Carpe Diem – Seize the Day Blog

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Epilepsy is one of the most common neurological disorders in the world. It can affect people of any age, and it has a wide range of causes. It also has no cure currently, although there are many treatments that can help to treat epilepsy, including medications and surgical procedures. Even the causes of epilepsy are not fully known.

Recently, researchers from the University of Queensland who were working on learning more about the function and capabilities of brain cells discovered something interesting. They found a genetic mutation that can cause epilepsy. This is a rare mutation, and it is an interesting and exciting discovery. Just how rare is it? At this point, there is only one reported case on the planet. Even though it is rare, it can provide valuable information to researchers.

According to Dr. Victor Anggono, from the Queensland Brain Institute at the University of Queensland, the team was trying to get a better understanding of how nerve cells communicate with one another. This communication is naturally essential in the way that a normal brain functions. They were not expecting to find anything like the mutation, as they were studying brain receptors and were not specifically looking for mutations or any new causes of epilepsy.

Dr. Anggono said, "We're both excited and astounded to make such an important contribution to the field of cellular and molecular neuroscience." The team published their research in Cell Reports in 2021.

The team found the mutation because they were working with receptors in the grain. The receptors are protein structures that are attached to cell surfaces. The mutation causes receptors to function differently than they would in a typical brain. This resulted in an "imbalance in brain cell communication".

When there is an imbalance such as this, disorders can occur. Too much activity in the brain is associated with epilepsy, as well as unwanted cell death. When there is not enough activity in the brain, it can cause other problems, such as problems with memory and learning.

If the brain cell communications are not in balance, it's believed that it can affect a range of different types of neurological conditions. This includes autism spectrum disorders and Alzheimer's disease.

The doctor said, "There are also many examples of other mutations in the same gene that are known to be associated with epilepsy. What we know is that this receptor is critical for brain function and can lead to epilepsy when its function is mis-regulated."

The discovery of this mutation, although it is rare, is exciting for those who are working in the field of epilepsy research. Each new piece they get to the puzzle, the better. It can lead to further research to help better understand and treat similar mutations. It may be possible to eventually develop personalized medicines that can target the mutation. He went on to say, "Receptor blockers which have been approved by the US Food and Drug Administration (FDA) are already available for human treatment, but the challenge is to find the right ones and see how patients respond."

Because there is still so much to learn about epilepsy, all the different pieces of evidence that are found can help researchers in the future make more discoveries. They can help to change the lives of those who are living with epilepsy. Having a better idea of the cause of epilepsy can help to make it easier to determine what types of treatments may work. This could include medications, surgical procedures, implants, etc.

Editor's Note: The Carpe Diem – Seize the Day Blog will be distributed and posted weekly.
Always remember – **CARPE DIEM – SEIZE THE DAY!**

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