

Peter Najm & KAT6A disorder; Living With a Little Known Genetic Mutation  
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Peter Najm is one out of only 311 people around the globe who were born with the rare genetic mutation more commonly referred to as KAT6A disorder. While he may appear to be a 12 year old, nonverbal boy on the outside, there is simply so much more to Peter.

What is KAT6A?

It is a gene mutation of the KAT6A protein in chromosome 8, which is a major protein involved in the packaging of DNA molecules.

So how does it work?

The KAT6A gene makes the KAT6A protein, which is involved in controlling the production of proteins from other genes. Therefore, when there is a change on the KAT6A gene, problems can occur in various parts of the body. Currently, scientists do not know all of the functions of the KAT6A gene, so our knowledge will increase as research advances (KAT6A Foundation, 2021). However, what we do know is that when a person has a mutated KAT6A protein, all of the cells in their body will be affected.

Interestingly enough, most of the time that a child is born with this mutation, their parents did not have or carry the gene, labelling it a ‘de novo’ gene mutation, as opposed to a typical genetic disorder or something that is hereditary and has been passed down. However, if a person does have the KAT6A mutation, there is a 50% chance that they will then pass this gene along to their children, as it is autosomal dominant.

How rare is rare?

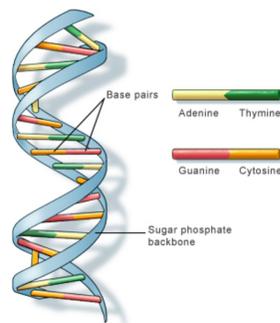
As of July 24th, in 2019, there were only 195 patients in the world who had been diagnosed with KAT6A, but as of February 23rd, 2021, there were 308 known cases, and the number is slowly increasing everyday. As of May 10th, 2021 there are 311 known cases. Doctors and researchers agree that as the awareness of this disease increases, the prevalence will increase as well. The prevalence of something is the proportion of a population who exhibit a certain characteristic, in any specific time period. One reason that there may be so few diagnoses of KAT6A mutation in the world is because in order to diagnose, whole exome sequencing must be done.

So let's break this all down.

It all begins within a microscopic structure inside of all of us, called DNA. It's more formal name is deoxyribonucleic acid, and it makes up all of the hereditary and genetic information in humans and just about all other living organisms. Some basic things about DNA to know, are that just about every single cell in our human bodies holds the same DNA sequence, no matter where the cell is in the body, or what its job is. Most of this DNA is housed in the nucleus of the cell, where the commands are made, and information is sent out from. The information that DNA carries is stored in the form of a code, made up of 4 unique chemical bases; Adenine (A), Guanine (G), Cytosine ( C ), and Thymine (T). The corresponding letters next to each base are the symbols that will be used for the bases when they are written or discussed later on.

Next, it is important to understand that the order or sequence that the bases will create when joined together determines what information the cells will have available for helping to build and maintain an organism. This can be compared to how when we arrange certain letters of the alphabet in different ways we can form words with them. When A+T are together, one protein is made that differs from the protein that is made when C+G come together. However, the bases A,T, G, and C, come together in a special way, as bonded pairs, and can not be rearranged in just any random order. Each of the four bases come together and pair up with their preassigned and corresponding partner, every time. A with T, and C with G. Once joined, the units they form are called 'base pairs'. Each base in the pair also attaches to both a sugar molecule as well as a phosphate molecule, to create what is known as a nucleotide. The nucleotides are then arranged into long strands that get twisted together and form the spiral double helix that we can all picture as DNA.

DNA is a double helix formed by base pairs attached to a sugar-phosphate backbone.



U.S. National Library of Medicine

Credit: U.S. National Library of Medicine

According to the US National Library of Medicine, DNA sequencing, or determining the order of every individual building block of a person's genetic code has greatly advanced the study of genetics ever since it was first discovered in the 1970s. While we were not yet able to read or sequence the DNA upon the earliest discovery of its three dimensional form, the conceptual framework for DNA replication and the encoding of proteins in nucleic acids began to develop in the coming years after.

“The order of nucleic acids in polynucleotide chains ultimately contains the information for the hereditary and biochemical properties of terrestrial life. Therefore the ability to measure or infer such sequences is imperative to biological research” (Chain & Heather, 2016).

With this information noted, scientists and researchers agree that determining the order of the nucleic acid residues in biological samples is an integral component of a wide variety of research applications. That doesn't mean that this is any easy task by any means.

Two methods that have advanced the study of genetics, and are “increasingly used in healthcare and research to identify genetic variations” include whole exome sequencing, and whole genome sequencing (U.S. NLM, 2020). Both of these methods rely on new technology that is able to rapidly sequence large amounts of DNA, or genetic material. Because of this reliance on technology and machinery, these methods are often referred to as Next Generation Sequencing.

Due to the speed and efficiency of these Next Generation resources, researchers are able to read and sequence larger amounts of individual's DNA in a shorter time frame, such as all of the parts of the DNA that provide instructions to the cells of the body for making proteins. The pieces of DNA that are grouped together and analyzed in this way are called exons, and are only thought to be a small fragment of an individual's entire genome, about 1 percent of it to be precise. All of the exons in a genome are referred to as the exome, and because of this, the method of sequencing them is referred to as whole exome sequencing.

Whole exome sequencing is how the KAT6A gene mutation can be discovered, because it allows for “variations in the protein-coding region of any gene to be identified, rather than in only a select few genes”. In addition, “because most known mutations that cause disease occur in exons, whole exome sequencing is thought to be an efficient method to identify possible disease-causing mutations” (U.S. NLM, 2020).

Dr. Natacha Esber M.D. who also doubles as Peter's mom, has put a tremendous amount of effort into funding research for this disorder, through her work with the KAT6A Foundation that she and her husband were founding members of in 2017. In an informational video now posted on YouTube, Dr. Esber explains that "the KAT6A diagnosis is only made with whole exome sequencing, which makes it hard to detect patients, because whole exome sequencing is not performed in all countries of the world yet".

So without reading the whole exome sequence, how would one know or be able to identify if a person had this disorder? What are some signs and symptoms of KAT6A? How are all of the cells in the body affected?

There are a wide range of features when it comes to this syndrome, because each person with KAT6A has a different mutation along the KAT6A gene, which may show itself differently. Common symptoms are developmental delays and disabilities, speech delays, gastrointestinal abnormalities such as constipation or obstruction, heart problems, epilepsy, and sometimes malformations (Esber 2019). Other common traits are feeding difficulties, acid reflux, vision problems, hypotonia (decreased muscle tone), and intellectual disabilities. Bolded on the KAT6A organization's website though, is this phrase: "It is important to note that many parents describe their children as happy and healthy despite global developmental delays". Less commonly exhibited symptoms of KAT6A syndrome include seizure disorders, frequent infections, sleep disturbances, autism, abnormal muscle tone, sensory issues, skull abnormalities, and distinct facial features.

So what therapies are available to help treat children and adults diagnosed with KAT6A? Currently, the therapies used to treat and manage KAT6A disorder are the same therapies used to treat any developmental delay such as speech therapy, physical therapy, occupational therapy, psychology, ABA therapy, special education in school, and even horse therapy! (Esber, 2019).

