

A KJELDGAARD LECTURE



Thursday 2 May 2019 at 13:15

1593-012 (iNANO)

Same location for the PhD session



Evan E. Eichler

University of Washington, Seattle, WA

New mutations, pathways and the genetic subtypes of autism

Autism is a neurodevelopmental disorder that affects approximately 1% of children and is characterized by deficits in language, social interaction, and repetitive behaviors. I will summarize our findings regarding the discovery of genetic mutations and their contribution to autism spectrum disorder (ASD) and developmental delay (DD). I will present evidence from sequencing of more than 10,000 families with autism and show how “new” mutations that are present in children but not their parents have been used to pinpoint specific genes associated with ASD and DD. These data have been used to discover rare, inherited forms of autism but also, more broadly, the genetic basis of the disorder. The results strongly argue that the development of the human brain is particularly sensitive to the timing and expression of many different genes; several hundred genes are responsible but in each family a different gene is disrupted; the different genes share neurodevelopmental functions related to long-term potentiation, chromatin remodeling, synaptic function, and Wnt signaling; and that the maternal and paternal contributions differ significantly. I will present data on how grouping patients based on a specific genetic etiology can be used to predict clinical subtypes of autism and specific gene targets for modelling therapeutics. Next-generation exome and genome sequencing data provide a powerful path forward for understanding the genetic architecture of these diseases but the heterogeneity demands an unprecedented level of global cooperation and networking for characterizing and the development of therapies.

Host: Christian Kroun Damgaard, Gene Expression & Gene Medicine
Department of Molecular Biology and Genetics, Aarhus University

The lecture will be followed by a chalk-board session for PhD students