



CHILD NEUROLOGY SOCIETY

## WEBINAR



### Why Care About Molecular Mechanisms in Neurogenetics

**Tuesday, August 8, 2023**

6:00 pm CST

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Genetic testing is a powerful tool to reach diagnoses in children with neurological disorders. However, the name of the affected gene is only part of the information on a positive test result. Can details about the patient's specific variant also be useful? Dr. Yano will discuss some ways research data on molecular mechanisms of neurogenetic disease can assist the clinician's judgment during diagnosis, prognostication, and treatment. Foundational concepts such as genotype-phenotype correlation and gain/loss-of-function will be illustrated using examples drawn from his research on sodium-potassium ATPase disorders (e.g. alternating hemiplegia of childhood) and from the literature on other neurogenetic diseases.

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#### Organizer

CNS Research Committee

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#### Speaker

**Sho Yano, MD, PhD**



Assistant Professor  
Section of Pediatric Neurology, Department of Pediatrics  
University of Chicago

Sho Yano is a child neurologist and medical geneticist who conducts translational research on disorders of ion transport. His primary research focus is sodium-potassium ATPase disorders, which include a growing spectrum of epileptic, motor, and neurodevelopmental phenotypes such as alternating hemiplegia of childhood. Using biochemical and electrophysiological approaches such as two-electrode voltage clamp, he searches for mechanisms of pathogenesis that could inform therapeutics development and prognostication in these conditions. His other activities in the field include participating in the medical advisory board of the Alternating Hemiplegia of Childhood Foundation and being an associate editor for a medical genetics journal, *Molecular Genetics and Genomic Medicine*.

**Register  
Now**

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***NOTE: CNS live-streamed webinars will be posted on the CNS website ("Craft" section) within two weeks following original presentation.***

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