

## Speaker

# Prof. Jin-Wu Tsai (蔡金吾)

Vice President of Research and Development

Distinguished Professor, Institute of Brain Science,

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National Yang Ming Chiao Tung University (NYCU)

## Title

Exploring Novel Genes and Mechanisms in Neurodevelopment and Disorders **Date & Time** 

## February 8, 2024 (Thu) 17:00-18:30 (JST)

#### Venue

Middle Auditorium, Clinical Lecture Building [A21] 2F, Seiryo Campus [MAP] https://www.tohoku.ac.jp/map/en/?f=SR\_A21

Format Hybrid (On-site & Online)

Registration Refer to the NGP Office

•Neuro Global プログラム生 (Neuro Global Program Students)

【脳科学セミナーシリーズEx】/【先進脳科学セミナーシリーズEx】セミナー 1ポイント

[Brain Science Seminar Series Ex] / [Advanced brain science seminar series Ex] 1 point

●医学系研究科(Graduate School of Medicine)

【医学履修課程】国際交流セミナー (アドバンスド講義科目)」 出席1回分

[Medical Science Doctoral Course] International Interchange Seminar (Advanced Lecture course) 1 attendance

●生命科学研究科(Graduate School of Life Sciences)

【単位認定セミナー】 【イノベーションセミナー(留学生対象)】 2ポイント

[Credit-granted seminar] [Innovation seminar (For international students)] 2 points

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## NEURO GLOBAL Seminar

## Exploring Novel Genes and Mechanisms in Neurodevelopment and Disorders Abstract

Recent advancements in understanding malformations of cortical development (MCDs), including microcephaly, lissencephaly, and focal cortical dysplasia, reveal significant progress in deciphering their pathogenetic mechanisms. Genetic variations and somatic mutations in key neurogenesis and neuronal migration genes provide crucial insights into our understanding for brain development. Novel genetic variants associated with lissencephaly, disrupting neuronal migration, were identified, emphasizing the complex relationship between abnormal cortical development and cognitive functions. Previously, we used a novel genetic screening paradigm based on transposon-mediated mutagenesis and identified dozens of candidate genes for MCDs, showcasing their potential clinical relevance. Additionally, a study on nuclear translocation during neuronal migration highlighted a BICD2 variant's role in lissencephaly, uncovering impaired nuclear translocation as a significant pathomechanism. Identification of CEP85L variants in posteriorpredominant lissencephaly underscores the centrosome's key role in this malformation. Our studies further elucidated the distinct roles of NDE1 and NDEL1 in nucleokinesis and MCD pathogenesis through the identification of a somatic mosaic NDEL1 variant associated with pachygyria and subcortical band heterotopia. These findings collectively contribute to a comprehensive understanding of cortical malformations, offering potential diagnostic and therapeutic avenues. The talk will discuss recent progress, presenting multiple pathogenic mechanisms elucidated in these disorders.

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Title