Developmental Synaptopathies Consortium (DSC)

Research Areas

- Tool Development

At a Glance

- Status: Active Consortium
- Year Launched: 2014
- Initiating Organization: NIH Rare Disease Clinical Research Network
- Initiator Type: Government
- Rare disease
- Location: North America

Abstract

The Developmental Synaptopathies Consortium (DSC) is composed of a group of 10 medical centers studying three rare genetic syndromes that often cause autism spectrum disorder and intellectual disability (ASD/ID), which are severe neurodevelopmental conditions with early childhood onset. Advances in genetics have indicated that ASD/ID represent a spectrum of rare disorders. While both ASD and ID have a variety of known genetic causes, some of them have been shown to impair similar cellular pathways in the brain. The three conditions to be studied by the Consortium are Tuberous Sclerosis Complex (caused by mutations in the TSC1 and TSC2 genes), Phelan-McDermid Syndrome (caused by mutations in the SHANK3 gene), and PTEN Hamartoma Tumor Syndrome (caused by mutations in the PTEN gene). These three rare diseases seem to affect certain shared pathways influencing the development of brain connections, or synapses.

Structure & Governance

This consortium is part of the Rare Diseases Clinical Research Network (RDCRN), an initiative of the Office of Rare Diseases Research, National Center for Advancing Translational Sciences (NCATS).
The RDCRN is designed to advance medical research on rare diseases by providing support for clinical studies and facilitating collaboration, study enrollment, and data sharing. Through the RDCRN consortia, physician scientists and their multidisciplinary teams work together with patient advocacy groups to study more than 200 rare diseases at sites across the nation.

Funding and scientific oversight for the RDCRN are provided by NCATS and 10 National Institutes of Health components: Eunice Kennedy Shriver National Institute of Child Health and Human Development; National Cancer Institute; National Heart, Lung and Blood Institute; National Institute of Allergy and Infectious Diseases; National Institute of Arthritis and Musculoskeletal and Skin Diseases; National Institute of Dental and Craniofacial Research; National Institute of Diabetes and Digestive and Kidney Diseases; the National Institute of Mental Health; National Institute of Neurological Disorders and Stroke; and Office of the Director. In addition, patient advocacy groups provide funds for many of the projects.

Patent Engagement

The RDCRN DSC Contact Registry is a way for patients with developmental synaptopathies and their family members to learn about DSC research studies they may be able to join. Participation is completely voluntary, and participants can withdraw at any time. There is no cost to join the Contact Registry.

Impact/Accomplishment

Journal Articles


Links/Social Media Feed
Points of Contact

Rajna Filip-Dhima, M.S.
Project Manager
Boston Children’s Hospital
300 Longwood Avenue
Boston, MA 02115
email: Rajna.Filip-Dhima@childrens.harvard.edu

Sponsors & Partners

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