Winter 2016-2017

HELPING HANDS

FOR FRIENDS, FAMILY, AND SUPPORTERS OF THE CHILDREN'S GAUCHER RESEARCH FUND


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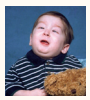
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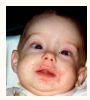
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INSIDE:

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IT'S ABOUT THE CHILDREN

In our last newsletter we said "Lots of science in this Newsletter". This is good news because there is lots of science happening and you, our donors, are responsible for much of the progress. But let us not forget why we do this … we do it for the children who are denied a chance at life. We dedicate this issue to the life of Xavier Edward Perusse… and to his parents… whose broken dreams transformed into a love and a commitment they never dreamed possible. ■


DEBORAH MACRES AND GREG MACRES

Xavier Edward Perusse

He will live strongly in our hearts forever...

Xavier Edward Perusse was born October 11, 2013. He was 7 lbs 8 oz, a beautiful, big, baby boy. Xavier was born with a full head of hair and deep, dark blue eyes. He was our first child and the day Xavier was bo rn, we were so happy to be welcoming him into a life of adventure, skiing, love and family. We initiated him early into the outdoor life, walking on the beach, sitting with him in our garden, and showing him the flowers and plants outside our house.

Our first six weeks with Xavier were a gift, blissfully unaware of the challenges that lay ahead. It wasn't until approaching his 2-month doctor's visit that problems started to become more obvious. Xavier was having episodes of arching his back and often had a posture of tipping his head backwards. He had developed increasing difficulty breathing (stridor) which was especially hard on him when he was crying. He seemed to also be having some difficulty feeding. At his 2-month appointment when the doctor discovered Xavier's weight was very low for his age, he recommended we see several specialists.

The ENT doctor told us that Xavier had laryngomalacia, a condition in which the cartilage in the throat is delayed in developing, causing his difficulty breathing. We were told that he would grow out of it, it might just take

a few years. In the meantime, we were told that his weight problems were likely a result of the increased caloric needs from this condition.

Over the following month, we worked closely with a pediatrician to monitor and supplement Xavier's caloric intake. Despite all our efforts, we could not get Xavier's weight up. Two days after Analisa's birthday in 2014, we took him to the hospital. While we thought we were just going in to get help with feeding and then going home, we would not leave the hospital again for the next 3 months, and Xavier would never again live at home.

As it happened, two days after his admission into the hospital, Xavier went into respiratory distress, and then moments after being transferred to the Pediatric ICU, stopped breathing. Doctors who were standing by sprung into action and he was intubated, but the roller coaster had begun. When the doctors took a look, they found that Xavier's vocal cords were closing instead of opening when he breathed in—the physicians had never seen anything like it in a baby, and nobody knew what to do.

Over the coming months Xavier was given a feeding tube and was repeatedly intubated, and had multiple respiratory arrests. In the PICU we were constantly fighting for any semblance of normal amidst the beeping, the constantly changing nursing staff, and the doctors coming through to re-examine Xavier with little in the way of answers. There were also a number of medical scares, and as anyone who has spent time in the ICU might tell you, we were constantly on alert. While being in the PICU was a very difficult experience, we did our best to support Xavier, hold him, and give him all our love amidst all that was happening. He was also never alone--we lived in the hospital, taking turns sleeping in a pullout couch upstairs in the hospital. One of us was always with Xavier, so our family pitched in to help us get food and sometimes sleep.

Despite daily exams, a litany of tests, and many consultations, the doctors at Lucille Packard Children's Hospital (Stanford) were unable to determine a diagnosis for Xavier. Because he was still having difficulty with his breathing, the doctors did suggest that the only way Xavier would be able to leave the hospital was to give him a tracheostomy. This would ensure his ability to breathe. It was a huge decision for us as it meant we would no longer hear Xavier's voice and learning to speak would be delayed--but ultimately we decided it was our only option.

When Xavier was discharged from the hospital in April, he was 6 months old. By this time, he had started having seizures and muscle jerks, and he failed his hearing test just before leaving the PICU. Because he had also failed a swallow test, he had to be fed his milk through a feeding tube to avoid the risk of aspiration. He also had an enlarged spleen, and odd eye movements. But still, he had no diagnosis.

Upon discharge that spring, Xavier was sent to a pediatric subacute care facility, the Children's Recovery Center (CRC), which specializes in babies and children with tracheostomies. Here, we were to learn how to care for Xavier's tracheostomy—a delicate task, especially with a baby. The idea was that once we were trained in his trach care, we could then go home with some support from home nurses.

Being finally out of the hospital, and out of the immediate threat of Xavier not being able to breathe, we focused our time on determining a diagnosis. With the assistance of a local neurologist who helped us to control Xavier's seizures, Xavier had specific tests done for Neiman Pick and Gaucher. The test came back positive for Gaucher Type 2, a rare genetic, fast progressing, lethal disease.

