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A study by Prof Goh and Prof Augustine has revealed that choline can help override physical and functional defects caused by mutations in nerve cells affected by Rett syndrome. PHOTO: DON WONG FOR THE STRAITS TIMES

Vitamin choline offers hope in search for Rett syndrome cure

Choline – a vitamin found in food such as broccoli, soya beans, seafood and chicken – could be the answer to curing Rett syndrome.

This neurological disorder is the second most common cause of intellectual disability in girls, ranking only behind Down syndrome.

In a new study, when choline was applied to a dish containing nerve cells, or neurons, with Rett syndrome, it helped override physical and functional defects caused by genetic cell mutations, leading to an almost complete recovery.

The research is at an early stage, and scientists plan to run further studies using three-dimensional (3D) models, such as lab-grown organs, which are more similar to the structure of the human body.

There is currently no cure for Rett syndrome, but the study's lead, Assistant Professor Eyleen Goh of the Duke-NUS Medical School, believes the findings could pave the way for the development of such a cure.

"One possibility is that pregnant women could be given choline supplements, similar to the folic acid supplements given to pregnant women to reduce birth defects," said Prof Goh, who is also with the National Neuroscience Research Institute Singapore.

She worked on the three-year study with Professor George Augustine of the Lee Kong Chian School of Medicine at Nanyang Technological University. The study was published in the science journal NeuroMolecular Medicine in July.

Rett syndrome is caused by a spontaneous mutation in the X chromosome, and affects the growth and development of neurons. It affects about one in 10,000 newborn girls, while male foetuses who develop this mutation often die before birth. Girls have two copies of the X chromosome, but boys have only one, which means they do not have a "backup" copy.

For those with the syndrome, symptoms include slowing development, loss of purposeful use of the hands, slowed brain and head growth, problems with walking and seizures. These issues are often treated with medication, including drugs to control seizures, and occupational therapy.

Neurons control our every thought, word, action and movement. They fire electrical signals and transmit information – what we see and hear, for instance – through the synapses that connect them.

Rett neurons are about 10-20 per cent smaller, and their cell membranes have a different lipid (fat) composition, which affects synaptic transmissions.

When Rett neurons were supplemented with choline, their shape and structure improved. Choline also alleviated synaptic transmission defects linked to the condition. Earlier studies had shown how choline could protect heart and brain functions and prevent cholesterol and fat from accumulating in the liver.

"We wanted to find out how nutrition affects neuron growth and brain development – it is a huge breakthrough, but the next step is to test this on a 3D-culture," said Prof Augustine.

Audrey Tan

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