

NBIAcure NEWSLETTER

-2022 RECAP-

Welcome back to the NBIAcure Newsletter from our team at OHSU! A lot has happened since our last issue in 2021 and we're excited to reinstate the newsletter, beginning with a recap of 2022. It has been a busy and gratifying year. We'd like to thank all of the members of the NBIA community.

Natural History Study Updates

Over the past year, the natural history studies of PKAN, PLAN, and BPAN have seen a lot of progress. These studies are crucial for the furthering of research and the development of new interventional trials. In fact, the FDA highlights the importance of natural history studies in its [publications](#) and its interactions with rare disease communities like ours. Natural history studies gather information about disease progression over time in order to identify disease markers and patterns, a necessary step in the research process. We so deeply appreciate everyone's generous participation at this stage of the work as it is an integral part of our trial readiness. If you're interested in learning more or enrolling yourself or your child in PKANready, PLANready, or BPANready, please visit [our website](#).

PKANready PLANready BPANready fast facts

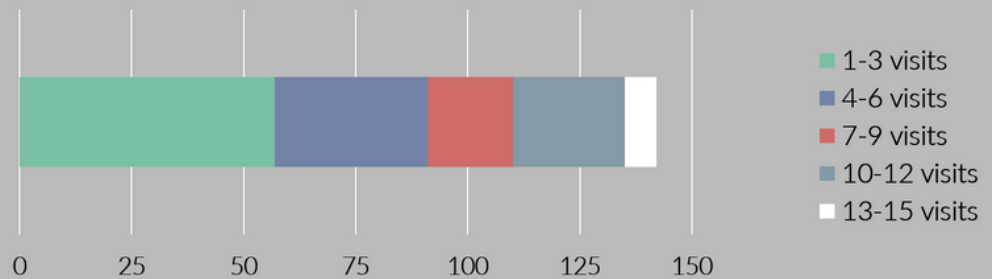
250+ participants enrolled

30+ countries represented

16+ years of total data collection

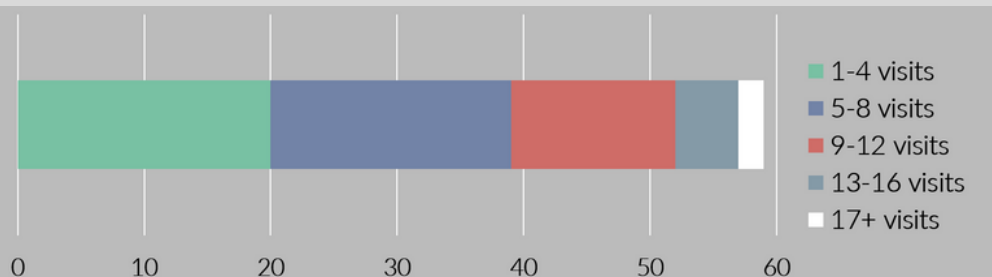
PKANready

PKANready currently has 142 participants actively enrolled.



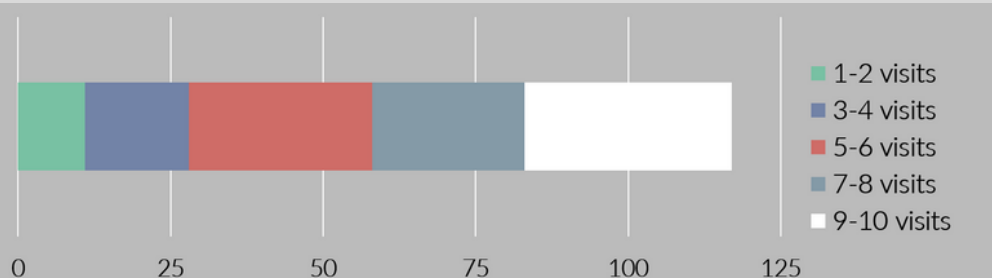
PLANready

PLANready currently has 59 participants actively enrolled.



BPANready

BPANready currently has 117 participants actively enrolled.