Speech therapy, which is performed by speech language pathologists (or SLPs), is used to assess and treat communication problems and speech disorders. If a child with KAT6A is nonverbal or has other communication needs or difficulties, speech therapy will aim to provide the child with techniques that can be used to improve his or her communication abilities. It can also help people who have articulation disorders, fluency disorders, resonance disorders,

expressive and receptive language disorders, cognitive communication disorders, as well as diagnose and treat disorders like Aphasia and Dysarthria (Luo, 2019).

In addition to therapy for speech, physical therapy can help ease pain, and improve body functions that help people to move around (Brakeville, 2019). For Peter or another person with KAT6A, their muscles may be weaker than those of the typical person. Physical therapy can help to strengthen their muscles, work on balance in order to prevent a slip or fall, control one's bowels or bladder muscles to prevent accidents, provide rehab after a stroke or other type of accident, injury, or surgery, and can even help to teach a person to use an assistive device such as a walker, cane, or brace.

Occupational therapy (OT) also helps people gain the abilities needed to participate in things they want and need to do everyday. It is called occupational therapy, because sessions include the therapeutic use of everyday activities which are also referred to as occupations. It is very common for an occupational therapist to help a child with disabilities be able to participate in school activities or other social situations, by teaching them methods of doing such activities that work best for the children's specific needs.

ABA or Applied Behavior Analysis therapy is another therapy technique commonly used for people on the Autism spectrum. This therapy is heavily based on the science of learning, and on the study of human behavior. Its goal is to increase those behaviors that are positive, and decrease the behaviors that may be harmful, or negatively affect the child's learning. ABA therapy has been around for years, (since the 1970s to be exact, where it got its roots from Discrete Trial Training), and has been proven to increase language and communication skills, improve attention, focus, social skills, memory, and academics in people with certain disabilities while also decreasing the behaviors exhibited that are viewed as problematic by other child peers or caretaking adults. ABA therapy is based on Skinner's idea of operant conditioning, and uses positive reinforcements as one of its main strategies of teaching. This works, because when a behavior "is followed by something that is valued (such as a reward), a person is more likely to repeat that behavior" (Autism Speaks, 2021), which over time fosters a positive behavior change. Using positive reinforcements such as food or candy can be good motivators, but therapists must be careful when using reinforcements such as those that the child they are working with is actually learning what is trying to be taught to them, or if they are only focusing on the piece of candy that is dangling in front of them. When this is the case, the skill being taught by the

therapist is not being learned or committed to the child's memory, and will not be able to be generalized later by the child but rather the child is only paying attention to doing what they must do in that moment in order to get the treat. When this happens, it is detrimental to the child or client in question, because the therapy is not helping to teach the child anything, which should always be the main goal of the session.

Another type of therapy that has been proven to foster positive behavioral growth is horse therapy, or hippotherapy, derived from the greek word hippo for horse! Horses have been found to calm riders who have Autism as well as other sensory issues. While calm and on top of the horse, the rider can think, focus, and accept the training they are getting from their instructors on skills such as speech, socialization, and fitness. Alicia Kershaw, who is the founder and executive director of GallopNYC, a therapeutic horseback riding organization, explains that:

“the desire [of the child] to ride, also allows us to encourage positive behaviors and gently discourage negative behaviors. And, yes, one of our goals is ‘just pure fun.’ Every rider learns some measure of riding and horse skills, and some riders become quite accomplished” (Kershaw, 2013).

This idea is similar to the ideas behind ABA therapy, where positive reinforcements encourage change, only now, being with the horse and the action of riding the horse is the positive reinforcement and rewarding behavior that will stimulate the child to behave well and pay attention to what they are being taught. If the child enjoys riding the horse, they will most likely want to win the approval of their instructor or therapist, by following their directions. In turn, if the child is able to generalize the skills they have learned, they will be able to take the listening skills as well as other things they were taught while on the horse, and apply them to many other aspects of their life.

In addition to these therapy methods, there are also food and vitamin supplements that Dr. Natacha Esber along with many other researching physicians have found to be helpful in the children diagnosed with KAT6A, which are referred to as a Mitochondrial cocktail. “For patients with mitochondrial disease, a mitochondrial cocktail is a personalized mix of prescribed high-dose supplements. The combination and dose of these supplements are unique to each

patient and designed to improve the patient's disease manifestations and support the function of organs" (Chemistry Rx Pharmacy, 2021). Over the years of researching KAT6A, evidence has been found of some "mitochondrial dysfunction in our children", which is also found in many children and people with different types of developmental disabilities. Major improvements in mitochondrial function have been noted among many of the children who are taking these vitamins and supplements.

So how has research in the field of KAT6A been going?

In 2019, there was a basic research trial on *kat6a* happening at UCLA in California, and a research pilot study being done at John Hopkins in Baltimore, USA about developmental evaluations of kids with *Kat6a*, as well as another basic study at McGill in Montreal, Canada. In Melbourne, Australia there was another study, this time on sleep habits in mice with KAT6A which is especially interesting, because they have found that many children with the KAT6A disorder have issues with falling asleep, staying asleep, and waking up at night. Additionally, there was a speech study being done at the Murdoch Children's Research Institute in Australia for children affected with *kat6a* internationally, which anyone can join worldwide (Esber, 2019). Beginning in 2020 and currently ongoing, are studies of "precision personalized medicine in KAT6A syndrome" out of the Centro Andaluz de Biología del Desarrollo in Spain, and the "Chan Zuckerberg Initiative, USA" where, "the KAT6A Foundation was chosen as part of the Chan Zuckerberg Initiative: Rare as One Network" and, "a group of 30 patient-led organizations are accelerating research and driving progress in the fight against rare disease" (KAT6A Foundation, 2021).

## PART 2: THE INTERVIEW

So how did I meet Peter?

I had the pleasure of meeting Peter on a hot sunny day at the pool one summer, many years ago. I saw him there with his mom, and his younger brother Paul. They were swimming and splashing in the water at the local town pool when *my* mom and I approached to say hi! Shortly after meeting, it became apparent that we were both regulars, and would be seeing a lot more of each other over the course of this summer and many many summers to come. We were drawn to Peter's family, because they attracted a lot of attention!

Anyone who visited the pool while Peter was there may have heard his loud voice, or booming lip trill accompanied by a smile when he was in the water. They may have been inquisitive, and looked over at him when they'd see his arms and hands flap at the water's surface creating lots of splashing in an otherwise calm and quiet environment. People may have taken notice of the boy who would run around from pool to pool, and then race into the showers in the bathroom for quite some time.

Peter and his younger brother Paul would tell us (my mom and I) to watch them, as they would show off their diving skills in one pool, and their lap swimming skills in the other! They would sometimes play in the kiddie pool, but not for long, as there was so much more exploring to be done, and *nothing* was going to hold Peter back from exploring.

Peter warmed up to my mom and I pretty quickly. We spent hours out of our days that summer getting to know him and his family, and every time we talked with him and his mom, I was always fascinated with the takeaway. Not only did it excite me to learn about Peter, and gain experience from our time together, but talking to his mom Natacha really expanded my horizons about the world, and opened my eyes to many different career options I did not even know existed, including speech therapy and applied behavior analysis, during what I would consider to be the most influential time in my *own* life, the summer before college, before I would need to decide my passion, calling, and what I wanted to do career-wise for the rest of my life.

