BASIC// DISCOVERY + CLINICAL// TRANSLATIONAL

TEAMING UP on AUTISM

AUTISM SPECTRUM DISORDER (ASD)—THE MOST COMMON neurodevelopment disorder of childhood—is characterized by challenges with social skills and communication, repetitive behaviors and sensory sensitivities.

The **Jefferson Autism Research Program** is comprised of a team investigating the molecular, genetic, synaptic and functional aspects of ASD. Program researchers and clinicians translate newly gained knowledge into treatments and behavioral interventions intended to improve the function and quality of life of those with ASD.

Roseann Schaaf, PhD, professor of occupational therapy, leads a multi-project study investigating ASD-associated sensory perception and integration difficulties. These often include hypersensitivity to sounds, sights and tactile sensations and trouble perceiving and integrating those sensory inputs—and can lead individuals with ASD to become overwhelmed and unresponsive. In addition, she is investigating whether occupational therapy designed to address ASD-related sensory challenges can create long-lasting improvements in everyday functional skills. Dr. Schaaf's lab is conducting a large, NIH-funded clinical trial of sensory integration interventions in collaboration with Albert Einstein Medical Center in New York. The project tracks both improvements in children's functional capacities and changes in their brains' neurological processing.

LEFT TO RIGHT: Matthew Dalva, PhD; Diane Merry, PhD; Roseann Schaaf, PhD; Judith Ross, MD

And she is developing and testing an assessment of sensory features in ASD, which should allow for more precise identification and treatment of an individual's sensory challenges.

ASD is more prevalent in males, but the reason is not clear. **Diane Merry, PhD**, professor of biochemistry and molecular biology, is working toward an answer by studying the basic biology of two proteins produced by sex-chromosome-linked genes. These proteins, which are critical to synapse formation and stabilization, have increased expression in boys with some types of ASD. For these studies, Dr. Merry is creating specialized lines of patient-derived stem cells, which can be turned into neurons in order to study the biochemical and functional characteristics of these synaptic proteins.

Mounting evidence indicates that ASD is a "synaptopathy"—a disease rooted in atypical function of the synapses through which brain neurons communicate. **Matthew Dalva, PhD**, professor of neuroscience, directs the Jefferson Synaptic Biology Center (SBC), which seeks to address how these building blocks of the brain form, function and are linked to disease. His laboratory uses leading-edge imaging and new molecular tools to study how synapses are made and lost, and what impact abnormal morphology and quantity of synapses have on brain function. Dr. Dalva's current studies address synaptic defects in ASD, but his work—and that of his SBC colleagues—will likely also shed light on a range of other neurological conditions including addiction, Alzheimer's and neurodegenerative disease.

Judith Ross, MD, professor of pediatrics, directs the Extraordinary Kids Clinic at Nemours/Alfred I. duPont Hospital for Children, which serves children with X and Y chromosome variations. She has more than 25 years of NIH-funded pediatric research experience focused on neurodevelopmental outcomes in children with X and Y chromosomal disorders such as XYY, Klinefelter and Turner syndromes. Her Jefferson lab applies that basic science expertise and her deep clinical experience to the challenge of uncovering ASD's genetic roots. ■