



October 30, 2023

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## **JACK'S TOMORROW FUNDS RESEARCH AT YALE TO IDENTIFY MOLECULAR THERAPIES FOR PURA SYNDROME**

[New York, NY] – Jack's Tomorrow, a 501(c)(3) whose mission is to fund research to develop treatments and ultimately a cure for [PURA Syndrome](#), today announced \$250,000 for two years of research support to the work of two Yale faculty members at the [Yale University School of Medicine](#). PURA syndrome is a neurodevelopmental disorder caused by a faulty copy of the PURA gene. Symptoms include moderate to severe developmental delays, loss of speech, epilepsy, and impaired movement function.

Leading this new research are two Yale scientists with a strong track record in precision medicine approaches targeted to rare neurodevelopmental disorders like PURA Syndrome. [Yong-Hui Jiang, M.D., Ph.D.](#), is Chief of Medical Genetics and an expert in clinical and biochemical genetics of rare and undiagnosed diseases in children and adults. [Jiangbing Zhou, Ph.D.](#), Professor of Neurosurgery and Biomedical Engineering, creates innovative molecular therapies for neurological disorders.

"We are thrilled to be collaborating with Jack's Tomorrow to find targeted treatments for this rare disease," said Jiang. "By developing a lab-based model of PURA Syndrome that contains patient cells, we can rapidly and efficiently monitor disease characteristics and test potential therapies."

Three projects from the Yale team will build a strong foundation for guiding development of molecular therapies for PURA Syndrome, by:

- Generating induced pluripotent stem cells (iPSCs), which are derived from PURA Syndrome patients.
- Analyzing in detail the molecular and behavioral features of mouse models of PURA Syndrome, toward enabling further study of newly identified PURA therapies.
- Understanding how the RNA molecule [MALINC1](#) controls PURA activity in human and mouse cells.

"Our research is highly treatment-focused," said Zhou. "Understanding isn't enough – we need to use what we learn to develop patient-focused therapies that cure disease, not just treat its symptoms."

Despite ongoing research, a lot about PURA Syndrome remains a mystery. The culprit PURA gene was discovered only about 10 years ago, and scientists still don't know how the [more than 100 disease-causing genetic variations](#) or PURA affect children. As such, key unanswered questions stand in the way of a cure. Last year, Jack's Tomorrow worked with opinion leaders in academia, medicine, and industry to develop a [strategic research approach](#) that will pinpoint the molecular culprits causing the disease and then develop precisely targeted treatments.

To date, several projects are underway in addition to the new Yale research. Scientists at Toronto's [Hospital for Sick Children](#) (Sick Kids®) are conducting PURA Syndrome drug-screening studies using zebrafish and worm model systems. [The Jackson Laboratory](#) (JAX) in Bar Harbor, Maine is making a new knockout mouse model that carries PURA Syndrome patients' genetic variants, taking advantage of the [latest, most advanced genome-editing tools](#). Jack's Tomorrow is collaborating with the [Center for Precision Medicine Models](#) at Baylor College of Medicine in Houston to study PURA genetic variants in fruit flies. This NIH Office of Research Infrastructure Programs (ORIP)-supported world-renowned center develops [precision animal models for rare genetic diseases](#) toward exploring personalized treatments.

"Jack's Tomorrow is leaving no stone unturned for children who live with PURA Syndrome like our 6-year-old son [Jack](#)," said Kyle Czepiel, co-founder of Jack's Tomorrow. "Research provides not only the hope all of us need but also the fastest path to a cure."

### ***About Jack's Tomorrow***

At Jack's Tomorrow, our mission is to fund research to develop a treatment – and ultimately a cure – for PURA Syndrome.

For more information, please visit [www.jackstomorrow.org](http://www.jackstomorrow.org) or follow us on [Facebook](#), [Instagram](#), and [LinkedIn](#).

### ***About Yale School of Medicine***

Yale School of Medicine is one of the world's leading centers for biomedical research, advanced clinical care, and medical education.

For more information, please visit [www. www.medicine.yale.edu](http://www.medicine.yale.edu).