Fast forward a few summers, and Peter and his younger brother Paul weren't the only Najm kids in their family any more. Natacha gave birth to another baby boy named John, and then after that to a beautiful baby girl named Mary. Now with four kids in tow, she still always seemed to be doing the most, and taking them all everywhere. With little to no help, she somehow still had time to bring them to the town pool almost daily, while working an intense job, and balancing all of her household's needs. Shortly after meeting her, I realized that Natacha was somewhat of a supermom.

And you don't need to just take my word for that either. It is evident from the numerous photos and videos that Natacha has shared of her children on the internet on sites such as Facebook, that Peter and his siblings do it all. While only one out of her four children have severe disabilities due to KAT6A, discovering her son's diagnosis has never stopped Natacha from allowing Peter to have as "normal" a life as possible! Though sometimes he exhibits loud

stimming behaviors which can be accompanied by putting his fingers in his ears, his mom says that this is mostly because he just wants to be near the excitement, and he loves to be at the front of concerts and entertainment, but it can be really loud for him. Peter has very sensitive ears and can actually pick up on more noises than the typical person. Regardless, from the pool to the beach, from museums to amusement parks, and from skiing to sledding, Peter enjoys the same activities as most kids his age. He even enjoys horseback riding, and fishing! You can find Peter out to dinner with his family, taking trips to the beach in Rhode Island and in Jersey, playing musical instruments to keep busy, and acing his schoolwork too. Peter and his three younger siblings are even in a band together! This past year, they performed in a Christmas recital, where Peter switched it up and took a turn playing not just one, but multiple different instruments.

Peter is a loving and helpful boy, who is never afraid to get his hands dirty in the kitchen, or play outside in the snow or mud! He can be found doing quite literally any activity or task, including painting and making art, swimming in their new backyard pool, bike riding down the block for ice cream and doing just about anything with his siblings, Paul, John, and Mary. Just about anything, including attending Arabic parties, where Peter enjoys listening to the live music and orchestra play!

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Peter's mom, Natacha Esber is a doctor of internal medicine, an area of primary care for adults. She specializes in diagnosing and treating cross-system illnesses that may affect multiple organ systems. She mostly works graveyard shifts at her hospital, which affords her that time during the day to really enrich her children's lives.

We sat down together and she was able to give me way more insight about Peter, his diagnosis, and his life story from birth until the present.

So what is Peter's story?

When and how did you first find out that Peter was different?

Natacha says that the first time she suspected something was different with Peter was when he was about 15 months old. Natacha believes that now, after having parental experience, she would have been able to detect a difference in her baby (or any baby) at a way earlier age,

but at the time Peter was her first child, so she had no prior experience, and limited references of development to compare him to.

Some of the things that led Natacha to believe that her first baby may be different, were the facts that he was barely making eye contact with her and his father, he was not walking, and he was not able to eat solid foods yet. He was only able to consume baby food, and would take his bottle, but he was unable to eat even a mashed up banana, as he would have trouble swallowing it, and would oftentimes choke on it if she tried to feed it to him.

What were the first steps in assessing what may have been wrong?

Natacha first took Peter to the pediatrician. She says that even upon examining Peter, the pediatrician was not too concerned at the time with the delays in his development. It was not until Natacha persisted that something was wrong, that the pediatrician referred her to a neurologist.

After undergoing the neurological testing, Peter was diagnosed with *something*. At now 18 months old, he had a diagnosis of Severe Autism Advanced, or Global Developmental delay. It is important to remember that at this point it was 2010, and there was no known disorder called KAT6A gene mutation that existed. Now, there is so much more knowledge of this specific genetic mutation disorder.

What did you do after finding out his diagnosis?

Right away, Natacha obtained a letter for him to get services. She contacted the state department of developmental disabilities to see what they could do for Peter, and how they could begin to help him. The state was able to set up appointments for his evaluations of Speech, Occupational Therapy (OT) , and Physical Therapy (PT).

By the time the first appointed PT session would come around, Peter would already be 21 months old. He was not yet walking on his own at 20 months of age, but could push something in front of him and walk with it. Somewhat miraculously though, the week right before his first PT session, he had gained the ability to stand on his own and walk, without holding anything like a wagon or push cart ahead of him, to stabilize himself.

The State department also referred Peter for ABA therapy, which he started at 20 months. At first, there were to be 10 hours of ABA therapy weekly, divided up into 2 hour daily sessions, 5 times a week. For this, therapists came to the house. He also received speech services 3 times a week, OT two times a week for half an hour each, and PT once a week, for half an hour.

What is Peter's Education History?

When Peter was about 24 months or 2 years old, he began to attend Special Education Preschool Program A<sup>1</sup>, which was at a school for half of the day. There, he would get ABA therapy in the afternoon. Peter's life began to revolve around the numerous therapies he was receiving, for almost 40 hours a week. Peter spent one year at Program A, in a 6 to 1 class with a one on one para for himself, which was thankfully covered by State aid.

When he turned 3, Peter's early intervention care switched from the State, to the public school district's jurisdiction. He initially stayed at Program A, but switched from a half to a full day schedule, from 8 am to 3 pm. Natacha originally believed this was a good placement for her son where the best services and therapies would be provided for him, but was extremely dismayed to find him restrained to a chair when she came in to pick him up one day. This was certainly not the type of care she wanted for Peter when he was at school, so she knew she needed to find a new place for him to attend as soon as possible.

There was one other place she was considering, that we will call Preschool Program B for the purposes of this paper. Preschool Program B had mostly mainstream classrooms with only one integrated special education classroom. Sadly, Peter's needs and behaviors were going to be more than this school was able to provide for with their current resources, so at first they did not allow his enrollment there. However, Natacha liked the idea of mainstreaming Peter, because she wanted him to learn and grow as much as possible, in the least restrictive environment for him. Mainstreaming is an "informal way of describing the practice of including special needs students in regular classrooms and giving them the exact same opportunities as any other kid to enjoy every aspect of the school experience – from academics, to socialization" (ABA edu, 2017). After Natacha was able to secure the funding for Peter to have a one on one paraprofessional with him at all times throughout the day, Peter was permitted to attend the school. Natacha would send Peter with several outfits packed in his backpack just in case, as he was not yet potty

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<sup>1</sup> Names of schools have been changed to preserve confidentiality.

trained, but this was not a problem at Preschool Program B. The teachers there were all so helpful, and did not mind changing him when necessary. There, they had all of the services Natacha wanted for Peter, while limiting the services she was not thrilled with for Peter's purposes, mainly ABA therapy to be specific. Applied Behavioral Analysis therapy, as mentioned earlier, is a type of therapy that aims to help improve one's social skills, communication skills, and learning skills, through the use of positive reinforcements.

While ABA therapy can be very helpful in many contexts and situations for people with Autism Spectrum Disorders as well as for children or people with other developmental disabilities or conditions, it involves positive reinforcements which usually consist of food like a piece of candy to get children motivated, and Natacha did not find it to be very helpful for Peter, as she notes that at that time, "he was gaining weight, and getting worse" (when referring to his behaviors).

