Clarifying molecular pathogenesis of mental disorders based on rare disease-susceptibility variants

We identified rare disease-susceptibility variants, such as copy number variants (CNVs) and single nucleotide variants (SNVs) with high effect size, including 22q11.2del and 3q29del for schizophrenia, PCDH15del for bipolar disorder, and SNV in MECP2 for Rett syndrome associated with autistic spectrum disorder. Now we are trying to elucidate the pathogenesis of these mental disorders by analyzing iPS cells established from patients with rare variants, as well as by generating animal models with these rare variants through the genome editing technology. One of the main symptoms in mental disorders is sleep disturbance; in particular, psychiatric patients with rare variants such as 22q11.2del and SNV in MeCp2 tend to show sleep problems. We should pay attention to sleep as a phenotype because we can directly compare sleep in animal models with abnormal sleep in patients.

Dr. Norio Ozaki
Department of Psychiatry, Nagoya University Graduate School of Medicine

Date: Thursday, January 25, 2018
Time: 14:00 – 15:00
Venue: 1F Auditorium, IIIS Building