GENETICS, BIOINFORMATICS, AND SYSTEMS BIOLOGY COLLOQUIUM

THURSDAY SEPTEMBER 30
12:00PM PST
LIVE @ LEICHTAG & ON ZOOM!

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PASSWORD: GENETICS

PRESENTED BY: IGM Institute for Genomic Medicine | UC San Diego Bioinformatics & Systems Biology

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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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