Preschool Program B seemed to be a good fit for Peter, as it had none of the ABA therapy with food rewards that Natacha wanted to avoid. After some time, he started Pre-K 4 there too! In addition, Program B was a nice, alternative environment for him where he could play and have fun, and work at his own pace. Peter enjoyed playing with the wii there as part of his alternative education program! This worked on his hand eye coordination, as well as engaged Peter with many different prompts. The teachers there didn't overlook his academic abilities either, and they began to do math work with him, which he very much enjoyed and excelled at.

When it was time for kindergarten, Peter started in one of the elementary schools in his district, which we will call Elementary 1. There, he was in a consorted class of 8 other students, to 1 teacher, and 2 aids or paras, where Peter had his own one on one paraprofessional once again. A consorted classroom is one in which a group of teachers or specialists are working together, and it is not just the one teacher to the typical 30 or so students. At Elementary 1, he was able to attend recess, lunch, and "specials" with the mainstream students as long as he had his one on one para by his side. In the public school district, "specials" are what is referred to for classes like art, gym, music, computer lab, and library. Peter's favorite parts of the school day were Music, Art, and Lunch. While this new school seemed to be working out just fine, it was still his first year in a public school, and parents are not always able to see what is going on in the classroom, behind closed doors.

Natacha says that the beginning of second grade is when the problems really started for Peter at that school, when unbeknownst to her, the school fired Peter's one on one para. After letting her go, they stuck Peter with any T.A. the building had available that day, and rotated them all the time. This was detrimental to Peter's academic achievements, because not only did the school now have inexperienced people working with him, but any attachment he had formed with his previous para had been ripped away, and now in addition, no one was properly advocating for him at the school.

During parent teacher conferences that year, when Natacha visited Peter's new teacher in the mainstream classroom, it was to her dismay to find the only thing the teacher had to tell her was that she did not want Peter in her mainstream class, and that she would not be permitting it any longer. And if he was not going to be in her mainstream classroom, then he was going to be put back in the isolated class all day. While he still couldn't properly hold a crayon at that time, Peter was bored there, simply playing with toys all day, and not being intellectually challenged at all. Now in this new placement, he couldn't even attend "special" with the rest of the kids. While Peter was nonverbal, and often exhibited stimming behaviors that were hard to control, his mind was neither idle, nor running rampant, and the teachers at his school were ill equipped to work with him. They disregarded him as if he did not know what was going on, but inside his mind, he did, and he had feelings about the way the teachers were treating him that would later be revealed.

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The breaking point was on October 5th 2015, when Peter was 6 years old, and the elementary school called Natacha. It was demanded that she must come down to the school immediately and pick up Peter within the next 5 minutes, due to the fact that "Peter had peed all over himself and was now eating a piece of the pull-up", which Natacha later discovered *never actually even happened* in the first place.

What had truly happened that day, was that Peter was scheduled to attend a speech therapy session, which he hated attending during the school day. And since the school would make him go to speech therapy sessions during his allotted recess time (his favorite time of the day naturally), he couldn't go out and play with his friends, which made him upset. As it would make any kid his age upset, regardless of having a condition. So Peter raced through his lunch to

go to speech class instead of recess, but decided he hated speech and did not want to go, so he peed on the floor. For someone who is nonverbal, and did not have any other means of communicating his message, that was his way of communicating his feelings.

Aside from Peter's being able to communicate this with his mom through typing, and telling her that he absolutely did *not* eat part of his pull-up that day, when Natacha got to the school after the whole situation occurred, she did not in fact find any evidence of a bitten pull-up, nor were his clothes even wet from pee. She noticed immediately that he was still wearing the same clothes that she had sent him to school in, and that they were still dry. Natacha wondered how the school could even get away with such allegations, when they were undoubtedly untrue, and her next steps were going to be to get a lawyer, as she would no longer settle for this sort of treatment from the district.

After the incident that day, the public school district wanted to send Peter to Elementary 2, which was a state school for children with severe disabilities, but Natacha didn't want or like that setting for Peter. Neither did he, as upon their walk through of the school, he expressed his dislike for their lack of desks, chairs, and toys for the students to use throughout the academic day in the classroom for students at Peter's age and grade level. Natacha thought that the classrooms in the high school setting at that same Elementary 2 school would have been more appropriate for Peter, as his mind was beginning to flourish beyond the material being taught in the lower academic levels, especially in the special education schools. From the work Natacha had seen him bring home, it appeared that  $1+1=2$  was the majority of the math being taught to Peter, year after year, when she knew that he could solve equations that simple in his sleep, but yet the lessons never accelerated beyond that.

It was when the public school district finally kicked Peter out for behavioral reasons and suggested a residential placement for Peter, that she and her husband finally asked for an independent evaluation. The school that Peter was previously attending had marked his IQ in the 20 range, but after the independent evaluation was complete, Peter received an IQ score of 145. Before this, Natacha and her husband did not truly know if Peter could understand them or not, but after getting these astounding and majorly discrepant results, Natacha knew that something else needed to be done to help her son receive the proper treatment and care that he deserved, because his current placement was not helping him to thrive.

That is when Natacha recruited the help of Dr. Catherine Lord, “a licensed clinical psychologist with specialties in diagnosis, social and communication development and intervention in autism spectrum disorders (ASD)” (Child Mind Institute, 2021). Natacha wanted Peter to take part in a study on Autism that was being done by Dr. Lord, not too far from their home in Congers.

Dr. Lord had recently moved to NY from Ohio, and opened a center in White Plains where she was conducting a daily study with her PhD students, on participants trying different therapies aimed to help them speak. The initial goal was for Peter to take part in this study on top of his regular school schedule, for 6 to 8 months, and maybe he would learn how to say some words by the time it was done!

Natacha was thrilled about the opportunity for Peter to participate in this program. At this point, he was already attending an after school care program whose focus was on helping children, adults, and families with intellectual/developmental disabilities, behavioral health challenges, and chronic medical needs, so Natacha suggested bringing Dr. Lord’s PhD students to *them*, at the aftercare program. The study was already going to be paid for, and Natacha thought that the teachers at the program could also benefit from Dr. Lord’s methods, but this could not end up being arranged.

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The day that Elementary 1 called Natacha to come pick up her son was also his last day in the district. Unable to come to an agreement with the school district on a proper placement for Peter, and not willing to settle for letting him spend his days restrained in a classroom, stunting and limiting his academic potential, a plan for Peter’s home instruction was underway.

After many negotiations and with a lawyer present, Natacha was at long last able to secure Peter but two hours a day of at home instruction. His speech therapy, OT, and PT therapy sessions would continue to occur at home as well. The home instructors that he had over the next several years were adequate, but when Natacha later wanted to send him to the district’s middle school over the summer for a half day program, the school was not in agreement with Natacha about how to best handle Peter under their care, so they decided not to let him come to their program.

Shortly after this, with the district *now* refusing to even send teachers to her home, Natacha decided it would be better to just send Peter back to a private school. After all, it was not fair or enriching to him to be sitting at home with no instruction or with poor instruction which was through no fault of his own. At first, the private school of their choice which we will call, Private School X, refused to admit Peter for not being potty trained. This was slightly discriminatory, as it was a part of Peter's disability that caused him to be unable to use the toilet on his own. However, if they too did not have the resources to properly take care of him at that time, Natacha understood. A year later, after he was potty trained, Natacha went back to Private School X, but was disappointed at the expensive price tag that came along with it. She had visited a few other private schools, but really did not feel like any of them would be the best fit for him.

