

## Genetics Writing Assignment #4- Kathryn Kuder

In this article, the researchers studied the genes that are often connected with intellectual disabilities and autism spectrum disorder. The technique that they used in this experiment was whole exome sequencing, which is a kind of genetic testing that uses biological samples such as saliva or blood to track down the genetic cause of a particular disease or disorder. The purpose of the experiment was to study the genes already known to be connected with autism spectrum disorder and these intellectual disabilities and to detect pathogenic variants of these genes. In other words, these pathogenic variants are genetic mutations that cause the carrier of these genes to be more susceptible to a certain disease or disorder. This type of testing is important because family members diagnosed with these conditions can be helped earlier on.

In order to carry out this experiment, sixty groups were selected to be tested, each of these groups consisting of a patient with a diagnosis of either an intellectual disability or autism spectrum disorder as well as their parents. The members of these groups were tested for pathogenic variants of genes already known to be linked to autism spectrum disorder and intellectual disability as well as new genes that could be linked to these disorders, especially since these disorders are associated with several genes.

According to the results of the testing, there were eight pathogenic variants found of genes that were linked to these disorders. In addition to these genes, there were four new genes that were found to be linked to intellectual disability and autism spectrum disorder, three of those genes resulting from frameshift deletions or duplications. Among the patients with these four genes, there were several diagnoses of other conditions such as epilepsy, facial dysmorphisms, language delays and ADHD. Finally, there were many other variations of unknown significance that were found to be connected to these disorders.

Bruno, Lucia Pia. et. al. New Candidates for Autism/Intellectual Disability Identified by Whole-Exome Sequencing. National Library of Medicine; <https://doi.org/10.3390/ijms222413439> (2021).

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Whole Exome Sequencing. *Yale Medicine* (2022). Available at: <https://www.yalemedicine.org/conditions/exome-sequencing#:~:text=%E2%80%A2A%20type%20of%20genetic,%E2%80%A2Involves%20Genetics>. (Accessed: 21st February 2024).