Individually *Rare*, Collectively *United*

By Kristi Wees, MS Chem

Would you consider something that impacts one-in-ten Americans a RARE occurrence? Can you imagine if lightning struck one-in-ten of us, that we would still consider this a 'rare' occurrence in nature? Probably not, but that is the number of Americans who have a rare disease and strikingly, over half of them are children. In and around Pittsburgh many families are impacted by rare disease, and while their journeys and diagnoses are individually rare, collectively in our community, you may consider them quite common. It is in this commonality of having a child with a rare disease, that many parents find support in their disease community as well as their local community.

According to NORD (National Organization for Rare Disease), any disease, disorder or condition that affects fewer than 200,000 people in the USA is considered RARE by the National Institutes of Health (NIH), the U.S. Food and Drug Administration (FDA) and NORD. We are honored to share with you the journey of three families who live in the Pittsburgh area and are living a life of rare disease with their children.

The *Hackwelder's*

Mom, Tara, and dad, Jason Hackwelder of Chicora have had a rollercoaster of a journey with their daughter, Elizabeth, age five, and her rare disease, ZTTK Syndrome. Elizabeth was diagnosed in 2015 with ZTTK and was one of the first six in the world identified with this genetic syndrome. Elizabeth's family asked to be put into contact with other families who were identified with this same syndrome and these families started an online parent group on Facebook, which now has close to 50 children identified throughout the world.

As you can imagine, a rare disease's impact on a child actually impacts the entire family. When asked about how Elizabeth's condition has impacted their family, Tara's response was so honest, transparent and heartfelt, there is no need to paraphrase:

"We refer to our journey with rare disease as a wicked combination of heartache and blessings. That is truly the best way to describe it. The heartache of rare disease is knowing that your days are numbered with your child. It's not knowing what your day-to-day will bring, and if you will make it through the day without having a medical emergency of some type. It requires our family to spend many days separated from each other during hospital stays. It is constantly making phone calls to doctors, the school, the insurance companies, medical supply companies, nursing agencies and pharmacies to ensure that your child is getting everything that they need to live their best life. It is the terror of watching your child have close calls with death over and over again, when you are literally helpless, and all you can do in those moments is hold your child, call their name, and demand that they stay with you, all while praying that the doctors and medical professionals working so diligently to reverse their critical condition are able to actually do so."

> "The silver lining is that when you find your rare disease community, or you are active within the community at large, you know that you are never alone. It is a comradery that cannot be found anywhere else."
>
> – *Tara Hackwelder*

Many individuals who are not impacted by rare disease may think that the impact of having a child with a rare condition may solely revolve around that child's health, medical needs and hospitalizations as Tara described above, but the impact can shake a family much deeper.

Tara describes the financial and social impacts, "It is the financial destruction of a family that faces rare disease to cover the costs of things that insurance doesn't cover and days of employment lost due to hospital stays and appointments or even having to quit working to stay home and care for your child. Even though being unemployed to care for your child is financially damaging, it is often what needs to happen, and this is what happened in our journey with Elizabeth. I resigned from my full-time teaching position to stay at home and become a full-time mom to our children, as well as a nurse and advocate for our daughter. I don't regret that decision, and I have treasured my time home with my children, but it did dig us a financial hole that we may never crawl out of."

Long-term financial plans like retirement are also impacted says Tara, "If the doctors are wrong, and you hope that they

are, your child will need care for the rest of their long life. Even if you are able to save money, you probably will not save enough to both retire and provide adequate care for your medically-complex child."

With all the hardships that a family experiences, though, many families like the Hackenwelder's have been able to see the bright spots and blessings in their daughter's diagnosis.

Tara shares her words of wisdom for others who may be experiencing rare disease: "Rare disease can be a heavy thing to experience. The key to a good life, and especially a life with a rare disease journey, is to always look for your blessings. You will find what you seek. If you are always focusing on the negatives and the burdens, then that is what will govern your perspective. However, if you focus on the blessings, then you will find them, and you will begin to see them without effort. When your blessings outweigh your burdens, your journey becomes a little bit lighter and brighter. Let the love, not the darkness, guide you along your way."

