Autism Spectrum Disorders: New Genetic Cause Identified

A team of researchers from IST Australia have recently discovered a new genetic link to autism spectrum disorders, along with the method that this newfound mutation uses to cause autism. These particular discoveries led the scientists to find that mutations in many other autism-linked genes behave the same way, showing that they may have also discovered an autism spectrum disorder subgroup. However, the scientists stated that the process to finding all of these mutations was long and difficult.

Through a group effort, scientists discovered a mutation in gene SLC7A5, a gene that transports branched-chain amino acids to the brain, in people who had syndromic autism. To further study this, the researchers removed the SLC7A5 gene from a group of mice, lowering the amount of branched-chain amino acids in their brains. They then observed their behavior, and found that these mice showed classic signs of autism such as reduced social interaction. The scientists wanted to see if these effects could be changed somehow, so they introduced branched-chain amino acids directly into the brains of the mice without the SLC7A5 gene. After roughly three weeks the mice started to participate in more social interaction and had an overall improvement in behavior, showing that autism spectrum disorders caused by an amino acid deficiency could possibly be reversed or at least treated. Dora Tarlungeanu, one of the researchers, supports that their findings here could be extremely beneficial, but changing the delivery method of the amino acids for humans would be tricky. However despite the difficulties, the researchers of this study have a positive outlook on the treatment for the future.

This article interested me at first glance because as a child I grew up attending school with numerous kids who had autism, and my dad’s cousin was also severely autistic just recently passed around the age of 45 due to complications. My whole life I’ve been surrounded by people with autism and have seen the effects the disorder can have on not just the individuals’ lives, but their families’ as well. I think it’s amazing and important that research is being done on the topic and that scientists are getting closer and closer to breakthroughs for treatments everyday.