While the diagnosis finally gave us some answers, it was devastating. We were told there was nothing that we could do and that the average baby with Gaucher disease lives 9 months to a year, without interventions, but that Xavier could live a bit longer because he had a trach. We were told to take a lot of pictures, and that our time with Xavier would be short.

After learning Xavier had Gaucher, we delved into finding out as much as we could about the disease, researching and speaking with the experts in the field. None of Xavier's local doctors had ever seen a baby with Gaucher 2. We spoke with doctors at the

NIH, as well as in Texas, Virginia and Boston who had seen more cases and had a better understanding of what we could expect during the progression of the disease, which causes neurodegeneration of certain parts of the brain. We also spoke with researchers in Israel, Japan and England, who were looking for a cure. Unfortunately, there was nothing available yet to treat Gaucher 2.

We soon realized that we had no options to cure Xavier—we would need to focus on helping Xavier's life to be the best it could be while he was with us. We made a conscious choice to spend as much time with Xavier as we could,

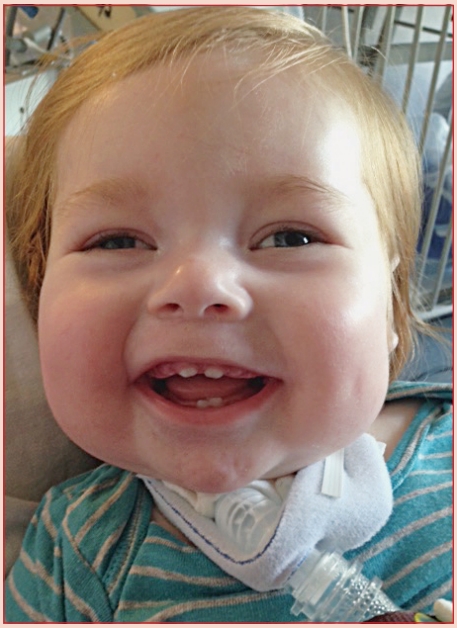
and to give Xavier the best life we possibly could give him. Family and friends supported this effort, visiting Xavier often and holding him as much as possible. We spent every day, all day with him, for the rest of his life.

Similar to the hospital, Xavier had his good days and his bad days as the disease progressed. The staff at the CRC was absolutely incredible and became like family. They loved him, and his special friends included some of the CNAs who knew just how to make him smile, and his occupational therapist, who worked closely with him and had a special relationship with Xavier. Xavier loved his bath-time and would often giggle as he got wet. He slept soundly each night with his stuffed animals arranged just so. He was such a smart and affectionate boy, and even as the Gaucher disease slowly decreased his ability to move his body, his face would light up in great big smiles – when he was held, when he heard his mom's voice, when he was sung to and cuddled and spoken softly to by his grandparents, aunts, uncles and friends, when Rinpoche and Christina came to see him, when he watched Giants ballgames with his dad (he was a natural Giant's fan with his orange hair!), and when he was enjoying the sunshine in the garden with his mom. His hair had grown from copper color, to bright orange, and he had beautiful blue eyes and the softest skin. Many people commented on how nice it was to sit and hold Xavier, and he would often put them to sleep cuddling!

We also had two special celebrations for Xavier – for his baptism and his birthday - where the whole family flew in to celebrate him. We gathered in the backyard at the CRC which had a beautiful grass lawn shaded by a tree with delicate blossoms. Everyone wanted to hold Xavier with his bright orange hair, and Xavier loved the energy and all the attention.

Summer with Xavier was a gift. While there were intermittent setbacks – generally he was doing pretty well. In between gatherings, we still worked towards a goal of going home with Xavier. However, as time went on and his condition progressed it became clear that going home would be very difficult. We needed the regular support of the nurses and the doctor at the CRC to help care for Xavier's needs.

As fall turned to winter, Xavier's condition started to worsen. He was sleeping more often, and smiling less and less. He became less and less able to move his hands, or sit up. He started needing ventilator support more of the time as his increasing seizures and myoclonus made it difficult for him to breathe sufficiently. He was having more and more difficult days. We worked with the CRC to obtain the support of a palliative care/hospice organization, who started coming to see us and Xavier several times a week.



In looking back, having the support of palliative care during that time felt essential, as there were many questions, and a lot of confusion over what would happen, what we could expect, the different choices we faced, and how we could make the best decisions for Xavier.

In March 2015, Xavier passed peacefully in our arms. His whole family was there, and he was surrounded by love.

Xavier's death was in some ways like his birth. It was extraordinarily peaceful. And he came into, and left this world on his own schedule, in his own way. He has left a huge and lasting imprint on the hearts of those who knew him, and the ripple effects of his life continues to manifest daily in our lives. He is so deeply missed. He will live strongly in our hearts forever. ■

Geoffrey and Analisa Perusse
Santa Cruz, California

Neurological Gaucher disease From Mouse to Human

By Raphael Schiffmann M.D., M.H.Sc.

