NBIAcure NEWSLETTER

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Rare Disease Day 2023

Top row (left to right): Puneet Rai, Penny Hogarth, Kira Anderson, Suh Young Jeong, Alison Freed
Bottom row (left to right): Caleb Rogers, Susan Hayflick, Dustin Le, Helena Loftus, Allison Gregory

Our team commemorated Rare Disease Day 2023 on February 28th by illuminating Portland's Morrison Bridge with the NBIAcure colors! We also sported our zebra stripes, handprints, and bright colors.

Statement About Rett Syndrome Medication

A new medication for Rett syndrome, called Daybue, was recently approved by the FDA. Since there are some clinical similarities between BPAN and Rett syndrome, including regression, seizures, intellectual disability, motor changes and certain repetitive behaviors, it is natural to wonder whether Daybue could help people with BPAN. We at OHSU continue to ask ourselves this question and are reviewing information and data that may help inform our thinking about this. It is important to understand that the common Rett gene, *MECP2*, and the medication in Daybue have a direct link and rationale that explain why Daybue works. This specifically relates to a protein called insulin-like growth factor 1 (IGF1) that is low in Rett syndrome and restored by Daybue. In BPAN, however, there is currently no evidence that IGF1 is reduced or plays a role in disease development. Still, it is worth exploring whether Daybue could benefit the BPAN population, starting with studies in BPAN animal or cell-based models. Even if it cannot, the lessons learned from the approaches to treating Rett syndrome, which is also X-linked, will likely be useful to BPAN investigators. For these reasons, we stay informed about Rett syndrome studies with an eye towards applications for BPAN.

Looking Forward to the 2023 Family Conference!

We are so excited to see all of your smiling faces at the NBIA Disorders Association Family Conference this May in Houston. Most of our team will be in attendance and presenting some of our recent work.
We will also be collecting research samples on Thursday and Friday at the conference. We deeply appreciate anyone's willingness to contribute a blood sample, and we have a particular need for healthy control samples from parents and siblings of someone with NBIA. The samples in our repository are an invaluable resource for the development of new treatments by researchers all over the world. If you or your family member are interested in contributing a blood sample, come find our team on Thursday or Friday!

For registration and conference information, visit nbiadisorders.org

NBIA Program at the Oregon Brain Bank

Although brain donation is a difficult subject, brain tissue has the capability to advance NBIA research in powerful ways. Past donations have answered questions that could not otherwise have been answered, and these precious gifts have provided crucial insights into the NBIA disorders. Our team would like to thank and acknowledge all of the families who have made the courageous and generous choice to contribute.
In our partnership with the Oregon Brain Bank, we take great care in keeping the process as smooth as possible so that families don't need to face additional stress during an already difficult time.
The Oregon Brain Bank registration form can be completed at any time. Having this information on file makes the process much easier when arrangements need to be made, but it is not a binding agreement and families may change their mind at any time. All costs for brain donations are covered by generous support from the lay advocacy community, for which we are grateful.

To learn more about the program with perspectives from families who have chosen to make this gift, please visit our website. If you have any questions or you are interested in registering, contact Allison Gregory at gregorya@ohsu.edu.

NBIA PROGRAM at the OREGON BRAIN BANK