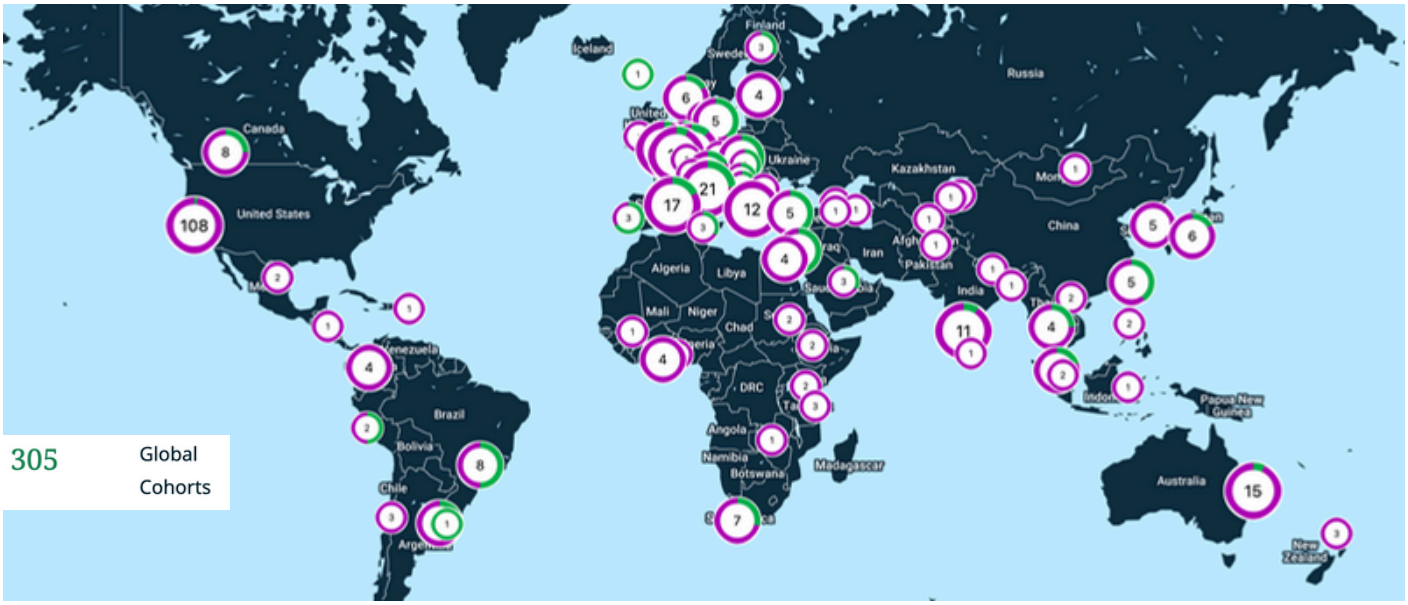
QIMR Berghofer

Recruitment update: We've come a long way!

Thanks to your support, as of 31 July 2025:

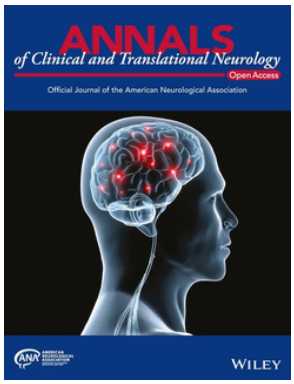
- Over **20,000 volunteers** have signed up to participate in APGS (including over 11,000 diagnosed with PD).
- Over **15,000 saliva samples** have been processed by our lab.
- Over **9,000 DNA samples** have been genetically characterised, and more are on the way.

Globally, the GP2 consortium has now analysed nearly **83,000 DNA samples**, including nearly 5,000 from APGS. This growing dataset is already helping scientists make important discoveries.



Largest study of **chronic pain** in Parkinson's

If you remember answering questions about chronic pain in the **APGS survey**, you helped shine a light on a symptom that often goes unnoticed in research and clinical care.



Using data from thousands of participants, we analysed how pain affects people living with Parkinson's. We're pleased to share that our study has just been accepted for publication in the ***Annals of Clinical and Translational Neurology***.

Here's what we found:

- Three out of four participants with Parkinson's reported living with chronic pain.
- The neck and back were the most common pain sites.
- Women were more likely than men to report pain.
- Pain was often linked to depression and other health issues.
- Only a small group said pain was their first PD-related symptom, showing it tends to appear later in most patients.

Why does this matter?

Chronic pain can significantly affect quality of life, yet it's often under-reported or overlooked during treatment. Your responses are helping us raise awareness, improve understanding, and push for more comprehensive care for people with Parkinson's. Thank you for being part of this important work.

You can read the preprint version of this study here:
https://www.medrxiv.org/content/10.1101/2025.02.09.25321969v2

Global Collaboration Uncovers **59 New Parkinson's Genes!**

The **Global Parkinson's Genetics Program** (GP2) has just published its **first major study**, and it's a landmark moment for Parkinson's research. This international project analysed **genetic data from over 63,500 people with Parkinson's and nearly 1.7 million controls**, making it the largest study of its kind so far. Thanks to your participation, ~3,000 Australian participants from APGS were included in this research and helped drive its success—and thanks to you, this number will increase greatly in the next version!

The study identified 134 genetic regions associated with Parkinson's disease, including 59 that had never been reported before, providing a better picture of the biological pathways involved in Parkinson's risk.

