Autism: Its Many Faces and Mysteries

(This picture is from HBO Documentary Films’ “Autism: The Musical.” From left: Adam, Neal, Elaine, Lexi, Henry and Wyatt. This documentary was inspiration for writing this paper)

Timothy Tunis

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“I have neither given nor received unauthorized aid on this assignment.”
“I might have autism, it might be an inch of autism where some people have a foot of autism and some people have a mile of autism. Or people have a millimeter of it. It doesn’t really have a meaning. It’s just a word.” -Wyatt

Part A:

Autism is a neurobiological disorder that usually starts around the age of three and continues throughout an affected individual’s life (6). It can be considered a developmental disability because, at first, a child appears to develop normally and then develops symptoms of autism usually before the age of three. It is also considered to be a “spectrum” disorder because it affects each individual with varying degrees of symptoms (6,8,9). Common behaviors associated with autism include repeating the same words or phrases over and over, repetitively playing with toys the same way, or arranging items in strange ways (6). Some other common symptoms of autism, associated with the communication side of the disorder, include lack of “babbling” during infancy, difficulty maintaining eye contact, unusual speech patterns, difficulty understand body language and tone of voice when expressing humor, sarcasm, irony etc (6,9). People affected with autism have difficulty seeing things from another person’s perspective. Autistic people have trouble with loud noises, bright lights, and large crowds (9). They usually have a passionate interest in either one or two fields and have difficulty keeping multiple friends (9). These are just a few of the many symptoms that people with autism suffer from. Not all of the symptoms listed above affect every autistic person. Furthermore, the symptoms have varying degrees of severity from person to person (6,9). This and many other factors make it difficult for scientist to understand an already complex disorder.

Most scientists are in agreement that autism is a result of both genetic and environmental factors (2,5,6). This, unfortunately, is one of the few things that scientists have been able to
agree on when it comes to autism. Through the next several paragraphs I will present various studies that have been conducted by scientists in an attempt to gain understanding on this mysterious disorder. These studies are just a few of many studies that have been and are being conducted right now.

The reason scientists believe that genetics plays a substantial role in autism is because of studies done on twins, both fraternal and identical, and families that have a history of autism (6). “When identical twins have autism, both have autism more than 60 percent of the time, depending on the criteria used. When fraternal twins have autism, both have autism between 0 and 6 percent of the time (6).” If there was no genetic component to autism then the chances of fraternal and identical twins both having the disorder should be the same. Furthermore, brothers and sisters who have an autistic sibling have between a 2 and 8 percent chance of being autistic themselves (6). While the chances are still low, they are much higher than that of siblings from an unaffected family. Another potential aspect of autism was covered in a recent report done by CNN which called into question a sperm donor, “Donor X (2).” After six women used “Donor X’s” sperm to conceive a baby it was found that three of the children had autism and another is beginning to show signs of being affected (2). If autism was not genetic then the chances of this sperm donor conceiving three and possibly four kids out of six with autism would be astronomically small.

There is scientific evidence that 12 or more genes located on different chromosomes may be involved in autism (6). Some of these genes may simply just place a person at greater risk of having autism while others may add to the symptoms. Scientists believe that chromosomes 2, 7, 13, 15, 16, 17, and the X chromosome may have some part in the disorder (6). Chromosome 2 is the location of the HOX genes, which play a crucial role in growth and development during early
life (6). If negatively affected it could lead to some of the symptoms consistent with autism. Chromosome 7, home to a region called AUTS1, is very likely to play a role in autism. This region of genes is known to be involved with speech and language disorders. In one study, “35 percent of families tested showed linkage for chromosome 13.” Scientists are now trying to replicate these results. Genetic errors on chromosome 15 occur in around 4 percent of people with autism (6). This chromosome is known to cause Angelman syndrome as well as Prader-Willi syndrome both of which share characteristics similar to the symptoms of autism. Chromosome 16 is known to house genes that if disrupted will cause symptoms similar to that of autism (6). Chromosome 17 may hold the most promise for researchers looking for clues into the disorder. “A recent study found the strongest evidence of linkage on this chromosome among a set of more than 500 families whose male members were diagnosed with autism (6).” This chromosome is known to cause problems such as galactosemia, as well as, contain genes for regulation of serotonin levels. Both galactosemia and serotonin levels affect neural development. Finally, the X chromosome is known to be the origin of disorders like Fragile X syndrome and Rett syndrome, both of which have common characteristics with autistic symptoms (6). Furthermore men are four times more likely to be affected with autism than women. Since men have only one X chromosome they are more likely to show the effects of a faulty X chromosome whereas women have two X chromosomes (1). If one of the X chromosomes is affected the other one may be able to make up the difference so that the woman can function normally (1).

