



HED: Tunes for Tosh fund-raiser draws a crowd of 300

DEK: The Bingham family's April 5K and family fun run raised \$14,300 for University of Utah Health's Penelope Program.

After four years and appointments with seemingly every specialist in the Salt Lake Valley, Shauna and Brook Bingham felt no closer to finding out what was causing their young son's serious medical issues. Tosh suffered from a range of developmental disorders including speech, cognitive, and behavioral delays. Doctors had performed a spinal tap, an EMG, and an MRI on Tosh, testing him for a range of maladies from cerebral palsy and muscular dystrophy to neurofibromatosis and various genetic disorders.

"There were a lot of tears, sleepless nights, and hours talking about it, stressing and wondering what the future held for Tosh," Shauna said. "To not know what's wrong with your child is challenging in so many ways."

Then, last fall, Tosh was selected to be part of the Penelope Program, University of Utah Health's pilot program for undiagnosed and rare conditions in the Department of Pediatrics. After sequencing the DNA of Shauna, Brook, Tosh, and his older brother, Koa, the medical mystery was solved. Tosh was diagnosed with an extremely rare spontaneous, or de novo, change in one of his genes that affects how a part of his brain works, in particular its ability to

control gait, muscle coordination, and speech. For Tosh, this meant frequent falls and difficulty speaking.

The condition, named for the effected gene, KCND3, is so unusual that at the time of Tosh's diagnosis, there was only one other known case, found in a boy in the Netherlands. However, thanks to the global network the Penelope Program is part of, 15 more people were found with the same gene change in just the first six months following the discovery of Tosh's condition. Doctors are now working to better understand this condition and pave the road for better treatments.

Named Penelope after the wife of Odysseus in Homer's the *Odyssey*—a symbol of strength and hope—the Penelope Program conducts a comprehensive clinical and genetic analysis of children with mystery ailments. As Brook explained, the Penelope Program provides “a deep dive into genetic testing for children” to provide answers. By offering faster diagnoses and better care to families, the program aims to end what can be long, confusing and painful diagnostic odysseys and sets patients on a path to an integrated care plan. In Tosh's case, that means intense speech and occupational therapy.

“As you can imagine, we were ecstatic to find out a cause,” said Shauna. “And because we were selected into this program, we discovered an incredible support network. All of our testing was free, which was incredible—we couldn't have paid for it otherwise. Even after diagnosis, the doctors are still involved, helping facilitate any testing or specialist we might need to see. They're really passionate about helping us.”

Having found their own answers, Shauna and her family set upon their specific treatment course for Tosh. But Shauna was haunted by the thought that others must be out there having the same experience: caring for a child with an unknown malady, feeling alone and helpless. She felt compelled to help them.

“I wanted people to know about rare genetic diseases,” she said. “I wanted people to know about the Penelope Program and that it will only continue to exist if people give private donations and grants. I'm committed to help other families be part of this wonderful program. So, my husband and I created Tunes for Tosh.”

Using her experience from fund-raising for a local charity, Shauna established the Tunes for Tosh 5K and Family Fun Run. On April 29, a sunny, picture-perfect day, more than 300 people arrived at Sugar House Park ready to support children like Tosh.

“I had so much support between family and friends and the community,” Shauna said. “I could have never done it without all of their help. I cried like a hundred times that day.”

According to Shauna, the event was such a success because of its namesake: Tosh.

“On the day of the event Tosh was running around and riding his bike, making it his goal to make people laugh and cry,” Shauna said. “And though he frequently fell down because of his poor balance and muscle control, Tosh didn’t get upset, but jumped right back up and continued on his way. His perseverance and love for life helped people be present and embrace the love and laughter of the day... People want to be around him and to support him in any way possible. We even had people who were just walking around the park start donating.”

After the event, the Bingham family presented the proceeds of Tunes for Tosh to Dr. Lorenzo Botto—a professor of pediatrics at the University of Utah School of Medicine, clinician, and director of the Penelope Program—and the team who helped in Tosh’s diagnosis. The gift of \$14,300—which far exceeded Shauna’s original goal of \$5,000—will benefit other families and young patients like Tosh through the Penelope Program.

“We are deeply grateful for the generosity demonstrated during this event,” Dr. Botto said. “So many people, in and outside the Penelope Program, have contributed to making Tosh’s diagnostic odyssey a true journey of discovery. We are humbled by and grateful for the support and passion of Shauna and the Bingham family. With support such as this, we can help more families discover the roots of mysterious conditions, make a comprehensive care plan, and—with the many collaborations we have in Utah and internationally—pursue new treatments. This is the future of medicine.”

The Penelope Program’s team includes experienced clinicians from multiple specialties, including the Comprehensive Care Program and molecular geneticists. They put considerable time and effort into the program, working behind the scenes without compensation. Much of the team’s work is supported by the Department of Pediatrics through the vision of Dr. Edward Clark, the department chair, and by Primary Children’s Hospital, the Utah Genome Project and the Sorenson Foundation.

The team’s goal is to serve 100 families annually, with complete work-ups, testing, and access to specialists to diagnose patients. Dr. Botto and his team continue to screen and evaluate families, providing previously elusive diagnoses and continuing to care for patients and their loved ones like Tosh, now five years old, his parents, and his brother.

As for Shauna’s goals, she and her family aren’t stopping with this year’s fund-raiser; they’ve committed to hosting Tunes for Tosh annually.

“Thanks to the Penelope Program, we found answers for Tosh,” said Shauna. “Our message to other families seeking similar answers for their kids is simple—there is an answer out there for your family; you’re not alone.”

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