令和 6 年度　第 66 回　大学院セミナー

令和 7 年 1 月 7 日

分 野 名 Area of Research (責任者名)(内線)	医科薬理学　分野 責任者名(　有賀　純　)　　内線(　7043　)
演　　題 Title	**The role of the glycine receptor α2 subunit in neurodevelopmental disorders**
講 師 等 Presenter	**Sean Fraser** *School of Health, University of the Sunshine Coast, Maroochydore, Australia.* *National PTSD Research Centre, Thompson Institute, University of the Sunshine Coast, Birtinya, Australia.*
概要 Abstract	Neurodevelopmental disorders (NDDs) featuring developmental delay (DD), intellectual disability (ID) or autism spectrum disorder (ASD) pose a significant challenge to public health, due to their lifelong nature, high management costs, prevalence and recurrence within families. Glycine receptors (GlyRs) are ligand-gated ion channels that have key roles in various neurological processes, with the GlyR α2 subunit in particular governing cell fate, neuronal migration and synaptogenesis in the developing cortex. Mutations within the X-linked gene encoding GlyR α2 (*GLRA2*) are associated with NDDs, with affected probands exhibiting substantial clinical heterogeneity (ASD, DD, ID, and/or epileptic encephalopathy (EE), with language delay, microcephaly or macrocephaly). To understand why GlyR α2 dysfunction causes different clinical presentations, it is essential to understand exactly how mutations affect GlyR function. Additionally, individual neurotransmitter receptors do not operate as 'stand-alone' units but rather associate with other proteins which influence synaptic localisation, homeostasis, signalling pathways and receptor function. Significant attention has been directed towards understanding the protein interactome of other neurotransmitter receptors, such as AMPA and $GABA_A$ receptors. Alternatively, little is known about the GlyR proteome and the physiological roles that these interactors mediate. In order to address these questions, my research endeavours to uncover the full spectrum of pathomechanisms associated with GlyR mutations, allowing correlation of clinical presentations with changes in GlyR function, as well as improving our understanding of the proteomic mechanisms which govern glycinergic signalling. The desired translational outcomes of this project are to gain insight into how to therapeutically target defective GlyRs, so offering a route towards personalised medicines, in addition to elucidating novel genetic causes of NDDs.
開 催 日 時 Date and Time	令和 7 年　1 月　9 日(木) 17:00　～　18:00
開 催 方 法 Online/Face to face	医学部基礎棟1階　第4セミナー室　対面 **4thSeminar room Basic Medical Science Building 1F**
備　　考 Notes	

■先端医療科学特論(基礎編)　　□先端医療科学特論(臨床編)
□先端新興感染症病態制御学特論　　□先端放射線医療科学特論
□日本語(Japanese)　　■英語(English)
■対面(Face to face)　　□オンライン(Online)