



Grounding Autism in the Basic Biomedical Sciences

The Nancy Lurie Marks Family Foundation (NLMFF) seeks to enhance the lives of people with autism by funding basic biomedical research, providing resources for advocacy and policy groups, and making available social services for families. We believe that access to improved medical diagnosis and treatment and to better means of communication is the surest path for autistic individuals to find increased enjoyment in social and family life, obtain meaningful employment, and discover ways of independent living and creative expression.

Achieving these goals means that autism must be approached from a fundamental scientific perspective. It is only when autism comes to be understood and defined in terms of off-kilter physiological systems and circuits, and not just their surface manifestations, such as self-injurious behaviors, seizures, tics, aggression, and frustratingly futile attempts at speech, that stigmatization and isolation will end. We believe that a proper description of autism will set the stage for powerful new technologies to be brought to bear on the difficult problems these children and adults face. Medical history has shown repeatedly that basic knowledge leads to practical invention, often in completely unexpected ways, but for this to happen in the case of autism the challenge needs to be expressed in terms of targetable physiological or molecular systems and processes, not just behaviors, failures to communicate, or socially awkward habits.

The principal goal of the scientific program at the NLMFF is to contribute to this deeper understanding of the biological basis of autism. The astonishing discoveries of the past few years in neuroscience allowing, for example, the electrical properties of individual neurons to be manipulated in living animals, enabling signaling properties of genotype-matched cells to be grown in culture, or revealing the subtle correlations in frequency coupling across brain regions as new tasks are learned, place demands on our scientific imaginations to probe the nature of autism more deeply than what appears on the surface in the form of behaviors.

Science is a conversation, across generations and disciplines, and for too long



autism research has been isolated. To help bring autism into the conversation of modern neuroscience, NLMFF conducts small workshops to which leading scientists, not necessarily engaged in autism research, are invited to share their insights with workers in the field. These meetings often inform our funding decisions and serve as an impetus for young scientists to enter into autism research.

Much of our funding at NLMFF is focused on genetics, synaptic chemistry, systems biology and the physiology of movement, all with the aim of exploiting the new insights of neuroscience to discover how behavior is organized, coordinated, and controlled in the human body, and how language is learned and speech produced. The scientific study of communication modalities, hand, eye, and ear, with the goal of producing new and useful devices and strategies for

social and intellectual engagement, is essential for developing support systems that could enhance the quality of life for many children and adults with autism by alleviating a principal source of distress. Autism is the 'Rosetta Stone of human neurobiology', presenting rich opportunities to study the neurobiology of speech, brain rhythmicity, and emotion, and explore the nature and potential of human thought and creativity as it evolves on a different, but no less endowed, developmental trajectory.

LURIE CENTER FOR AUTISM

Nancy Lurie Marks and the Nancy Lurie Marks Family Foundation recently made a gift to the Massachusetts General Hospital (MGH) to establish the Lurie Center for Autism, an integrated and multidisciplinary clinical, research, training and advocacy program dedicated to treating individuals with autism spectrum disorders and other developmental disorders across the lifespan. In addition to clinical care, cutting-edge translational research, advocacy and public policy analysis, the Center is committed to providing training and clinical experience for a new generation of medical doctors to meet the comprehensive needs of those with autism.

Persons with autism, like the rest of us, face new and different medical challenges as they age. Little is known about what health concerns might be unique to this population. Medicine has always advanced from the hunches and conjectures of physicians who see hundreds of patients, often for periods spanning many decades, and large clinics devoted to serving this population are needed.

To address these and many other questions, the Lurie Center will maintain comprehensive longitudinal biomedical databases, integrating clinical and research observations, enabling qualified researchers to assemble research cohorts with informed consent. Inherent in these databases may be patterns and subgroupings that will spark physician-researchers,

LURIE CENTER FOR AUTISM



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collaborating with bioinformatics specialists, to form new hypotheses and generate new ideas for treatments. Through referrals to specialists at the major clinical departments at MGH, as well as collaborations with basic scientists at the Martins Center for Biomedical Imaging and the Center for Human Genetics Research, the Lurie Center is in position to ground the study of autism in one of the most advanced biomedical treatment centers in the world.

