

Psychiatry and Neuroscience Seminar Series 2023



Pr Ype ELGERSMA

(Host H Rebholz)

Erasmus MC University, Rotterdam, Netherlands

Mechanisms underlying Angelman syndrome and the promise of antisense oligonucleotide (ASO) treatment

Friday, April 7th, 2023, noon

Room D Levy, 102-108 rue de la santé - 75014 Paris & VISIOCONFERENCE

Pr Ype ELGERSMA

**Department of Neuroscience, Erasmus MC University Medical Center, Molecular and cellular mechanisms
underlying neurodevelopmental disorders, Rotterdam, Netherlands**

My lab seeks to get insight in the molecular and cellular mechanisms underlying these disorders, with the ultimate goal to develop treatments. Our research is divided into three research lines: (1) Improving genetic diagnosis, (2) Understanding the mechanisms underlying neurodevelopmental disorders (3) Translational studies (i.e. clinical trials) to improve the quality of life of the affected individuals. (1) To improve genetic diagnosis, we have developed together with van Woerden lab a functional genomics screen (PRiSM) to rapidly determine if a genetic variant is pathogenic. This screen is not only important for providing a diagnosis, but also allows us to get more insight in the genes underlying neurodevelopment. (2) To get more insight in the pathophysiology of neurodevelopmental disorders, we typically make use of genetically engineered mouse models. Mouse models are analyzed at the biochemical, cellular (electrophysiological) and behavioral level. By analyzing the mouse models at all these levels we try to understand the specific function of these genes and proteins in brain development and learning and memory. Besides mouse models, we are currently also exploring the value of iPS cells to study these disorders. The genes and proteins that we specifically focus on are proteins associated with the RAS-ERK-MTOR signaling pathway and the proteasome. (3) To translate our findings to the patients, we are part of the ENCORE expertise center for neurodevelopmental disorders, I am the scientific director. ENCORE has large expertise outpatient clinic for Angelman Syndrome, Neurofibromatosis, Tuberous Sclerosis, Fragile X and autism.

Keywords:

Neurobiology

Cellular Neuroscience

Molecular Neuroscience

Behavioral Neuroscience

Synaptic Plasticity

Electrophysiology

Brain Diseases

Behavioral Testing

Learning and Memory

Neurobiology and Brain Physiology

ZOOM Meeting ID/ 832 4821 2814 Passcode: 202304

LINK: <https://u-paris.zoom.us/j/83248212814?pwd=M3dQM21mL2gzS2ZmK0FPQmpueFo5dz09>

Stay tuned