To learn more about Elizabeth Hackwelder's journey, her blog is teamelizabethjeanne.blogspot.com

Some Pittsburgh (and beyond) - *Resources for rare disease families*

- Center for Rare Disease Therapy - Children's Hospital of Pittsburgh • chp.edu/our-services/rare-disease-therapy
- National Organization for Rare Disease (NORD • rarediseases.org
- Global Genes • globalgenes.org
- Syndromes Without A Name (SWAN) • swanusa.org
- NYMAC - Find a genetics clinic near you • NYMACgenetics.org • NYMAC@ferre.org
- Genetics and Rare Disease Information Center: 888-205-2311 • rarediseases.info.nih.gov/about-gard/pages/23/about-gard
- Reference for facts and figures on rare disease • cdn.rarediseases.org/wordpresscontent/wp-content/uploads/2014/11/NRD-1008-FactSheet_5.pdf

continued on page 10

The Conley's

> "We quickly became part of the special needs community in our area, but could never find anyone very much like Pierce. We would always wonder what the future held for our son."
> *- Audrey Conley*

When Audrey Conley of Upper St. Clair recalls back eleven years ago, she remembers that they had no warning signs that there would be anything wrong with their newborn son, Pierce. She recalls how she and her husband, Ted, were quickly thrust into a world of testing at UMPC Children's Hospital of Pittsburgh and she remembers being so overwhelmed by all the extra medical attention their son needed that she did not have time to really comprehend what was going on. Audrey says, "We quickly learned to trust the doctors and listen intently to every word they would say, then we would lay awake at night trying to make sense of it all."

Audrey recalls the first five years of her son's life as a blur. The world of rare disease is full of appointments, therapies, evaluations along with all the other things that go along with childhood. For Pierce, the typical childhood illness or cold would always put him in the hospital, usually for a few days. In fact, for ten years the Conley's did not know what was causing Pierce's challenges, they were living in the unknown. During that time, they came to accept the differences, the oddities and the uncertainties of the unknown condition.

This past May, through genetic testing called whole exome sequencing, the family received a call that Audrey describes as: "A call that actually has given us peace and a new sense of hope!" Pierce had KATA6 -a mutated gene that had only been recently discovered in 2015.

Audrey remembers that day a few months ago and describes it as one of the highlights of their journey with their son. She recalls, "Within an hour of the diagnosis I was requesting to be part of a closed social media parent group for KAT6A parents. At this point, there had only been 123, now 124, children in the world with this diagnosis! They had also already started a website where I saw children so very similar to our son, developmental disabilities, spasticity, most were non-verbal, feeding issues and so on. I was immediately welcomed and even met a mother that had a son Pierce's age. We felt welcomed and relieved. Over the next several weeks I read and commented on so many similarities, as well as gave advice!"

With the highs come some lows as Audrey shares one of the toughest parts of the rare disease journey, "Our group desperately needs to raise money for on-going research to help the quality of our children's lives. Just another thing special needs parents should not have to worry about." But even on their worst days when the Conley's may be weary from all the care their son needs: feeding, toileting, therapies, the constant attention, they remind themselves, "This was NOT our plan but God's. There is a beautiful purpose for our child. The medical community needs our RARE children, our RARE children teach, encourage and spur them on for answers," says Audrey.

To learn more about the rare disease that impacts Pierce you can visit this website: kat6a.org or Facebook: KAT6A Support Group, KAT6A Foundation.

In(cluded) Community

When parents of children with a rare disease/special needs were asked on Facebook:

"What is your biggest piece of advice for your community, city, township, etc. on how they can be more supportive, inclusive, helpful or sensitive to your child and your family's needs?"

These are their responses:

- I would encourage others to ask questions instead of staring or pointing. I'm always happy to answer questions about our daughter. I would much rather you ask, than point or feel awkward around us.
- Don't talk in front of our kids like they do not understand. They understand and remember everything. (Presume competence)
- Educate the police about how to recognize people with special needs.
- Have public bathrooms that allows for changing adult sized children's diapers.
- Quit having every event revolve around food. Even crafts for kids have food ingredients. It would be so nice to have non-food-based family events.
- The community needs to select after school care programs at school where kids with special needs can be included. Many are not welcoming or equipped to provide care for kids with special needs. Many families have only one parent working for this reason.
- Lower stimulation (sound, lights, commotion) areas within public areas.

continued on page 12

INDIVIDUALLY RARE *continued from page 11*

- Have more opportunities for vocational/supported work
- Be mindful of germs and sickness. Stay home if you're sick. Cover your cough. Wash your hands. Things like a simple cold can be so much more serious to kids like my son. A simple cold or fever could mean 48 hours in the hospital.
- Just because a child "looks ok" doesn't mean they are ok. Parents know their child best. If they tell you something has drastically changed, believe them.
- Talk to my child - not me. (When asking about them.)
- If you say you are handicap accessible, you should have automatic doors. It's so hard getting into places with super heavy doors trying to push a large child through with a wheelchair.
- Do unto others as you would have them do unto you.
- Don't count me out, keep asking even if I say no a lot.
- We've always desired empathy and compassion over pity or charity. Our children desire to belong as any other child.
- Take time to notice the little things in your business, such as flickering lights that can be very harmful for a child with epilepsy or others with sensory issues.
- More handicapped parking spaces.
- Special needs shopping carts at the grocery store - like Caroline's Cart.

