Center for Neuroscience, ART

第50回脳神経科学コアセンターセミナーのお知らせ

日時　2017年3月6日（月）17:00−18:30

会場　医学部第2セミナー室（医学部仮設校舎2階）

演題　22q11.2染色体数変異 – 統合失調症と自閉スペクトラム症に関与する遺伝子、行動表現型の解体 –
(Deconstructing 22q11.2 copy number variants into dimensions of schizophrenia and autism spectrum disorder)

演者　廣井　昇　教授 (Prof. Noboru Hiroi)
Albert Einstein College of Medicine
Department of Psychiatry and Behavioral Sciences
and Department of Neuroscience

Many genetic variants with association with neuropsychiatric disorders are now known, and they have been recapitulated in genetic mouse models. Because of their exceptional degrees of association with schizophrenia, autism spectrum disorder (ASD) and intellectual disability (ID), copy number variants (CNVs), a few hundred kilobase to megabase hemizygous deletion and duplication of the human chromosomes, have emerged as promising entry points to delve into neuronal and cellular mechanisms underlying neuropsychiatric disorders. Many mouse models of CNVs have been--and are being-- developed since 2007. However, the robust association of 22q11.2 CNVs with schizophrenia, ASD and ID has been known since 1992, 2002 and 1998, respectively, and a number of 22q11.2 CNV mouse models have been analyzed in detail. Our group has identified the transcription factor *Tbx1* as a driver 22q11.2 gene for dimensional elements of schizophrenia, ASD and ID in mouse models. Consistent with this observation, several studies have reported individuals with *TBX1* mutations and ASD and ID diagnosis. I will specifically highlight ASD-related phenotypes of *Tbx1* heterozygous mice to illustrate a lack of individual variability and its impact on social communication between neonatal pups and mothers. Several issues have emerged from mouse models of 22q11.2 and other CNVs, including their reproducibility and genuine relevance of behavioral and neuronal phenotypes to human psychiatric disorders. I will illustrate pitfalls of CNV mouse models and strategies to circumvent them.

連絡先：発生発達神経科学分野 大隅 典子（内線8203）