

GENETICS, BIOINFORMATICS, AND SYSTEMS BIOLOGY COLLOQUIUM

THURSDAY SEPTEMBER 30

12:00PM PST

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PASSCODE: GENETICS

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ETHAN GOLDBERG, MD
THE CHILDREN'S HOSPITAL OF PHILADELPHIA
U.PENN SCHOOL OF MEDICINE.....

“AN EPILEPSY NEUROGENETICS INITIATIVE: FROM DIAGNOSIS TOWARDS MECHANISMS AND THERAPY”

Epilepsy is a severe neurological disorder associated with seizures as well as non-seizure comorbidities including developmental delay, intellectual disability, and increased mortality. Epilepsy-associated cognitive impairment is not amenable to treatment, and even seizures remain resistant to therapy in a significant proportion of cases. In children, at least half of epilepsy has a genetic basis, and establishing a definitive genetic diagnosis can be used as leverage towards the development of new treatment approaches. In this talk, we discuss the current landscape of epilepsy neurogenetics and a new clinical initiative aimed at providing access to genetic testing for all children with epilepsy. We then discuss novel insights into disease pathomechanisms in advanced experimental model systems with a focus on Dravet syndrome (DS), the canonical genetic epilepsy, due to heterozygous pathogenic variants in the sodium channel gene *SCN1A*. We use mouse genetics, bioinformatics, electrophysiology, pharmacology, optogenetics, and in vivo two-photon calcium imaging in awake, behaving *Scn1a*^{+/-} mice in vivo to identify and manipulate key cells and circuits to dissect the mechanistic basis of key DS endophenotypes.

Organization Committee: J. Gleeson, J. Sebat
GBSBC Seminar Coordinator: R. White

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