第４６３回 難研セミナー
第３６回 難治疾患共同研究拠点セミナー

下記により難研セミナーを開催しますので、多数御来聴下さい。

記

日　時：平成２３年１２月１４日（水）13:00～14:30

場　所：M&D タワー 22 階北側　難治疾患研究所セミナー室

演　者：**Alysson Renato Muotri, Ph.D.**
(Assistant Professor　University of California San Diego)

演　題：**"Shared molecular pathways in human neurons derived from autistic patients"**

要　旨：Autism spectrum disorders (ASD) are complex neurodevelopmental diseases in which different combinations of genetic mutations may contribute to the phenotype. Using Rett syndrome (RTT) as an ASD genetic model, we developed a culture system using induced pluripotent stem cells (iPSCs) from RTT patients fibroblasts. Neurons derived from RTT-iPSCs had fewer synapses, reduced spine density, smaller soma size, altered calcium signaling and electrophysiological defects when compared to controls (Marchetto et al, Cell 2010). Finally, RTT-iPSC differentiated into neural precursor cells supported an increase amount of somatic mutations caused by de novo L1 insertions (Muotri et al, Nature 2010). We also observed defects in a subset of ASD neurons. These data suggest that perturbation in common molecular pathways can lead to neuronal alterations involved in the etiology of ASDs. The overlap phenotypes observed in the iPSC-derived human neurons between ASD patients provide evidence of an unexplored developmental window, before disease onset. Our model recapitulates early stages of a human neurodevelopmental disease and represents a promising cellular tool for drug screening, diagnosis and personalized treatment.

連絡先：　エピジェネティクス分野　石野　（内線 4863　）

共催：　幹細胞制御