Lexi Fae's Story

March 2019

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Our daughter Lexi Fae (3) was born with rare genetic disorder called BPAN. There are only 100 children diagnosed with this disorder in the world. There is no medication and no clinical trials in progress to date. Without any advancement in modern medicine, Lexi will lose all abilities to walk, talk, eat and function in daily living. Your donations will fund scientists to continue research and hopefully have a remedy for better outlook for the future. Thank you in advance for your support.

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What is BPAN?

BPAN stands for Beta-Propeller Protein-Associated Neurodegeneration. BPAN is caused by mutations in the gene WDR45 which is located on the X chromosome. The majority of the people who have BPAN are females - the mutation is lethal for most males before birth, however, there are a few males living with BPAN. People with BPAN are developmentally delayed with slow motor and cognitive gains. Typically they are also non-verbal and develop little to no words. Seizures and sleep disorders are typically common with the disorder. During adolescence or adulthood, affected individuals experience a relatively sudden onset of progressive dystonia-parkinsonism and cognitive decline and they cannot regain skills once they have lost them. The average life span varies for individuals with BPAN, but due to improvements in medical care, more affected individuals are living well into middle age.

The future for those diagnosed with BPAN.....

This disorder was only discovered in 2012, and currently, there is no treatment for this disorder. There are doctors and researchers that are currently trying to understand BPAN and they need the money to keep funding their research in hopes to find a treatment or even a cure for this disease. They rely on donations from organizations, corporations, and fundraisers to keep their research going because they do not receive any US Government funding. This is why we want to raise money for BPAN Research.

Let's start from the beginning……

Lexi Fae was born on March 8 2016. She was a healthy beautiful baby with a head full of hair, and no complications. Lexi was very cute and sweet from the moment she was born. Her first few months of life were typical however she was unusually quiet for a newborn. Most mothers would be thrilled that their baby was barely crying or fussing in the middle of the night. At 7 months, I recall waking up my husband Tommy at 3AM in the morning to tell him I just know something is not quite right with Lexi. Sadly, my intuition was right. Due to the lack of motor skill development and hitting typical milestones, we had her evaluated by numerous doctors and early intervention team. Lexi qualified for Early Intervention. It was a bittersweet moment. We were happy Lexi was getting support from many therapists but our gut instinct told us this is only the beginning. The revolving door of Physical Therapy, Speech Therapy, Occupational Therapy, ABA Therapy, and Music Therapy continued until Lexi was 3 years. All of these therapy sessions were good for Lexi. These therapists are moms and sisters and aunts. They genuinely cared about our little girl and wanted to see her succeed. In those first 3 years, Lexi was able to finally sit and stand at 18 months and take her first steps at 25 months. My favorite, treasured moment was Lexi's first smile at 9 months! That memory still warms my heart!! No first words yet but I know they will come eventually.

During all of these therapy sessions and regular life activities, we had doctor upon doctor visits, hospital stays, blood tests, Seizure evaluations, basic genetic testing and the list goes on. This is a lot for an adult let alone a 3 year old. Lexi handled everything like a champ. Majority of the test results came back normal yet inclusive for explaining Lexi's delays.

The one test that took our insurance many months to approve was whole exome sequencing (WES). This test cost more than $20,000 but we felt it was necessary and fought hard to have this test approved. The test was done Dec 2018 and in March 2019, we received a call that the results were in. Tommy and I were not prepared at all for what we heard. Our flippant attitude going into this appointment was because we were so conditioned by all of the other doctor's appointments in the first 3 years of hearing the medical community could not explain why Lexi is the way she was.

On March 18, 2019, our lives turned upside down. The genetic testing revealed that Lexi had an ultra-rare genetic disorder. It was a mutation on gene WDR45, also known as BPAN. My head was spinning and the walls were closing in as the doctor was speaking. I experienced my first ever anxiety attack. Thank goodness, Tommy was able to stay calm and gather the important details of the condition. The good news if there is such a thing at this appointment was that this

gene mutation is not hereditary and our 5 year old son would not be affected. The bad news and there is lots of that…… is that this is a neuro degenerative disease which will limit Lexi in many ways. She will lose ability to walk, communicate, eat and have Parkinson like effects as she grows older. There is no cure and no clinical trials. Lastly there are only 100 or so children in the world that have this rare disorder.

Even though I am surrounded by such a great family and friends I still could not function and spent 2+ days crying….. the tears would just not stop. My family reminded me that Lexi is still the same beautiful, sweet girl that has the best curls and longest eye lashes ever! Hearing Lukas say to Lexi, "stop giving me hugs and kisses, I am trying to watch my show!"…….. that is when I realized everything is still the same. This diagnosis does not change the fact that we are blessed.

What's happening today.... In April 2019, Lexi started a full day special needs school program She seems to be liking it and I am adjusting to the new routine. She now gets her occupational therapy, speech therapy, and physical therapy all at school. She is currently learning how to use a device to ask for things. She still loves to give out free hugs and kisses to anyone that wants a wet one!

In the area of research and clinical trials, there is not much happening today. This disorder is so rare that any large pharmaceutical will not invest the time and money to save these 100+ kids. The tenacity and drive of friends and family is the main method to create awareness and raise money for individuals with BPAN. I firmly believe together we can raise awareness and money to make a difference and improve the quality of life for these children. We cannot stay idle and do nothing! Please come together and help make a change.

Please click on below link to make donation. Please also forward to your friends, family members, communities, etc.

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