Edwin Cook, Jr., M.D.
MIND Institute Distinguished Lecturer Series – January 10, 2018

Biographical Information

Ed Cook attended Southern Methodist University and the University of Texas Medical Branch at Galveston. He trained in Adult and Child and Adolescent Psychiatry at the University of Chicago. After completion of training, he was promoted to the role of Professor of Psychiatry, Human Genetics and Pediatrics at the University of Chicago until his move to the University of Illinois at Chicago in 2005.

Ed is now the Director of Child and Adolescent Psychiatry and the Earl M. Bane Distinguished Professor of Psychiatry at the University of Illinois College of Medicine. His program of research focuses on collaborative molecular genetic studies of autism spectrum disorder, with an emphasis of studies of relationships between genotype and phenotype. He also focuses on study of the biomarker, hyperserotonemia, in autism. The goal of Dr. Cook’s research is the development of improved pharmacological treatments of autism. He has assessed and treated children, adolescents and adults with autism for over 30 years, including following many patients for over 25 years.

Presentation Abstract (4:30pm presentation)
Translating the complex genetics of autism

Evidence supporting the role of genetics in the multifactorial etiology of autism will be reviewed. There is evidence for both polygenic (small effects of many genetic variants) and larger effect and infrequent de novo (spontaneous) mutations involving single genes or segments of chromosomes with many genes. To further add complexity there is evidence that the polygenic risk that applies to most cases of autism also has an impact when there are larger effect de novo genetic findings.

Substantial progress has been made in identifying specific single gene and copy number variants in autism, with the pace of these findings accelerating over the past decade. Although more are predicted, at least 72 loci have been identified, including 66 affecting a single gene (e.g. CHD8, SCN2A) and 6 affecting the copy number of several genes (e.g. maternal chromosome 15q11-q13 duplication). Specific knowledge of such genetic events in 3-5% of individuals with autism may provide important information about risk for comorbidities (e.g. ADHD, epilepsy). Finally, study of these de novo genetic variants is providing information about complexity of alteration in brain development contributing to autism. Finally, such findings are guiding development of new therapeutics for autism and related comorbidities.