



Breakthrough discovery finds specific protein linked to intellectual disability and depression

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A research team led by Queen's, in collaboration with the Center for Regenerative Therapies, Dresden, has found how a specific protein plays a crucial role in the generation of neurons at a specific time and location during brain development.



They discovered how its disruption can lead to intellectual disability and depression in adults.

The findings have been published today (Thursday 20 August) in *Genes & Development* (<http://genesdev.cshlp.org/content/early/2020/08/20/gad.333906.119.abstract>).

It is expected this breakthrough will have a major impact on our fundamental understanding of brain development and lead to earlier diagnosis and better treatments for people with certain brain disorders.

Brain development is a highly complex process that involves generating various types of cells at defined time points and locations during embryonic development. Any kind of interference during these processes is known to cause diseases. Among these brain cell types, neurons are the working unit of the brain, designed to transmit information

to other nerve cells and various tissues in the body, such as the muscles as well as storage of memory in our brain.

While the field has rapidly advanced, the mechanisms creating the birth of neurons from their mother cells, called neural stem cells, in time and space during development has not been well understood, until now.

To conduct their study, the researchers looked at brain samples to closely determine the development of various cell types within the brain.

The study showed how the presence of a specific protein (called Phf21b), within a defined time window of brain development and in a specific location in the brain, signals the birth of neurons from neural stem cells in the right place and at the right time.

The researchers found that removal of protein Phf21b stopped the production of neurons from neural stem cells and led to severe defects in brain development. The researchers also found the importance of this protein, in particular in the folding of DNA in cells going on to form neurons.

Dr Vijay Tiwari, Reader at **[Wellcome-Wolfson Institute for Experimental Medicine at Queen's University Belfast](https://www.qub.ac.uk/research-centres/wwiem/)** (<https://www.qub.ac.uk/research-centres/wwiem/>) and corresponding author on the study, said: "Our study reveals the key role this protein plays during the birth of probably one of the most important cells in our body - the neuron.

"Understanding how a cell type in the brain is born at a specific point and in a specific place during development is crucial in our understanding how neurological issues arise later in life. We hope this discovery will pave the way for earlier diagnosis, earlier interventions and better treatment for people with a brain disorder, such as depression."

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