



SickKids®

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JACK'S TOMORROW FUNDS RESEARCH TO IDENTIFY A CURE FOR PURA SYNDROME

[New York, NY] – Jack's Tomorrow, a 501 (c)(3) whose mission is to fund research to develop a treatment – and ultimately a cure – for PURA Syndrome today announced a new grant to The [Hospital for Sick Children](#) (SickKids) in Toronto, providing \$180,000 to support cutting-edge science to identify candidate therapeutics for PURA Syndrome. This research aims to help patients worldwide who have been diagnosed with [this rare disease](#).

“We are thrilled to support SickKids research to help define a better future for children living with PURA Syndrome, like Jack, and many others with rare disease conditions,” said Kyle Czepiel, co-founder of Jack's Tomorrow and Jack's father. “Through this funding and more to come, we aim to expand the number of scientists who can uncover new knowledge about PURA Syndrome – and how to find treatments and cures.”

PURA Syndrome can be traced to a faulty version of the PURA gene. Affected children may have serious developmental delays, hypotonia, seizures, sleep apnea, and orthopedic problems. PURA Syndrome is considered a new disease, because the culprit PURA gene was discovered only about 10 years ago. There are currently 620 people living with the disease globally, and new cases are being diagnosed every week due to increased access to sequencing technologies. Strengthening basic and clinical research on PURA Syndrome is essential in order to identify a treatment for PURA Syndrome and ultimately a cure.

This new research will conduct large-scale drug-screening in zebrafish and nematodes, led by SickKids pediatric neurologist and scientist [James Dowling, M.D., Ph.D.](#), to identify potential treatments that can be used in animal models of PURA, and later, in humans. Dr. Dowling is a world-renowned clinician-scientist specializing in inherited neurogenetic disorders. This new work will harness the power of these model organisms, which are well-suited to accelerate clinical therapeutic development and improve the lives of patients with PURA-related disorders.

- Dr. Dowling and his team are creating a zebrafish model to study PURA Syndrome. Their preliminary research shows that “knocking out,” or eliminating, one of two copies of the fish versions of the genes (*puraa* and *purab*) leads to swimming defects, suggesting that *pura* genes in fish help control muscle behavior. The new funding will extend this work to remove both versions of the two fish genes and identify the best combination of gene alterations to perform additional testing. The scientists will then use an automated swim-measurement platform to perform large-scale drug screening and validation testing toward identifying potential new treatments for PURA Syndrome.
- The second project will employ *C. elegans*, a popular research organism for studying human disease. Using this nematode model of PURA Syndrome and a gene-silencing method called RNA interference, or RNAi, this research will identify molecules and cellular processes disrupted by loss of PURA. The researchers will then test the effects of various potential drugs

on fixing these problems in the worms. Those potential drugs can later be tested in a model more similar to humans.

Collaborating with Dowling are Brent Derry, Ph.D., of SickKids, and Peter Roy, Ph.D., of the University of Toronto. Derry's past research has identified therapeutic targets that reverse disease symptoms in animal models of rare neurological diseases affecting blood flow in the brain. Roy pioneered the use of roundworms to discover potential new drugs for potential application to human diseases.

To date, scientists have found [more than 100 disease-causing versions](#) of the human PURA gene, but more research is needed to understand how these gene variants cause PURA Syndrome or contribute to its symptoms.

In addition to this new research in zebrafish and worms, Jack's Tomorrow is partnering with [The Jackson Laboratory's Rare Disease Translational Center](#) to create a mouse model for advancing research on PURA Syndrome. Jack's Tomorrow is eager to continue to establish research collaborations toward treatments, and cures, for rare diseases like PURA Syndrome, said Czepiel.

"We need many smart minds tackling PURA Syndrome – groundbreaking research tools means that a better tomorrow for our son Jack and hundreds of others will be here sooner than we ever thought possible."

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 or follow us on [Facebook](#), [Instagram](#), and [LinkedIn](#).