In addition to its core services, the Center offers speech, occupational, and physical therapies to autistic individuals. The clinical program, specializing in diagnosis and referral, includes experts in pediatrics, adult internal medicine, gastrointestinal evaluation, augmentative communication, nutrition, audiology, and optometry. Large hospital systems can sometimes present daunting challenges for persons with autism just to get to the right place, compose oneself, and then try to communicate what's not feeling right. Families are assigned a dedicated 'Autism Resource Specialist', often a medical social worker, a person who will come to know them well, to help with the planning and coordination of their visits with clinicians, therapists, researchers and school counselors.

NANCY LURIE MARKS CLINICAL AND RESEARCH FELLOWSHIP PROGRAM IN AUTISM

If the field of autism is to be transformed by the discoveries of biomedical science in pediatrics, neurology, and psychiatry, in addition to the basic sciences of genetics, physiology, and structural biology, training in autism must become integrated into the curricula of medical schools. Physicians specializing in autism are in precious short supply. To help meet this need, and increase awareness of the difficulties autistic individuals may face in the clinic, The Nancy Lurie Marks Clinical and Research Fellowship Program in Autism has been established to support Harvard Medical School faculty and students interested in autism and related neurological disorders. Participants are selected from

all levels of the training program: medical students staying on for a fifth year, advanced postdoctoral scientists seeking clinical collaborations, and doctors who have completed their residency and are undertaking specialty fellowship training.

LURIE INSTITUTE FOR DISABILITY POLICY

We are also keenly interested in addressing progressive public policy and planning initiatives relevant to adults with autism including employment, residential planning, family and community supports, and the transition to more independent living. Through a grant to Brandeis University, the NLM Family Foundation helped establish the Lurie Institute for Disability Policy and endow a professorship at the Heller School for Social Policy and Management. The Lurie Institute for Disability Policy conducts research on disability policy in the United States with a special emphasis on autism, focusing on the lifespan of persons with disabilities and their families. The Institute explores how human services and public benefits are provided to persons with developmental disabilities and their families and also how persons with disabilities can become self-advocates. By offering courses on disability policy, the Institute fosters and develops careers in this field. The Institute also shapes public policy on issues related to autism and other disabilities as these issues affect children, the aging population, special education, health care, and transitions across the lifespan for persons with disabilities and their family caregivers.

AUTISM AROUND THE GLOBE

Autism around the Globe (www.autismaroundtheglobe.org) is a global public awareness project of the NLM Family Foundation. The goal of this project is to provide a platform for sharing stories from contributors around the world whose lives have in some way been affected by autism. These compelling narratives, often translated into multiple languages, provide insight into the cultural factors that affect those living

with autism in different parts of the world. There are many surprising and original observations on how autism is perceived in highly developed and populous areas as well as in remote regions of the world. Detailed descriptions of referral and diagnostic procedures in places where medical care is so different from that typically found in advanced countries vividly illustrates the universality and devastating impact of autism on daily life and the long-term prospects for adults living with this condition. Many of the authors of these articles are medical researchers or directors of caregiving agencies, who are passionately interested in joining the world-wide research effort to understand how autism arises and what can be done about it.

AN INVITATION TO AUTISM RESEARCH

The pace of discovery in autism research now matches that in many other areas of clinical neuroscience, including schizophrenia, addiction, Alzheimer's disease, and depression. In some cases, such as the convergence onto common synaptic mechanisms involved in learning and habit formation, the large number of implicated genes has, remarkably, led to simplifying and testable hypotheses at the systems level. The Nancy Lurie Marks Family Foundation encourages scientists, young and old, basic and applied, to take up the challenge of understanding autism and, in the process, discover how molecules, cells, and circuits give rise to learning, memory, and human communication.

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