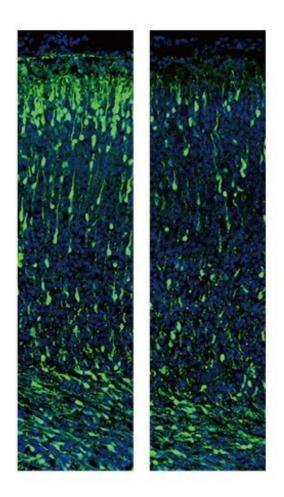


Researchers explore role of CHAMP1 gene in neuronal development

August 30 2022



Immunofluorescence images of fetal mice brain sections show a delay in the migration of neural cells (green) towards the surface of the cerebral cortex (upper) when expression of CHAMP1 is suppressed (right) compared with a normal condition (left). Credit: Nagai et al.



Intellectual disability affects roughly 2–3% of the world's population. Scientists understand the genes related to intellectual disability, but how the mutations of these genes cause this developmental disorder remains largely unknown. As a result, effective therapeutic interventions are lacking.

Now, a research group led by two Tohoku University researchers has uncovered the role of CHAMP1—a gene whose <u>mutations</u> are associated with intellectual disability—in neuronal development.

Details of their research were published in the journal *Brain Communications* on August 30, 2022.

Kozo Tanaka from the Institute of Development, Aging and Cancer, and Noriko Osumi from the Graduate School of Medicine and their group studied mice with the CHAMP1 gene "knocked out," where one or both of the genes from father and mother were inactivated.

When both of the CHAMP1 genes were deleted, mice died soon after birth. And despite no apparent brain structure abnormalities, the researchers found that neural cell migration towards the brain surface was delayed, indicating a delay in neuronal development.

When a single CHAMP1 gene was lost, mice demonstrated slight memory defects, impaired social skills, and depression-like behavior. These mild behavioral problems resembled some aspects of individuals with CHAMP1 mutations.

While many genes are related to intellectual disability, CHAMP1 is one of the most commonly unearthed mutations, with more than 100 cases worldwide.

"We need further research of CHAMP1 so we can understand how



intellectual disability occurs, and to ultimately find a way to treat it," said Tanaka.

More information: Masayoshi Nagai et al, Deficiency of CHAMP1, a gene related to intellectual disability, causes impaired neuronal development and a mild behavioral phenotype, *Brain Communications* (2022). DOI: 10.1093/braincomms/fcac220, academic.oup.com/braincomms/ad ... omms/fcac220/6678139

Provided by Tohoku University

Citation: Researchers explore role of CHAMP1 gene in neuronal development (2022, August 30) retrieved 2 May 2024 from

https://medicalxpress.com/news/2022-08-explore-role-champ1-gene-neuronal.html

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