

## A Recap of 2018

### BPAN Progress

#### Studies

- Natural history study, BPANready, was launched in May and 65 people have enrolled so far.
- Remote sample collection was started for BPANready

#### Publication

- Expanded phenotype paper (IN PROGRESS)

#### Collaborations

- We shared samples with Munich and University College of London investigators
- We are exploring a collaboration with TGEN and Melbourne Institute

### Dr. Hayflick's 60th Birthday (And no, she's not retiring!)



### Collaboration being explored

Our team had an initial call with Dr. Timothy Yu and his associate, Dr. Darius Ebrahimi-Fakhari, after his new work in Batten disease was highlighted on social media and seen by many NBIA families. Dr. Yu attempts to treat rare genetic disorders with oligonucleotide treatment and is interested in applying the technology to other disorders. This call was an initial exploration of how we can work together. We've agreed to share de-identified mutation information with him from cases in our NBIA repository. He hopes to identify one or more individuals whose genetic change(s) would be amenable to oligonucleotide treatment. We will keep the community updated if this collaboration moves forward. You can read more about Dr. Yu's work [HERE](#).

### MPAN Progress

#### Publications

- Autosomal dominant paper with insights into C19orf12 protein (UNDER REVIEW)
- Gene Review updates (IN PROGRESS)

#### Studies

- Autosomal dominant and autosomal recessive MPAN neuropathology studies done with Dr. Woltjer and Marlous van der Weijden

### PKAN Progress

#### Studies/Trials

- We are developing CoA-Z for a clinical trial starting in early 2019
- Biomarker development
- 90 people have enrolled in natural history study, PKANready

#### Publications

- Mouse spermatogenesis paper (IN PROGRESS)
- CoA-Z Paper (IN PROGRESS)

#### Collaboration

- We are working with an early career scientist on gene therapy in the PKAN mouse model

Our team is busy preparing for the CoA-Z clinical trial. We know that many of you are waiting anxiously. While you wait, there are some steps you can take to speed up your enrollment in the trial. First, make sure you are enrolled in PKANready. If you are already in PKANready, please make sure to respond promptly to the PKANready surveys and emails. As we approach the trial start date, we will be contacting people about updating their PKANready surveys and getting any missing genetic testing or MRI records in order to smooth the enrollment process.

# PLAN Progress

## Studies

- 20 people have enrolled in natural history study, PLANready
- A letter was released about Desipramine as a possible treatment for PLAN

## Collaboration

- We shared information from medical records for a retrospective PLAN study with Dr. Manju Kurian in the UK

# MePAN

## Publication

- MePAN Gene Review, including reporting on the expanded phenotype (IN PROGRESS)

## Collaborations

- We shared samples with researchers in Finland
- We shared samples with the University of Utah
- Samples were shared with us from Stanford

# FAHN Progress

## Publication

- Gene Review was updated with new clinical information (LINK)

## Winston Meets Santa!



# Plans for 2019

## NBIADA Family Conference

We are excited to see families in Charleston for the 10th International NBIA Disorders Association Family Conference! We are looking forward to the disease specific group meetings and break-out session, as well as meeting new families and reconnecting with everyone we have met in the past.

## OHSU Clinical Appointments

- 30 patients were seen in 2018
- New visits are booked out until March 2019



Left to Right: Puneet Rai (genetic counselor), Dolly Zhen (research assistant), Tanya Brooks (administrative coordinator), Katrina Wakeman (study coordinator), Allison Gregory (genetic counselor), Suh Young Jeong (research assistant professor), Caleb Rogers (geneticist), Penny Hogarth (neurologist), Alison Freed (study coordinator) and Susan Hayflick (geneticist)

## CoA-Z Trial: *In Progress*

*Some steps are still in progress or pending before the trial can begin. Many of these are happening concurrently.*

- Contract with Quest Labs for safety lab blood draws
- Design/Build database for clinical trial
- Continued biomarker research
- Completion of shelf life study
- Final FDA approval of clinical trial design
- Logistics planning for shipping CoA-Z