Scientists at Johns Hopkins University in Baltimore believe that maternal antibodies may contribute to autism (5). Dr. Harvey S. Singer and his colleagues note that, “children with autism have antibodies in the blood that react against brain tissue.” During fetus development,
maternal antibodies can sometimes cross over through the placenta and attack the developing brain tissue of an embryo (5). In a study of mice injected with neural antibodies, they were found to exhibit behaviors consistent with that of someone affected with autism (5).

Many parents of children who have autism believe that vaccines, specifically MMR (Measles, Mumps, and Rubella), administered during childhood have something to do with the disorder. Case studies done by the Institute of Medicine (IOM) and The American Academy of Pediatrics (AAP), found that evidence does not support the claim of a link between MMR and autism (7). Another case study showed that cases of autism were steadily rising since 1979 before the MMR vaccine was available in 1988. Furthermore, there were no sharp increases in autism rates after 1988 (7). Children who were vaccinated exhibited signs of autism at the same time as children who were not vaccinated. If MMR had a role in autism then children who were vaccinated would exhibit symptoms before children who were not vaccinated (7).

The age of the parents could also have an affect on the likelihood of having an autistic child. A study published by the Archives of General Psychiatry in 2006 reported that children whose paternal father is over the age of 40 are six times more likely to have autism than children whose paternal father is 30 or younger (4). This could be due to the fact that genetic material loses its ability to repair itself as an individual gets older. This increases the chances of faulty genes being passed from parent to child (4).

Another study was done on pregnant women from specific parishes in Louisiana from 1980 to 1995 which were directly hit by hurricanes or tropical storms (3). The study showed an increased occurrence of autism in children of mothers that were affected by a hurricane during the middle or end of their pregnancy. This may suggest that children are at an increased risk for developing autism if stressful situations occur during the pregnancy (3).
“Why is my daughter in the 8th grade and learning how to wash dishes? I want the world to value her and they don’t. And I can make them. I can share with people who look at her funny and try to enlighten them but I can’t make them value her. And I can kill myself and feel horrible all the time or I can focus on loving her.” – Lexi’s Mom

Part B:

Neal, from “Autism: The Musical”, cannot speak without the aid of typing machine and has trouble controlling and expressing emotions (10). Neal is autistic. Wyatt has verbal skills equal to those of kids his age. He struggles in a mainstream school system but does not belong in a “special education” class (10). He can vividly express his emotions and understands certain aspects of life that most people do not learn until their late teens. Wyatt is autistic. Lexi has trouble processing verbal questions, often just repeating the question back to the person asking the question, as her answer (10). Yet, she can sing entire songs on key and while keeping up with the rhythm. Lexi is autistic. One word and three people with completely different symptoms and problems. How can one word mean so many things? While there are labels such as Asperger’s syndrome, PDD-NOS (Pervasive Developmental Disorder-Not Otherwise Specified), and ASD (Autism Spectrum Disorder) among others, to denote the severity of the disorder, autism is most often used to be all encompassing (6,10).

It seems through my research that I have more questions now than I did before I started. There is extensive research currently being done on autism at universities and foundations around the world. With the numbers of autistic children being born increasing everyday it seems this could not come a moment to soon. Autism has become, “the fastest-growing developmental disorder in the United States (13).” But has the number really been rising? A few decades ago many of the symptoms consistent with autism were thought to be normal childlike behavior (13).
Now that scientists have realized that there is in fact a problem and early intervention is a must, a wider and wider net is being cast to ensure that they are getting every child that may be affected (11,14). In fact, due to concerns of doctors that they are not identifying all the cases of autism, “The American Academy of Pediatrics… stressed the importance of screening every kid – twice – for autism by age 2 (13).” Based on this evidence I am not sure I believe that the cases of autism are increasing as much as some people believe. It may be merely that the once “normal” kid’s behavior is now something more sinister.

With early intervention being a crucial part of controlling the disorder I believe it is of the utmost importance that scientists be looking for new and earlier ways to diagnose autism. Unfortunately, as things stand now, the disorder is rarely diagnosed before the age of three (14). A new study being done by the director of the Kennedy Krieger Institute in Baltimore, Rebecca Landa, PhD, is diagnosing children with autism at 14 months of age with a 70 percent success rate. She says, “At 6 months of age, babies with autism were no different than anybody else,” but, “By 14 months, though, kids with autism are different in both language development and motor control. They are not globally mentally retarded. Whatever is wrong with them, it influences their motor system as well as the development of their language system (14).” If more can be learned from Dr. Landa’s research it may be possible to increase the diagnoses time by several years giving the child an early start on combating the symptoms (14).