Finally, with the help of lawyers, Natacha was able to come to an agreement with the school that worked out for them and for her, and would allow Peter to attend Private School X. At first, he could only attend for a modified half day until 12 in the afternoon, because Private School X was not a school approved by the public school district, and funding for him to attend was not entirely approved yet. However, after continuing to visit other private schools, Natacha decided that no matter the cost, Private School X was the best placement for Peter, because he was now potty trained, and *there*, he would be able to receive speech therapy, PT, OT, physical education, *and* music class, which was Peter's personal favorite!

Private School X is a Waldorf school, whose "pedagogy strives to develop pupils' intellectual, artistic, and practical skills in an integrated and holistic manner" (Waldorf, 20121). Private School X utilizes its own unique approach to education, and even implements its own curriculum. It offers an alternative environment to a typical public school, where the children who attend are nurtured and creativity is fostered. Infinitive play is emphasized, and children are encouraged to explore and be open minded individuals. At Private School X, there is no set time or grade level that a writing or math curriculum must be completed by, and in the early academic levels including kindergarten, first, and second grades, the children mostly play outdoors and learn about nature, how to grow plants and vegetables, and things like weaving.

Additionally, all of the food that is either served or to be eaten at Private School X is organic, and no junk food or pre packaged snacks are allowed. This is extremely beneficial to a child like Peter, or another child with a special or restrictive diet, because it not only limits the

temptation that may cause behavioral issues from other children's snacks that may be harmful to them, but it also helps to improve Peter's symptoms and behaviors when he eats healthy foods that are low in sugar and sodium which many prepackaged snacks are high in.

Natacha also loved that at Private School X, the kids get to play outdoors all year round, even in the winter, because Peter is the type of kid who always loves to run around and get his energy out! He started attending school there in January, but ever since June 22nd 2020, Peter hasn't missed a single day, nor does he want to. Natacha says he loves it there. There is a pond he enjoys to swim in, horses he enjoys to ride, and maple syrup to be made from the sap of the trees.

Peter enjoyed this placement so much, but the finances of sending him there were somewhat of an issue for Natacha and her family. She ended up having to pay an educational lawyer who was able to help advocate for Peter against the school district, as in her heart Natacha knew that Private School X was clearly the right placement for Peter, and was the best place for him to learn and grow academically as well as socially.

It should be noted that in America, children are entitled to an education, no matter their disability, or need for accommodations in the classroom. There are several laws that provide for these accommodations too, including The Individuals with Disabilities Education Act (IDEA), and the Americans with Disabilities Act (ADA) which both outline specific criteria for children to be eligible for special education and related services, which Natacha knew Peter was deserving of. In addition, Section 504 of the Rehabilitation Act "requires schools to provide disabled children with reasonable accommodations... covers all programs or activities, whether public or private, that receive any federal financial assistance... giving this law [a] very broad and powerful application" (HG, 2021), which meant Natacha knew she had the law on her side, and she would not stop fighting until she received the funding needed for Peter. Alas, there was a light at the end of that tunnel, and after much back and forth, finally an agreement was made.

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What are Peter's health complications?

On top of being a nonverbal child, Peter was born with a slew of other health issues that can be credited to his being born with the KAT6A genetic mutation. One of the worst symptoms for him before taking the correct supplementation was severe constipation and an inability to

pass proper bowel movements. His parents know about this constipation now, and have been able to help him treat it, but when he was much younger Peter was admitted to multiple different hospitals, on numerous occasions because he was yelling and screaming out in severe pain. Natacha says that she felt in her gut it was the pain Peter was enduring that was creating his bad behavior. At that time, his parents both knew he was having trouble pooping, but did not realize the severity of the backup that was accruing. Even after administering 7 bags of miralax as prescribed by his doctor, when he was finally able to poop, the pain did not subside. He would not sleep for 4 or 5 days at a time, and be screaming in pain all the while, but when his parents brought him to the doctors, they still just could not figure out what was wrong.

Instead of admitting this, of course, the doctors told Natacha that all of his screaming was due to his “autism”, and that he wasn’t really in pain at all, but that his body and his brain are just *telling him* he is in pain. After hearing that, Natacha once again knew that she needed another opinion, and to look somewhere else to find Peter some answers, and to put him out of this misery he was living in. That’s all a mom ever wants anyway, to get her baby out of pain and into comfort.

At this point, Natacha did admit to feeling slightly hopeless and discouraged by the doctor’s remarks. Peter’s behaviors at home had gotten worse too, and at one point he was physically getting so out of control that he had knocked a kitchen chair down on top of his baby brother John. Accidentally ofcourse, but even still, it was in that moment, when a thought crossed Natacha’s mind. Should she and her husband maybe start looking into a residential school for him? That was obviously a last resort scenario, and she would not have dreamt of that for him, but she also knew that she couldn’t risk having a child in the home who could be a danger to her other three children. Instead of dwelling for long on this idea, she continued to search for alternatives that would enable her to help her son and keep her family together.

So what were the next steps?

Well, this was when Natacha brought Peter for yet another evaluation (the independent evaluation previously mentioned), this time with Diane Sloan, which provided astounding results, that alerted his family to the fact that there was more going on up there in his brain than they might have originally thought. At 8 years old, Peter’s receptive english score was off the charts. He scored a competency that was above a high school level, or that comparable to the

score of an 18 year old! This was especially astounding, seeing as at the same point in time he was still unable to sit still, was still not potty trained, and could be seen ripping his shoes off without reason at inappropriate times.

Following this evaluation with Diane Sloan, Peter's IQ was then again evaluated. Natacha had found a connection in David Salsberg, a neuropsychologist from Manhattan, who was at that time the director of the PALS organization, which stands for Pediatric Assessment, Learning & Support. Peter went to evaluations with Salsberg three times, for three hours each time. Though he was still nonverbal, it was when *this* evaluation was done that Peter received an IQ score of 145. This is considered to be a score of someone who is Highly Intellectually Gifted. Now, in Peter, Natacha knew she was dealing with someone who didn't just have a mental capacity to understand, and to learn, but an excellently advanced capacity at that, and she knew that her fighting to give him a better quality of life should not be quelled, and was what he deserved.

Shortly after receiving this new IQ score, came the psychological and neuropsychological testing previously mentioned. While doctors had not initially recommended this to Natacha when she had brought her *18 month old* son in with concerns, there were new tests available now, and his persistent symptoms lended themselves to further testing, including whole exome sequencing of his DNA. It would later turn out that Peter Najm was the 39th person known in the world to have the specific genetic mutation, KAT6A.

After getting the results from the geneticist, with new answers and a new diagnosis of KAT6A in mind, Natacha's next steps were to see what treatment methods were available. However, even though the etiology was now known, and Natacha and her family now had the diagnoses of a mutated gene being the cause for Peter's symptoms, solace was not around the corner, due to the rarity of his disease and the lack of a "cure", nonetheless treatment options, that were available.

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Simply not willing to give up hope in finding relief for her son, Natacha and her husband turned to the internet community to find others who had been diagnosed with the same disorder, and could hopefully provide some answers. However, when no such community could be found,

Natacha and her husband Emile started from the ground up, in creating their own non profit organization, for the research and support of families whose lives were also touched by KAT6A.

In 2019, their organization, the KAT6A Foundation, held its third annual conference that was attended by doctors and families around the world who are all hoping to be able to bring a higher quality of life and one with less pain to those diagnosed and affected by the disorder.

Thankfully, through advancements of technology and social media, Natacha was able to connect with other families around the world who also had children with this disorder! Upon getting in touch with several of them, Natacha was finally led to new developments, full of information that could help her and Peter out on their KAT6A journey. In addition to finding other families with this disorder online, the Najm family also loves to meet other families who are affected in person, and they have been down to Washington D.C. on numerous occasions for the Walk for Rare Diseases, where they have met a larger community that they have something in common with!

One family that Natacha connected with back in 2016 had a daughter who also had the gene mutation, and was diagnosed with it several years prior. This girl had been prescribed a cocktail of different vitamins and supplements to manage her behaviors and symptoms, and when her parents said that she had been doing very well on this cocktail of supplements, Natacha wondered if they might be beneficial for Peter.

Soon after, she was able to consult with Professor Richard Kelley, who had been doing research specifically on these mitochondrial cocktails. After analyzing Peter's blood work, sure enough Professor Kelley found evidence of mitochondrial dysfunction, and subsequently prescribed a mitochondrial cocktail to Peter. Natacha bought all of the vitamins that were prescribed in the cocktail and gave them to Peter, which included the vitamins C, D, and E, all in high doses, which could be purchased at the local supermarket.

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After just four weeks of taking the mitochondrial cocktail that was given to him by his mother, Peter was all of a sudden able to hold in his urine, and was no longer having accidents! In five weeks, he was able to ride his two wheel bike by himself, with no training wheels, something he definitely couldn't do before! This development in Peter's new found abilities led

Natacha to believe that maybe the way Peter was reacting to his environment were not simply behavioral responses after all. Maybe, the symptoms were due to his having weak muscle structure before taking the cocktail of supplements. This turned out to be true.

Now able to hold his bladder long enough to use the bathroom, Peter was going every ten to fifteen minutes. He had to pee, and would go to the bathroom. So they now knew that his urinary tract muscles were getting stronger, but there was still the question of why he was having to go so often.

After more hospital testing, high levels of acid were found in Peter's blood that had been ignored or 'overlooked' for years. He had been tested at the hospital so many times, and yet this was never an issue brought to Natacha's attention. The doctors at the hospital told her that Peter was most likely just dehydrated, when she brought him in yet again, this time for *holding in* his urine for too long, which turned out to be a newfound inability for him to pee. This was now causing a new problem for his kidneys, because when too much urine is held in, the body's toxic waste product is stuck inside, which left Peter still in pain.

Knowing this new information, Natacha had to put all of these puzzle pieces together on her own. First, she had to fix the acid issue in his blood which she thought could maybe help to ease some of his pain. Natacha used a combination of bicarbonate acid, and citric acid to try and remedy it, which worked, and now Peter is able to hold in his pee if he has to, and can urinate freely when he has to. It was hard to tell where he was when it came to potty training when he was wearing a pull-up all of the time, but now with Peter wearing big boy pants, Natacha was fairly confident that this part of the problem had been resolved.

On top of those remedies, Natacha says that giving Peter celery juice every morning helps to keep his bowels moving along with the citric acid treatments, and limiting his junk food has also worked wonders for his behavior, along with providing his diet with plenty of meat and other protein rich foods to keep him strong, sleeping well, and properly growing.

First, Peter's muscles were not strong enough to hold his pee in, causing accidents, but the mitochondrial cocktail helped that. Then, with the acid problem on top of it causing Peter pain, Natacha had become a detective now too. Especially with her son being nonverbal, and having no way of expressing the symptoms he was experiencing in an articulate way that could help her to figure out what was wrong, Natacha was determined to keep looking for answers.

Aside from his bladder and blood acidity issues, Peter also suffered from terrible belly aches that Natacha refers to as ‘abdominal migraines’, and that she believes are most likely caused by abdominal obstructions. As a doctor herself, Natacha knew that if they were not treated fast that he would need a colectomy (a surgical procedure to remove all or part of the colon), and feeding tubes, which she did not want for her son if he didn’t *have* to have them. At this point, Peter was in so much pain that doctors suggested putting him to sleep in a medically induced coma while they worked on cracking the code and solving his medical issues. Luckily though, this did not have to happen, as together, the doctors and his mom were able to figure out how to drastically soothe Peter’s symptoms and improve his quality of life.

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How does Peter feel about music?

If there is one thing that Peter Najm loves about life, according to his mother, it is music. She says that she is unsure if it is due to his KAT6A diagnosis or not, but Peter has exceptional hearing abilities. His ears are very sensitive to music and other loud noises, but this doesn’t stop Peter from putting on his headphones, and walking right up to the front of a band when they are performing or up to the speakers at a party, to get to the best spot to listen. Ever since he was younger, Natacha says music is the one thing that Peter would be calm and quiet to listen to. She took her family to many concerts while the kids were all growing up, and always thought to herself that if he enjoys it so much, why not try to teach Peter to play music!

Doctors suggested Natacha take Peter for music therapy, but he was not improving in the therapist office’s clinical setting. At least there, Peter was exposed to some instruments he could grab on to and try to play for himself, and when his parents saw him take such an interest, they knew what they had to do. Natacha immediately bought some instruments for Peter to play with at home, including drums and a piano keyboard, and sat back as he excelled.

Soon after, she hired a young man named Alex<sup>2</sup>, to work with Peter privately in her home. Alex worked with Peter day after day, and eventually taught him to play the piano at just 5 years old! At first, it was hard for Peter to learn how to separate his fingers in order to press on the correct piano keys. He needed his wrists to be held steady, so the only thing he had to think

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<sup>2</sup> Names have been changed for the purpose of confidentiality.

about moving would be each finger, one by one. After a few months like this, Peter was able to separate his fingers well enough for his parents or Alex (whoever he was playing music with that day) to stop holding his wrists while he played, and move up his arm to now just hold his elbows. Since it is still somewhat challenging for Peter to gain control of his muscle movements, having someone there to hold his arms steady, and then over time gradually move further up his arms to his elbows and then his shoulders to keep steady while he played, he was able to flourish at learning to play the piano's keys.

Peter had no problems when it came to reading the music, and was easily able to learn all of the scales and notes and correlate them with what keys to press on the keyboard. Alex helped teach him this, but it was surely impressive to any person to see just how *fast* Peter had learned. To this day, *every day*, Peter plays the piano, drums and bass guitar under guidance of Alex, as well as his dad. He feels better while he is playing if someone comforting who he associates with that task is near him when he is doing it. And that is fine with Natacha, as she feels as though one goal she has for Peter is that if he can play an instrument for an hour, without her sitting right near him, that he will be able to have a job on his own one day, without her at his side.

Stability and routine are something Natacha never takes for granted when it comes to things that are beneficial to Peter. Music is something he enjoys and is successful at, so she emphasized how imperative it is that he keep up with it, and continue to hone his skill. His father Emile practices playing piano with Peter for at least 45 minutes every day too. This is excellent bonding time that reinforces one area that Peter is very talented in. He loves music, and is amazing on the piano now, which Natacha credits partially to the fact that he never skips a day of rehearsing. Peter's first accomplishment at this skill was when he was 7 years old and in first grade. Now he is 12 and a level 5 NYSSMA scorer, which is extraordinary, considering most kids even older than he is have a hard time receiving such a high score.

