

Down syndrome is a condition when a child is born with an extra chromosome, specifically the 21 chromosomes. This condition is also referred to as Trisomy 21 because Trisomy is a medical term used to describe having an extra chromosome. Cases of lower levels of hypertension and certain types of cancers are also common in people with down syndrome.

An example of how down syndrome and genetics play a role together is how Alzheimer's disease has an increased occurrence in people with down syndrome because of the increased production of beta-amyloid proteins on the 21 chromosome. The article continues to explain how four of the six human interferon receptors, which are encoded by genes that were identified in their research, are found on the 21 chromosome. Most people only have two copies of these genes, however because people with down syndrome have 3 copies of the 21 chromosome, they have an extra pair of this gene, and this leads to the overproduction of interferon receptors.

The researchers wanted to figure out whether or not this extra pair of genes are contributors to the features of down syndrome, so they tested this along with the other couple hundred genes found on this chromosome. To do this, they used CRISPR gene editing which included them reducing the number of receptor genes from three, found on people with down syndrome, to two, found on people without down syndrome, despite them decreasing the number of this gene from three to two, they leave all the other genes at the 'normal' amount for people with down syndrome, which is three. After conducting this experiment on mice, they found that correcting the number of these receptor genes significantly reduced abnormal gene expressions of multiple tissue types. Along with that, the immune responses, memory, ability to learn tasks, skull and facial morphology all were more regulated and 'normal'. This data concludes that the additional pair of interferon receptor genes do play a role in key traits of down syndrome.

In summary, the researchers found that the additional pair of interferon receptor genes found on chromosome 21, which leads to hyperactivity of immune responses, do play a role in some of the key features of down syndrome. The supporting article I chose, infections and immunodeficiency in Down Syndrome, helped support the article by also stating that genes along with other conditions that are located on the chromosome 21 or are linked to that chromosome become more prominent by adding a whole extra pair or set of it because there is a third chromosome 21. This review article also expands on the topic covered in the news article by giving more examples of different genes that are attached to the 21 chromosome and also behave in similar, if not the same, ways.

References

Espinosa, J. Several Down syndrome features may be linked to a hyperactive antiviral immune response – new research. *The Conversation*, (2023).

Ram, G. Chinen J. Infections and immunodeficiency in Down syndrome. *Clinical & Experimental Immunology* **164**, 9-16 (2011).