Many parents of children with autism seem to be convinced that vaccines played some role in their child developing the disorder. Doug Flutie, the retired NFL great, son Doug Jr. was diagnosed with autism soon after birth. Flutie has since developed the Doug Flutie Jr. Foundation for Autism. While watching a special on Autism on Larry King Live, Doug Flutie stated he thought that the vaccines played an important role in his son developing autism. Many
scientists and doctors are not supporting this theory and through their extensive research on the subject it is hard for me to accept that vaccines play a major role in autism (7). I think it is human nature to seek out a solution to a problem as a means of comfort. To tell a parent of an autistic child that it could be genetically linked, it could have been something they were exposed to, it could be antibodies during fetal development, it could be vaccines, or it could be a combination of all these factors, is just not an adequate answer. Furthermore, for me at least, it would be much more comforting to think that my genetics had nothing to do with my child’s problems. Since the disorder is usually diagnosed around the age of three it is easy for parents to finger the vaccines as the culprit to their child’s problems. When in reality, their child was probably exhibiting signs of autism well before then, possible even around six months of age but these signs went unnoticed as typical kid behavior. Doug Flutie also states in his interview that his son was saying full sentences and playing tee ball then regressed to not being able to speak or play sports. While this is difficult to explain, and probably even more difficult for a parent to understand, a child with autism who is not treated at an early age can regress back from speaking and other activities.

There are ample amounts of compelling evidence linking genetics to autism. At the very least genetics puts someone at an increased risk for developing the disorder. I personally believe that genetics is the main reason for someone developing autism. Genes can be turned on at any time during a person’s life (12). This could help explain why at first a child may look normal then develop the symptoms of autism. With evidence suggesting that at least 12 genes on multiple chromosomes having something to do with autism it would be foolish to think that genetics does not play at least a role(6). There needs to be more studies on the chromosomes in
Part A in order to further our understanding of autisms origins as well as work towards a potential cure.

The real inspiration for this paper came from Autism: The Musical which aired on HBO’s Documentary Series (10). It was amazing to see the spectrum that this disorder covers. Some of the kids were not able to speak on their own while others would look like normally functioning kids if you just took a quick glance at them (10). This is why it is so hard for me to accept that all of these kids are affected by the same thing. In actuality these kids are each affected by very different disorders which lead to very different symptoms. But it is the label of “autism” that makes them all the same. Scientists will need to be able to differentiate between the disorders and what causes each symptom in order to help these kids. Even if there is a major breakthrough with one of the contributing factors to autism it is unlikely that that breakthrough will be able to help every person affected. It is also important to be able to differentiate the symptoms in order to specify the treatment to meet the needs of each child.

Probably the hardest part of documentary was seeing the toll that this disorder has on the parents. To go from rejoicing in having a “healthy” baby to the realization that all those dreams you had for your kid will not be possible. In the film multiple parents of the kids say at one point or another they thought about killing themselves because the emotional toll was too great. Lexi’s mom at one point says that she hopes Lexi dies before she does because she worries about what type of care Lexi will get when her and her husband are no longer there to take care of her (10). It is every parent’s nightmare that they die before their kids do but in her case it may not be such a bad thing. Many of the marriages in the film were either very rocky or they were divorced. Kids affected with autism are very high maintenance both financially and emotionally (10). While the kids should take priority I think it is important for families of those affected to get
counseling. Hopefully, in the near future, the government will provide aid in the form of counseling to families affected.

Through all the turmoil you see during the documentary, all of the hardships, sadness, doubts, and questions that cannot be answered, you see what these kids are capable of. The musical was certainly not ready for Broadway, by any means, but it was still something special. If nothing else it should give people hope and that these kids are worth fighting for. They did not ask to be in this position and they should not be thrown under the bus like so many other people with disorders are. With the number of autistic cases rising to an estimated one in every 150 kids being affected it is going to be of the utmost importance that we develop new testing methods to identify kids with autism at an early age (14). It is also going to be important to figure how to effectively treat these kids so that they can make a contribution to society. If not we are going to have a large part of the population who posses no job related skills and then what do we do?

Autism is an extremely difficult disorder to understand. It has many unknowns that need to be answered before we make any real progress. What is known about the disorder only makes it more complex. It is easy for people to get frustrated with scientists because they want a quick answer. We have sent a man to the moon, developed a vaccine for polio, and even mapped the human genome. The human race is an amazing species but all of these things did not happen over night. When it comes to the complexity of the human genome, sending a man to the moon seems like child’s play. Scientists need adequate funding from the private sector as well as the government in order to study this disorder thoroughly. Thankfully it seems people are beginning to understand the severity of the situation and action is being taken. With April being autism awareness month you can not turn on the news without seeing something about autism. Progress
is going to be slow and it is going to take time to understand this disorder. Unfortunately time is not something that these kids have (11,14).
References:

Part A


Part B:


