Alexander: A Case Study in Autism

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Objectives:
• To compare normal and abnormal child development with respect to autism
• To determine factors scientific research has and has not implicated in causing autism spectrum disorders
• To analyze inheritance patterns within families
• To explain possible treatments available for Autism Spectrum Disorders

Part 1 – Brian and Alexander

Brian was born February 8th 1997. He weighed 9 lbs. 2 oz. and was 21 inches long. He screamed at the doctor and nurses and turned bright red. Eventually he settled down and his skin returned to a wrinkly, white color. When he was not sleeping, usually at 2 and 4AM you could see his brown eyes. At two months he was smiling and making a cooing sound his parents, John and Patty, loved. At 4 months Brian enjoying playing games with people and became upset when they stopped. He appeared curious and would watch things move from side to side. He also began to roll from his belly to his back. At 6 months he could distinguish between people he knew and those he did not know. He also responds when his parents called
out his name. Patty and John argued whether the vowel sounds he made were “mama” or “papa”. Neither was right. His curiosity continued to grow and often reached for objects just out of reach and sometimes within reach. Whatever he grabbed he tried to put in his mouth. One of Brian’s favorite games was to pretend talking on the phone. He was always pointing at things. At 9 months Brian learned an important phrase. It was “NO BRIAN”. He was into everything he could crawl to and Brian could crawl faster than many people can walk. He began plays his favorite game, peek-a-boo and was able to pick up his favorite cereal, Cheerio’s®, between his thumb and index finger. He loved his plastic dump truck and became agitated when it fell from his high chair. At one Brian began handing Patty his favorite book, The Very Quiet Cricket, for her to read over and over. Brian is clearly able to say “mama” and “dada”. At 18 months Brian is off and running – well – walking. He is eating with a spoon and drinking from a cup.

Brian not only knows what the word “no” means, he now says it while shaking his head vigorously. Outside the house he begins to explore the world but only when Patty or John is around. At two Brian was excited when there was other children around which made John and Patty happy. They hoped he would enjoy having a brother to play with since Patty was pregnant.

Brian was 2½ when Alexander was born on July 18th 1999. The due date for Alexander was July 30th and he was smaller, weighing 7 lbs. 5 oz. and was 17 inches long at birth. Alexander was a beautiful boy with thick dark hair and olive colored eyes. Patty knew they would darken with time. All the babies on her side of the family did including her sister Kate’s beautiful baby who was recently diagnosed with an Autism Spectrum Disorder (ASD). Kate’s husband Dan took a leave of absence from Binghamton State University where he was a Math professor. At family functions, John, an economist with People’s Credit Union, and Dan loved to talk numbers while everyone else did fun things like bocce. Alexander grew but Patty’s instincts had her concerned. At 3 months Alexander did not smile or make any sounds. At 4 months he became upset when Brian made loud noises which he did often. Other times when Patty called Alexander’s name it seemed as if he couldn’t hear a thing. Even after 6 months Alexander was not making any sounds. He did not smile when others smiled at him and hated being bounced on people’s knees, something Brian always loved. When Patty tried to read to Alexander would stare off, into space and at 9 months he would crawl away but not towards anything specific. In fact, the curiosity Brian showed while growing up was almost completely absent. Patty had heard people talking about “refrigerator moms” and worried that she might not be as affectionate with Alexander as she had with her first born, Brian. Dividing time between two children was difficult, especially when John was away on business. John had heard on the radio that older fathers, he was 51, had an increased risk of having autistic children. At 10, Alexander was crawling towards the cat litter box. When Patty said, “No Alexander” he kept moving and she had to drop a hot pan in the sink to stop him from touching it. She would have yelled but it always led to his yelling and shaking his hands uncontrollably. When Alexander was calm he would make unusual movements with his hands near his face. Patty and John discussed the development of Brian and Alexander often and didn’t know
exactly what they should do. Both Brian and Alexander had been vaccinated for Mumps, Measles, and Rubella. Didn’t they hear something about vaccinations and autism? They thought about talking to their family doctor. They trusted her but when they spoke with her at their 6 month check-up she dismissed their concerns by saying, “Each child develops at their own pace, give Alexander time.”


- **Activity 1.2**: Use the Autism Speaks M-CHAT diagnostic tool ([http://www.autismspeaks.org/what-autism/diagnosis/mchat](http://www.autismspeaks.org/what-autism/diagnosis/mchat)) to determine whether Patty and John should seek a specialist for diagnosis and possibly early intervention therapies for Brian and/or Alexander.

- **Activity 1.3**: Research science references and the popular media for stories linking maternal care, age of parents, and vaccines to autism.

**Part 2 – From a Molecular Point of View**

Not content with waiting, Patty and John did some research on their own; looking for any medical explanations as to their son Alexander’s delayed development. In looking through scientific articles, they were intrigued by a graph from a *Science* Perspective showing the expression of various genes during human neurodevelopment (see below).
Activity 2.1: Working in pairs, choose one of the genes listed in the diagram above and research it using [http://ghr.nlm.nih.gov](http://ghr.nlm.nih.gov). Include the normal function of the gene, how changes in the gene relate to health conditions, the chromosome number where the gene is located, and the base pair length of the gene. Report out your findings to the entire class.

Activity 2.2: Explain how Alexander can show symptoms of Autism Spectrum Disorder, while John, Patty, and Brian do not. Include a Punnett square with your explanation.

Activity 2.3: Construct a pedigree showing Alexander and his immediate family, as well as his Aunt Kate, Uncle Dan and three sets of grandparents.

Part 3 - Emerging treatments for Alexander’s social symptoms

“Worms? How will worms help Alexander socialize?” John is looking at the computer screen in disbelief. “Did you hear that honey? Worms! Pig worms! They might help Alexander” he calls to Patty who was in the living room with their sons Brian and Alexander. After a couple years the couple learned a lot about the
possible causes of their son’s autism. They also learned that they were not alone in their search for answers. 1 in 88 children are diagnosed with autism. The statistics are more staggering for boys. Groups such as Autism Speaks® provided their family with answers when Alexander was first diagnosed. The specialists told John and Patty that each autistic child is unique and that they would work together to create a plan that best met Alexander’s needs and that the plan should be revisited frequently. Early intervention included a variety of behavioral therapies and some slow progress was being made, especially with immediate family members and therapists. The pace of improvement tested the couple’s patience at times. Alexander spent 25 hours a week at a special learning center for autistic children. Alexander’s social skills were improving along with his social skills. He really enjoyed spending time in the pool and became upset if his swim schedule was disrupted. John and Patty adored Alexander and hoped that some of the research being done around the world might offer additional treatments for some of his sleep disturbances, digestive system problems and especially the social symptoms. Alexander was uncomfortable at larger family gathering and would not play with his cousins. Alexander’s doctor told them that according to the Fifth Edition of the Diagnostic and Statistical Manual of Mental Disorders (DSM-5) his social symptoms fell on the more severe end of the autism spectrum. Some days it even seemed Alexander was losing some of his hard earned social skills. Either John or Patty attended lectures about the state of research. John remembers a psychiatrist from Stanford University saying “I won't be talking about the state of pharmacology in autism, but about the MISERABLE state of pharmacology in autism.” Currently there are no FDA approved drugs for autism. There are some that treat the symptoms but these were mostly developed for other psychiatric conditions. So John was excited when he read a Nature© article by Michael Eisenstein which said doctors and scientists are eagerly pursuing drugs that target the social symptoms of autism. He was especially excited about a preliminary study of oxytocin and how it changed brain patterns. John had heard about oxytocin. “Was this the so the love hormone he heard people talk about? Could it increase Alexander’s trust of others?” He hoped it would but what was this thing about worms?
The prevalence of autism like autoimmune disorders and allergies has increased over time. Some people have begun to wonder if there may be a connection. Some parents, including John and Patty, noticed that their autistic child’s social interactions improved when they had fever. Others with autoimmune disorders had found that intentionally infecting the gut with pig whipworms as alleviated their symptoms. A formal clinical trial has begun at a leading autism center. John thought, “Maybe the worms can do what other scientists have not been able to do.” A man named Stewart Johnson tried it on his son and reported some positive results. John and Patty were ready to try anything that might help.

- **Activity 3.1**: A wide variety of medications have been used to treat irritability in children with Autism Spectrum Disorders (ASDs). Describe how each of the antipsychotic pharmaceuticals, aripiprazole (marketed as Abilify) and risperidone, decrease irritability in patients with ASDs. What are some of the negative side effects of using these drugs, especially in younger children?

- **Activity 3.2**: Some autism specialists prescribe anti-depressants from a family of drugs called Selective-Serotonin Reuptake Inhibitors or SSRIs. Serotonin is an important neurotransmitter released by neurons in the brain. Define neurotransmitters and how they allow neurons to communicate with other neurons. Describe the structure of neurons. What are the roles of serotonin in the brain? How do pharmaceuticals from a family of drugs called Selective-
Serotonin Reuptake Inhibitors which include fluoxetine (Prozac) and citalopram potentially alter behavior in children with autism? What are some potential negative side effects?

- Activity 3.3: Autism researchers are about to begin a five-year study on 300 autistic children examining whether oxytocin helps their social function. Oxytocin is a hormone released by the posterior pituitary gland. How do hormones differ from neurotransmitters? How does oxytocin affect behavior?

- Activity 3.4: Research additional treatments for patients with Autism Spectrum Disorders (ASDs). Suggest a course of action for Alexander and his family.