Vienna, 31 May 2022

More than 87,000 people in Austria living with autism
How modern genetic research is aiming to improve treatment in future

In many cases, the causes of neurodevelopmental disorders such as autism, epilepsy and mental disabilities are genetic. With the help of genetic models based on mouse models as well as stem cells, detailed research is being conducted into 15 special genes and the brain mechanisms behind them, with a view to developing new therapies. The research centring on the BCKDK gene proved to be a success story. This and other scientific breakthroughs in the field of human genetics will be under discussion at the 2022 European Society of Human Genetics (ESHG) Conference from 11 to 16 June at the Austria Center Vienna.

“Usually, neurodevelopmental disorders such as autism, epilepsy and mental disabilities are not discovered until children miss specific milestones in their cognitive development – such as learning to walk or talk – entirely or only reach them with a significant delay. In many cases, the causes are genetic. And this is exactly where we start with our research to find out which genes are responsible for neurodevelopmental disorders. It is only once we know what exactly is happening in the brain and we understand the mechanisms that take place there that individual therapies can be developed as personalised treatments, which will also help to bring about a significant improvement in the children’s cognitive functioning,” explained Prof. Gaia Novarino, Vice President of Science Education at the Institute of Science and Technology Austria (ISTA) and keynote speaker at the ESHG 2022 Conference.

Success story - the BCKDK gene: from initial discovery to nutrition therapy

A major research breakthrough in the field of neurodevelopmental disorders was the discovery of the BCKDK gene, which can be found on the sixteenth chromosome. BCKDK is short for **b**ranched **c**hain **k**etoacid **d**ehydrogenase **k**inase. “This gene can cause an amino acid metabolism disorder which subsequently leads to neurodevelopmental disorders. Specifically, the BCKDK gene normally encodes an enzyme that is responsible for regulating specific amino acids and which builds up proteins. People with the BCKDK genetic anomaly break down these particular amino acids more,” Novarino confirmed. In the case of BCKDK, a simple and effective therapy was identified that involved increased intake of these special amino acids through nutrition or infusion, which significantly improved the cognitive functions of children affected by this form of autism and epilepsy.

Basic research into 15 genes using mini brains

Research into the causes of neurodevelopmental disorders and the development of therapies to treat them continues to prove challenging in many cases, as both causes and forms

Internationales Amtssitz- und Konferenzzentrum Wien, AG
T +43-1-26069-0 I F +43-1-26069-303
Handelsgericht Wien, Firmenbuch Nr: FN 102030 w
A-1220 Wien I Bruno-Kreisky-Platz 1
office@acv.at I www.acv.at
ATU 15663100, DVR-Nr. 0484865

can vary hugely from one case to the next. “As a result, we focus on smaller groups of very similar cases. At present, in my research group we are looking at 15 different genes which are also representative of various gene types. Some genes regulate proteins, while others are responsible for transcription – in other words, the process governing how a segment of a gene’s DNA is copied before being translated into a protein.” Another focus of the research is not just to look at the gene in isolation, but to understand what it does on various different levels – molecular, cellular, in neurological networks and in the brain. To help achieve this, researchers are using genetic models from mice and stem cells to create neurons and mini brains that show how the human brain works. “Since genetic research is such a new science, it is extremely difficult to say when findings might eventually lead to specific therapeutic applications,” said Novarino.

Genetic research the key to personalised medicine

“It is only when we really understand what is happening with the genes and what is going on in the brain that we can develop effective treatments as we take the next step towards precision medicine,” Novarino noted. This is particularly important as many development disorders of the nervous system are resistant to treatment. While some neurodevelopmental disorders such as various types of epilepsy can be treated with drugs, getting the dose right still involves an amount of trial and error and they often end up suppressing other brain activities. Novarino hopes that genetic research will help to bring about significant improvements in treatment outcomes. After all, close to 2 in 100 children in Austria have autism. The Dachverband der österreichischen Autistenhilfe an umbrella organisation providing support for autism sufferers, puts the total number of people living with an autism spectrum disorder in Austria even higher at 87,000. An additional 80,000 Austrians are estimated to suffer from epilepsy.

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Contact

IAKW-AG – Austria Center Vienna
Claudia Reis
Deputy Press Officer
Tel: + 43-676-3199523
Mail: claudia.reis@acv.at