Raphael Schiffmann M.D.,M.H.Sc.
Baylor Research Institute
Chairman – CGRF Scientific Advisory Board

Funding provided by the CGRF

Our understanding of the neurological aspects of Gaucher disease has progressed greatly thanks to the work of Weizmann Institute researcher Professor Tony Futerman. This work was made possible largely thanks to CGRF funding. Professor Futerman's laboratory discovered fundamental disease mechanisms that cause brain cell death and inflammation in mice affected by Gaucher disease. However, we do not know whether the findings in the mouse model for Gaucher disease also apply to the human patient.

Testing in Human Brain Cells

Thanks to recent scientific developments, it is possible to "make" human brain cells from skin cells (also called fibroblasts) of patients affected by this disease. The first step is usually to produce stem cells from the patient's skin cells. These are called "induced pluripotent stem cells" or iPSC. This method allows creating and then testing mechanisms and therapies in the patient's own nerve cells without being invasive.

Professor Ricardo A. Feldman

Late last year the CGRF funded a study, "A novel iPSC-based system to elucidate the molecular mechanisms that cause neuronopathic Gaucher disease", by Professor Ricardo A. Feldman of the University of Maryland. Using a specialized cell culture method Professor Feldman's laboratory will investigate whether the patients' inflammatory cells release molecules that harm the nerve cells. His laboratory will also study the mechanism by which Gaucher patient brain cells die. Professor Feldman's laboratory will use these findings in the effort to develop new therapies and protective measures for neuronopathic Gaucher disease. ■

3rd Annual Chasen'Tailz

That's a Wrap! 3rd Annual Chasen'Tailz Grew by 110% Making it one of the largest KDW's in The South East and Donating $29,000 to the Children's Gaucher Research Fund. To date we have donated almost $70,000 in the last 3 years!

We knew this year was going to be special and coined #BESTYEARYET but our expectations were blown away. We had 116 boats, 90 sponsors and almost 500 Anglers support us on our journey to help sick children in memory of our son Chase. The venue and weather couldn't have been any better. Harbourside Place offers plenty of dock space & the amphitheater served as the weigh station and awards ceremony. We had some of the best teams in the south east fishing from North Carolina to Islamorada. All on the hunt for the monster fish to win them $5000 and crowned Chasen'Tailz Tournament Winner. Our tournament has become one of the most competitive yet if you ask any of the teams they will tell you it's the most fun too.

Tournament day started with a festival of 35 vendors, steel drum band, vintage car show, water slide & dunk tank. Families enjoyed the festivities while cheering on our teams as they weighed in. Almost 60 boats weighed in some of the largest fish caught in this year's tournament series. Fish that would have won other tournaments didn't place in ours. The tournament winner was a 49.2lb Kingfish caught by team Holdem Hook on a 23 Contender single engine, proving its anyone's game.

We couldn't be more proud of our tournament this year. We worked for 10 months with the most dedicated committee and amazing sponsors. We miss Chase more than words could ever describe but we are thankful that his legacy and short life is now helping so many children and families. ■

Parents to an Angel,
Summer & Jay Warren
Jupiter, Florida

In May we flew to Washington DC to meet with the founders of The Children's Gaucher Research Fund, leading scientist working on a treatment for Gaucher and other families who tragically have lost their child as well. It was inspirational and difficult all at once. We learned where our funds were being allocated and how advancements were being made to have a treatment in the future.

ONLINE DONATIONS

can be made by visiting
www.childrensgaucher.org
OR
www.cgrf.org
All family stories can be read on the web site.

100% TO RESEARCH

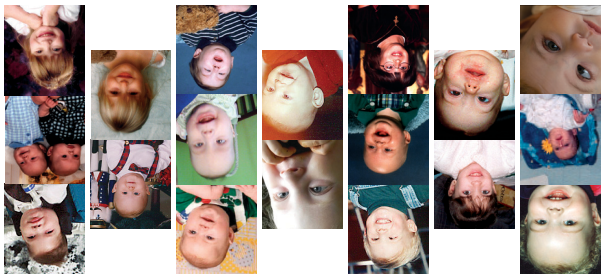
You need to know:

1. The CCRF is a *legitimate* IRS approved 501 c3 non-profit organization.
2. *100% of every donation* goes to medical research.
3. We *do not* hire professional fundraising companies who keep 50% of donated funds.
4. We have talented volunteers who *donate* their time and talent for a variety of our needs.
5. All administrative costs are paid for by the *founders.*

Simply put: if you send your hard earned dollars - *It ALL goes to medical research.*

Visit our web site at:
www.childrensgaucher.org
All family stories can be
read on the website.

Contributions Payable To:
Children's Gaucher Research Fund
P.O. Box 2123
Granite Bay, California 95746-2123



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