Additionally, as his behavioral issues subsided, Peter was now able to sit still and focus on more fine motor and creative tasks presented to him, such as writing and typing literature, that was not only nominated but was also voted first place (!) when submitted to the all grade special education competition of the New York State PTA Reflections program!

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You may be wondering how it is possible that Peter has all of this talent and almost hidden seeming abilities, and yet the public school he was attending seemed to not know what he was capable of. Natacha attributes this to the fact that the school was never patient with him. They never gave him the one on one time or benefit of the doubt that he deserved. No one who was ever qualified enough ever sat down with him and gave him a chance to show them what he could do and how much he really understood. Natacha knows that his behaviors can sometimes be difficult. She is a mother, and she is not kidding herself. However, if the right person such as his amazing music teacher Alex were there with the right training and attitude a long time ago, the world of education that Peter experienced thus far may not have been as grim. Music saved Peter's life and showed his family that he could do it, his brain was in there and it was working, he just needed a little time, a little push, and a lot of love and support to bring it out.

Covid and KAT6A; How has covid affected Peter?

The Coronavirus lockdown has not been too hard on Peter and his siblings! His household had to change their activities up a little bit, but they never actually stayed home! If they did, Natacha says that Peter would go crazy, and probably not sleep. He needs to be outside and doing things to keep stimulated. Over this whole pandemic, Natacha made it clear that they did not stay home in the house for a single day. When the library was closed, they walked around BJ's. When the mall was closed, they hiked around Rockland, and discovered some cool new trails. When the same public pool that I met Peter and his family at was now enforcing too many pandemic restrictions for Peter to handle, they swapped their daily routine to swimming at the beach! One day over the summer of 2020, Natacha took Peter and his brothers and sister to a *different* public pool in the county though, which she says the kids very much enjoyed. While their capacity may have been limited, there were less stringent restrictions that Peter and his siblings would need to abide by.

The Najm family was able to go out and do things that were open, and they took advantage of every single opportunity, such as going roller skating and to the ice cream place. Natacha says that she had to shift gears a little bit and get creative. She branched out socially, expanding her usual circle, and was able to find plenty of friends for her kids to hang out with, who were also less strict about social distancing rules. This was because Natacha did not want

her children to feel more isolated than they already were going to be, from having to stay home from the closed public school.

Peter is also somewhat claustrophobic, so for him, being able to still attend his school and speech therapy as well as other therapy sessions in an outdoor setting was highly beneficial. His Private School X remained open during the pandemic, as it served the special needs population which was legally allowed to carry on throughout the height of the pandemic's otherwise would be restrictions. Having a large outdoor space with a tent and a bonfire at school made the learning that continued fun, and something that Peter looked forward to every day.

Something else that this coronavirus pandemic has made us all pay attention to is making sure we don't leave our houses without our masks! As it turns out, special needs kids are legally exempt from wearing masks. If someone like Peter were to put one on, he would probably eat the mask, or be constantly putting it in his mouth. In addition to this, he could have pulse oxygen breathing issues that would supersede any requirements or mandates to wear a mask, because wearing one may be more detrimental to his health than *not*. Children with special needs who cannot wear their mask in a proper fashion can suffer from way more issues by wearing dirty masks and breathing in the bacteria from the perfect breeding grounds that is a warm, moist, ill fitting mask strapped to a child's chin or face. Data has already been recorded and reported on, claiming that children who wore masks daily during the pandemic have suffered from dry mouths, cavities, sinusitis, as well as other breathing difficulties from their noses being covered or even closed up by too tight masks that are blocking their airways.

In addition to being open legally to cater to the special education population that they serve, Private School X, being an alternative education setting, was full of parents and teachers just like Natacha, eager for their children to return to as normal as possible in person instruction, and stray away from socially isolating policies. Some teachers at the school chose to wear masks to work while others did not, and some students chose to wear them, while others did not. Ultimately, masks were not mandated for anyone attending the school, which worked out quite well for Natacha, as Peter was a student who would not have been able to wear one in the first place.

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What has the impact been on your family from Peter having Kat6a?

Peter's siblings are amazing at helping him get through his daily tasks. Paul, John and Mary all help get Peter the things he needs throughout the day with ease, because they just know him, and can understand what he wants, and what he doesn't want at this point. Natacha says that they work well together, especially when they all do their chores together, which makes them more fun. When they each do their reading and writing on top of their homework, they all get 50 cents a day, which adds up to an allowance of \$15 a month!

Peter's little sister Mary's favorite thing about him, when asked, is when he plays ball with her and shares his things with her. She also told me that she likes how nice he is to her, and when he doesn't hit. He and his siblings also love to ride the bumper cars together at Spin Zone at the mall (which I have forgotten to mention earlier), and Natacha is hoping that the strong bonds her kids have now will carry into adulthood.

At age 12, Diane Sloan wrote about him in one of her books, claiming that someone with Peter's symptoms due to KAT6A would not be able to live in a house on his own. Now, after years of therapy and supplemental treatments, Natacha hopes that in the future Peter *will* be able to talk to anyone he needs to in order to communicate his needs that have to be met with the assistance of others. Even if he can't work a full time job in order to support himself, his family wants him to be more independent. They envision him living with a roommate or two who could be a caretaker, or potentially his sister or one of his other siblings after Natacha and her husband pass away. Natacha says that while Peter can generalize certain skills from one lesson or idea to the next, he can not be trained on everything. One important thing that Peter *does* have committed to memory, is that he must put on his seatbelt every time he is going for a ride in the car. Things such as that which involve his own safety are of utmost importance for his future, and any hopes of independent living his family has for him.

Lastly, What are some other noteworthy things about raising a child with KAT6A?

One child with KAT6A is most definitely going to be different than any other child with KAT6A, and this is important to remember. Natacha says that she can tell us some other interesting things about Peter though, that are most likely due to his diagnosis.

1. Peter tends to be very specific about the people who work with him on specific tasks, meaning that he likes to do specific tasks with the people who he knows are specific to that task. For example, His dad and Alex are his music teachers, so he loves to play piano with their help. However, when it comes to swimming laps at the pool, he wants Mommy all the way. Peter used to be involved in swim lessons and was even on a swim team. Natacha wanted him to participate in the Special Olympics swim competition, but he did not want to swim laps without her there with him.
2. Peter has anxiety about many things, but one of those is typing even though it is his main form of communication. He used to not even be able to type for his teachers at school, but now he types more for them and is able to better express himself. When asked, “who is Jesus” he typed back, “it is god”. When asked “who is god”, Peter responded that “he is creator of the universe”.
3. Learning to communicate has saved Peter’s life. He can now say what is going on to alert others to get the right help or things that he needs. Natacha tried to teach him to communicate through the ipod when he was younger, but he was unable to accurately press such small keys and convey a proper message. It is especially easy for him to type when using large keys such as those from a desktop computer, or a special AAC communication device.
4. Though he is classified as someone who is nonverbal, Peter does have some ability to sing and talk, which he does sometimes at home and often works on in therapy. Currently, his vocal attempts are very breathy, and consist of separate monosyllabic utterances that have louder and stronger beginnings, that tend to trail off and not demonstrate any ending sounds.
5. When he needs to get someone’s attention, Peter tends to yell loudly as well as close his ears. If a stranger saw this behavior they may not know that this is Peter’s way of expressing that he wants something, but his mom is able to read him by now and tell what he wants.