The *Clegg's*

Jillian and Brian Clegg, of Bel Vernon, gave birth to their first child, Tyler, in 2002. As first-time parents, they began to recognize that he was delayed when he was about five-months-old. Devastated, they began seeking doctors in search of what was wrong with their baby boy. He did not roll, have purposeful hand or eye movements, he screamed almost continuously, slobbered a lot and just looked sick to his concerned parents. They eventually began treatment with neurologists and had numerous tests completed. The end result was being told, "Well, we will have to wait and see". But, the Clegg's not being the wait-and-see type of parents, jumped into finding out what they could do for their son. In 2004, they welcomed a "healthy" baby boy named Noah. They delighted in seeing him develop normally and described it as miraculous.

In May of 2008, Jillian was finishing her last semester of nursing school, caring for her father who was very sick, getting ready to deliver twins, and caring for Tyler, six (complete care) and Noah, four. In July, Jillian delivered the twins ten days following her father's passing. Luke and Aiden. Two beautiful babies! Jillian knew instantly that Luke had the condition, the same condition as his brother Tyler. Everyone told her, "You are just worried, he's fine," but she knew in her heart he was impacted.

As Luke began progressing the same as Tyler had, the family began seeing doctors again, never any concrete answers but now knowing that it had to be genetic. A relative told them of a program at the National Institute of Health called the Undiagnosed Diseases Program. They applied with their neurologist's help and were accepted. They traveled there in June 2010 and spent one week packed full of multiple tests and appointments. The family was told that there was minimal chance of finding a diagnosis but that they would try and keep them updated.

Finally, in May of 2013, the Clegg's got the call they had been waiting nearly ten years for. Tyler and Luke were diagnosed with a rare gene defect, with only two confirmed cases worldwide. The genetic condition known as Med23 (the gene) causes intellectual disability in an autosomal recessive fashion which means both parents carry some type of mutation on the same gene and any child has a 25 percent chance of inheriting the disease.

Tyler and Luke are impacted by Med23 in that they do not speak, walk or use arms in purposeful manner. They are severely delayed and require assistance with all activities of daily living with 24-hour care. They have very high muscle tone in their arms and legs. Along with many sensory issues. But with all their daily challenges, they are a joy and a blessing to their family.

Jillian shares the bright spot in their journey has been the people: "Meeting other people who share in an amazing yet difficult journey. I wouldn't have half of the people that I have in my life without my boys. Good and kind-hearted people that like me get to share in the purest and deepest love that there is. There is a special beauty behind the eyes of a kiddo with special needs that many never get to experience."

> "Seek out others. Don't let this journey be a one-lane road. Drive next to others and when you are in need of repairs or a break, hitch a ride with someone else. Let others build you up, educate you, teach you and support you. Those people are hard to find and not worth losing!"
>
> – *Jillian Clegg*

As a mother, nurse and advocate, Jillian's words of encouragement for others on the rare disease journey are inspiring and heartfelt: "Life is not about waiting for the storm to pass, it's about learning to dance in the rain. The hard times are there. Some days better but for the most part, it's a tough life. Enjoy the little things, take time for yourself and continue to give even when it's a struggle! We all carry different crosses... just different sizes."

To learn more about the Clegg's journey you can visit their webpage: hope4tylerandluke.com and their Facebook page: hope4tylerandluke

With over 7,000 rare diseases identified, one would think there must be thousands of treatments for all these rare conditions. However, the sad reality is there are less than 500 rare diseases that have FDA approved treatments, less than five percent of them! With this startling reality, many families, like these three from Pittsburgh, find themselves connecting with other families, sometimes across the globe, to help navigate to hope for their children.

Are you a parent of a child with a rare disease? Do you have a child with an unknown condition that baffles doctors? Are you concerned about your child's development?

There are many resources for families and physicians alike, some close to home and other national organizations, as well. As we have learned from these families, once you have a name, a gene or a diagnosis, it is easier to connect with parents whose child may be similar to your own.

Even if your family is not directly impacted by a rare condition, we all live IN COMMUNITY with families who are on a rare disease journey with their child. We all can take part in making these families lives a bit brighter by being good neighbors. Please see the In(cluded) Community list for some ideas from parents of children with rare disease and special needs, to get you started! ■

Kristi Wees is the Chief Advocacy Officer at www.EmpoweredAdvocacy.com. She advocates along-side families each and every day for the best quality of life for their children, specializing in rare diseases such as mitochondrial and metabolic conditions to the not so rare conditions such as Autism, ADHD, PANDAS, and developmental delays.