Some of the key insights include:

- ***Immune system involvement:*** The results strongly reinforce the role of immune-related genes, including those involved in lysosomal function and antigen presentation—suggesting that immune system dysfunction may be central to how Parkinson's develops.
- ***Cell types and brain regions:*** Using follow-up analyses, researchers pinpointed specific brain cell types (like microglia and dopaminergic neurons) and brain regions (including the substantia nigra and putamen) where Parkinson's risk genes are most active.
- ***Targetable genes:*** Several risk genes may be promising targets for future therapies. These include genes that regulate inflammation, support protein balance, and help cells clear out or fix damaged proteins.

This study provides the strongest genetic foundation yet for future work in risk prediction, early diagnosis, and new treatment strategies. While the manuscript is currently undergoing peer review, you can read a preprint online: https://www.medrxiv.org/content/10.1101/2025.03.14.24319455v1

Major **Scientific Breakthrough** in Cell Therapy for Parkinson's

Two independent clinical trials have reported encouraging progress in a potential new treatment for Parkinson's disease: **cell therapy**. These breakthroughs were featured on the **cover of *Nature* magazine**—one of the world's most prestigious scientific journals—on **22 May 2025**.

Parkinson's is caused by the gradual loss of brain cells that produce dopamine, a chemical essential for movement and coordination. While current medications can ease symptoms, they don't stop or reverse the underlying disease—and their effectiveness tends to fade over time.

These new trials in Japan and the US explored whether replacing lost cells with lab-grown, dopamine-producing neurons could help.

Both teams successfully transplanted these cells into the brains of people with Parkinson's. The results were promising: no serious side effects were reported, and there were early signs of improved motor symptoms in some participants.

While much more research is needed to confirm long-term safety and effectiveness, these trials mark a major step forward. They bring us closer to the possibility that cell therapy could one day slow, stop, or even reverse the course of Parkinson's disease—not just manage its symptoms.

What's Happening With the **DNA Results**?

By September 2025, we will have completed genetic analysis for more than 9,000 APGS participants, with results for the remaining participants available by the first half of 2026.

If you're interested in **receiving a summary of your research results**, we're developing an **opt-in process** that will allow you to request this information through your nominated clinician. Your clinician can help explain any findings we make that are relevant to you, and if appropriate, refer you to a genetic counsellor or arrange for clinical-grade genetic testing to confirm the results.

It's important to note that these are **research-level findings**. So they shouldn't be used to guide medical decisions unless they are validated in a NATA-accredited (or equivalent) clinical laboratory. Research results can offer insights—but they are **not a substitute for clinical testing or advice**. We'll share more details about this process as it becomes available.

Your **Voice** Matters: Studying **Speech** in Parkinson's

Speech and language changes—like reduced volume, unclear speech, or word-finding difficulties—are common in Parkinson's, but not always well understood or monitored.

To learn more, we've partnered with Prof. Adam Vogel, a speech pathologist at the University of Melbourne, using AI to study how speech may help track disease progression.

Over 2,000 participants have already taken part—thank you! If you haven't yet and have received an invitation, it's not too late. If you have not heard from us yet, you should get an email in the next month or so. The study involves a few short voice tasks done online, from home. It takes less than 10 minutes and is safe, simple, and could shape the future of symptom monitoring.

We've also published a literature review on this topic in npj Parkinson's Disease, available here: https://www.nature.com/articles/s41531-025-00913-4

THE UNIVERSITY OF MELBOURNE

REDENLAB

Young Onset Parkinson's Sub-Study Launching Soon!

MONO PDAUS
MONOGENIC PARKINSON'S DISEASE INITIATIVE - AUSTRALIA

Parkinson's that begins at a **younger age** (typically before 50), especially when there's a **family history**, is more likely to have a strong genetic cause. To better understand this, we're launching the Monogenic PD Australia (MonoPDAus) Initiative.

Thanks to a $2.95M grant from the **Medical Research Future Fund**, we're working with a national team of neurologists, researchers, and genetic counsellors to make this possible. **If you're eligible, we'll contact you directly**. Participation is voluntary.

MonoPDAus participants will receive **whole genome sequencing** (covering all 3 billion letters of DNA) and a telehealth **clinical assessment with a Parkinson's specialist**. You'll also have the option to talk to a **genetic counsellor** and **choose whether to receive your results**. This initiative is especially timely, as new treatments targeting specific genes are in development—raising the possibility of more personalised therapies in the near future.

Coming Soon: **Wearables** to Track Parkinson's in Real Time

We're working on an **exciting new project** that will use wearable sensors—like **smartwatches**—to monitor Parkinson's symptoms continuously, unobtrusively, and remotely. This will allow us to better understand how symptoms change over time and why people experience Parkinson's differently.

By combining this real-world data with genetic information, we hope to uncover how genes influence symptom type, severity, and progression. This could be a major step toward more personalised care. Stay tuned—**APGS participants may soon be invited to take part!**

Your **Involvement** Matters

Thank you for your continued trust, time, and generosity. Research is a long journey, and it's only possible thanks to people like you. Every questionnaire, sample, and story brings us closer to understanding Parkinson's and improving lives.

This is a true team effort—scientists, clinicians, and participants working together toward a shared goal. Your contribution isn't just valuable—it's essential.

As always, we welcome your feedback, questions, and ideas. Thank you for being part of this important work. Together, we're unlocking the genetic puzzle of Parkinson's and building hope for the future.

Get in touch!

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