CoA-Z Clinical Trial Updates

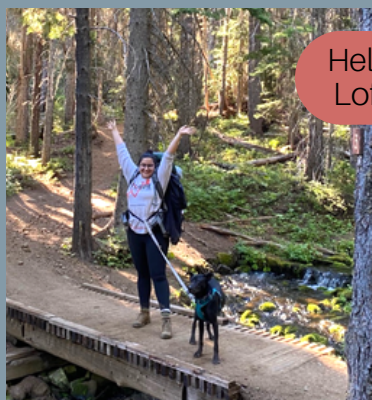
As many of you know, the CoA-Z in PKAN clinical trial has now come to an end. We were happy that so many in the PKAN community joined the trial, and in fact, the early enthusiasm encouraged us to change our original plan for the study, extending its planned duration from two to three years and expanding the number of people enrolled. Of course, that was before the full impact of the COVID-19 pandemic was known! As the world shut down in 2020 and we saw other clinical trials closed or delayed, we were so pleased that our unusual home-based approach enabled our own CoA-Z trial to continue successfully. Sadly, though, the pandemic did eventually catch up with us: a supply chain problem (compounded by that Texas deep freeze in 2021) led to production delays of CoA-Z, forcing us to make the difficult decision to return to the study's original design. This did not affect the length of the 6-month "double-blind" portion of the trial, which is key for analyzing the data (this is the time when nobody knows who was taking active CoA-Z and who was taking placebo), and in fact, many participants were able to complete two years in the study as originally planned. Now, we are underway with the important next steps: 1) running multiple blood samples from each participant for biomarker analysis and 2) cleaning up all the data we have collected over the past 2+ years so that it will be ready for statistical analysis. We plan to stay in touch with the PKAN community as we proceed.

We are grateful to each individual with PKAN and their amazing families and caregivers who helped make the trial work. We could not have done it without you!



—Meet the Newest Team Members—

The past 18 months have seen a lot of new faces since Dolly and Katrina have moved on to begin medical school!



Helena
Loftus

Lab Research Assistant

Where did you grow up?

Ashland, OR

What are your hobbies?

Reading, ceramics, hiking,
watching soccer

Favorite place you've traveled?

Paris, France



Dustin Le

Clinical Research Assistant

Where did you grow up?

San Jose, CA

What are your hobbies?

Basketball, swimming,
photography, traveling

Favorite place you've traveled?

Nha Trang, Vietnam



Kira
Anderson

Clinical Research Assistant

Where did you grow up?

Minneapolis, MN

What are your hobbies?

Mountain biking, reading,
gardening

Favorite place you've traveled?

San Salvador, El Salvador

Gene Therapy Project

Three NBIA family organizations have supported an important gene therapy project in a PKAN mouse model. In PKAN, the altered *PANK2* gene usually means the body's cells cannot make a working PANK2 protein. This project aims to test if providing a correct copy of the human *PANK2* gene will allow cells to produce a complete PANK2 protein. In 2022, we finished a preliminary trial of this project, which primarily focused on testing efficacy of treatment. We are very encouraged by the results of that work and are moving forward on this project with involvement of additional parties. While we are very excited about the prospect of gene therapy in PKAN, we still have a long road ahead of us before this potential treatment might move into human trials. We sincerely appreciate family organization support of research projects exploring potential treatments for NBIA disorders.



Dr. Hayflick and Helena (Research Assistant) look on as gene therapy injection is performed by Dr. Shelley Winn



Rare Disease Day 2023!

Rare Disease Day 2023 is coming up on February 28th! Rare Disease Day is an international advocacy event for the more than 300 million people worldwide who live with a rare disease. Our team has already begun planning some exciting festivities for the occasion. We'll be illuminating Portland's Morrison Bridge with the NBIAcure colors to raise awareness for NBIA disorders. Join us in commemorating Rare Disease Day with pink, blue, green, purple, and all things Zebra. For more information, visit rarediseaseday.org



Share your colors
and show your
stripes!

Mark your
calendar for Feb
28th!

Study coordinator pets Bucky and Phoebe eagerly preparing for Rare Disease Day. They heard there would be treats.



RARE DISEASE DAY®