Judith H. Miles, M.D., Ph.D.

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Biographical Information

Judith H. Miles, M.D., Ph.D., graduated from Mount Holyoke College; received a Ph.D. in maize genetics from Indiana University with Marcus Rhodes; and then completed her M.D. and pediatric residency at the University of Missouri in Columbia, and a fellowship in medical genetics at UCLA with David Rimoin and Michael Kaback. She was the director of Medical Genetics at the University of Missouri until 2006. In 1995, she and Richard Hillman established the first Autism Clinic/Center in Missouri. This clinic/center had the dual purpose of providing a medical home for families with autism and pursuing research into "who gets better and why," with the ultimate goal of discovering effective treatments for each of the autism spectrum disorders. The Thompson Center for Autism and Neurodevelopmental Disorders morphed out of our original "self-proclaimed Center" thanks to a generous gift from the Thompson family. Dr. Miles served as the first William L. Thompson Chair. In addition to delineating precise physical and behavioral phenotypes and using them to sort out autism subgroups, Dr. Miles' research interests have included the provision of genetic services to rural areas, prenatal diagnosis and cancer cytogenetics.

Presentation Abstract

Delineation of Etiological Subgroups within the Autism Diagnosis

The transition of autism from a diagnostically inclusive, behaviorally-defined disorder to an etiology (i.e., cause) based classification system mirrors the history of medical genetics and mental retardation. Beginning in the 1960s, geneticists used physical dysmorphology, biochemical and other laboratory data to delineate the heterogeneity within multiple behaviorally-based phenotypes. To accomplish a similar shift for autism, we identified "phenotypic features" that are present in some but not all individuals with autism, are relatively discrete, quantifiable and pathophysiologically relevant. Morphologic (ex. dysmorphology, facial structure), physiologic (ex. pupillary light reflex), medical (autonomic nervous system) and family history all provide helpful biomarkers that distinguish subsets of autism. Proof that a biomarker splits out an etiologically distinct subset depends on showing differences in outcome measures, clinical course, response to therapy, genetic indicators (sex ratio, recurrence risk) and ultimately identification of separate genes or genetic pathways. I will describe phenotypic variables that may define autism subgroups with emphasis on physical dysmorphology, physiology and family history.