6. With his muscles getting stronger and stronger everyday but still not able to execute very many fine motor skills, it is easiest for Peter to write with chalk on a chalkboard attached vertically to the wall, instead of writing horizontally on a piece of paper flat on the table. This, he would have more difficulty with.
  
7. Peter has apraxia of space. This can be compared to Limb apraxia, where “a wide spectrum of higher-order motor disorders result from acquired brain disease affecting the performance of skilled, learned movements” (Leiguarda, 2000). His mom describes this for Peter as having a major disability to find things in space. He can point to his head, ears, eyes, nose, and mouth sometimes, but other times he can not direct his body movements to point to the right facial parts. It is not that he does not know which parts are which, because cognitively speaking, he does. I have witnessed this myself first hand with Peter.

Another example of this was when I witnessed his mother ask Peter to go to the fridge and get out the almond milk. He could not follow these simple instructions, even though he does know what the fridge is, what the almond milk is, and that the almond milk is sitting on a shelf in the fridge.

This is something that I found very interesting about Peter and his unique situation, and his mother does too. It is apparent that Peter understands the commands that are spoken to him, because he can type a response that answers a question correctly, or explain something to us, but when he has to put it all together at the neurological level and send a signal from his brain to his limbs to complete an action and make a body movement happen, he is unable.

For example, when his mom asked Peter where the milk is, he answered that it is in the fridge. Next, when asked where the fridge was, Peter responded that it is next to the microwave, (which it was, at his house). This demonstrated that Peter did indeed know what milk is and where it was, and he did in fact know where the fridge was too, but he just could not put all of the parts together and move his muscles in the proper way to go retrieve it on command.

So what if Peter was hungry or thirsty, and there was no one around to help him? First of all, Peter is never typically left alone. He needs one-on-one adult supervision always. However,

Natacha says that if he feels hunger or thirst on his own, he is in fact able to (and welcomed to) go to the fridge, open it up, and take something out for himself. So how does that work? Hunger and thirst are survival instincts. Peter's body can respond to instincts like those, and use muscle memory to complete the task and fulfill his needs. I was fascinated by this development.

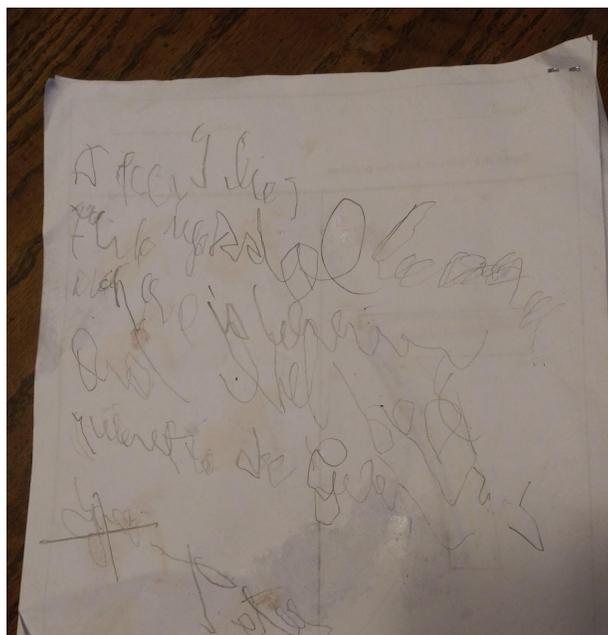
Natacha also attributes his speech issues and non verbal communication style to this form of Apraxia, as she says that he can't speak because his brain does not know where his tongue is in space, or how to properly move it into the proper location to make certain sounds. As a student of communication disorders, something I have learned at the beginning of my journey in the field of speech is phonetics, including how to produce each specific consonant sound in the mouth through placement, manner, and voicing, and where the tongue needs to be placed in the oral cavity in order to produce proper vowel sounds. For Peter, regulating his tongue movement is almost impossible, as his brain does not properly send the signals to the body parts it has to, most likely due to his having KAT6A disorder.

Similarly, if Peter were asked to point to an object such as the toaster oven, he is unable to use his hand to point to its location. He would be able to describe the location of the toaster oven to you through typing, but he is unable to gesture to where it is in person.

8. Natacha thinks that Peter is able to play the piano because its notes are continuous, but she has observed that if there is a song that requires a key that is farther away and disconnected to the other keys he is using for a particular song, he has to reach farther away, which causes him difficulty.
9. Following more than one step directions is nearly impossible for Peter. He can bring his laundry downstairs to the washing machine, but can not follow or perform the direction of bringing down his clothes **and** putting them in the machine.
10. Peter is extremely adept when it comes to math problems, but can not control sensory triggers that bother him and may cause him to lose his composure. Going off of ability to keep his composure, one not so positive behavior Peter exhibits, is that if he sees a bathroom and needs to go, he may run off to it, and if he doesn't see it he may pull his pants down and scream for it.

11. At 12 years old, Peter is able to dress himself and put his own clothes on if they are laid out before him. He is also able to put on his shoes by himself if they don't have laces.
12. Previously, Natacha was not able to have a babysitter due to Peter's severe behaviors before starting the supplements, but now after much improvement, he and his siblings are able to be home without her and her husband if they ever want to go out!
13. Peter is a smart boy. He has instincts for safety and keeping himself out of harm. He does not run off of her property or into the road, as he knows better and is scared to get hurt.
14. Fun Facts about Peter:
  - a. He knows and understands some French and Arabic!
  - b. He takes Religion classes with his siblings where he is able to behave.
  - c. He loves to cook, and make cupcakes! He picked up this hobby this year in the pandemic.
  - d. He likes to help do his laundry downstairs!
  - e. He can go to *lulu's*, a local restaurant, by himself with some money and buy a cupcake!
  - f. He has a very advanced reading level.

Here is a sample of a correspondence that was written from Peter to myself during the process of writing this thesis paper!



Additional questions for Peter:

- a. What is your favorite color? BLUE
- b. What is your favorite type of music, or singer? Arabic music, or Nancy Ajram!
- c. What is your favorite instrument to play? DRUMS
- d. What is your favorite thing to do in the summer? SWIMMING
  - i. In the winter?
    1. Playing in the snow, and eating the snow!
- e. Which do you like better, reading, or math? MATH
- f. What's your favorite food? HAMBURGERS
- g. Where is your favorite place in the world? THE BEACH

Through the power of modern technology and science, proper diagnoses, therapies, and modern treatment techniques, people who are born with and live with rare genetic conditions can live happy, fulfilling lives, which may not have been the opportunity of those who lived before such advancements and developments in DNA sequencing came about.

Hopefully, more and more research about KAT6A will continue to be done in the future, better therapy options will become available, and people who work in the field of education can find sympathy/empathy in their hearts when faced with people who may act or 'behave' differently than them, because no one ever knows what reason a person may have for "acting" or responding in the way that they do, so have compassion when interacting with people with disabilities.

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