

Synaptopathies Clinic

at Kennedy Krieger Institute

Synaptopathies are neurodevelopmental disorders that result from changes in genes that function at the synapses, the points where brain cells, also called neurons, meet and “talk” with each other. These genes and their corresponding proteins play important roles in brain development and functioning. Changes, also called mutations, in these genes can cause a spectrum of neurodevelopmental disorders, including childhood-onset epilepsy, developmental delays, cognitive difficulties, repetitive and challenging behaviors, and social and communication difficulties. The symptoms a child experiences and the severity of the disorder can vary widely.

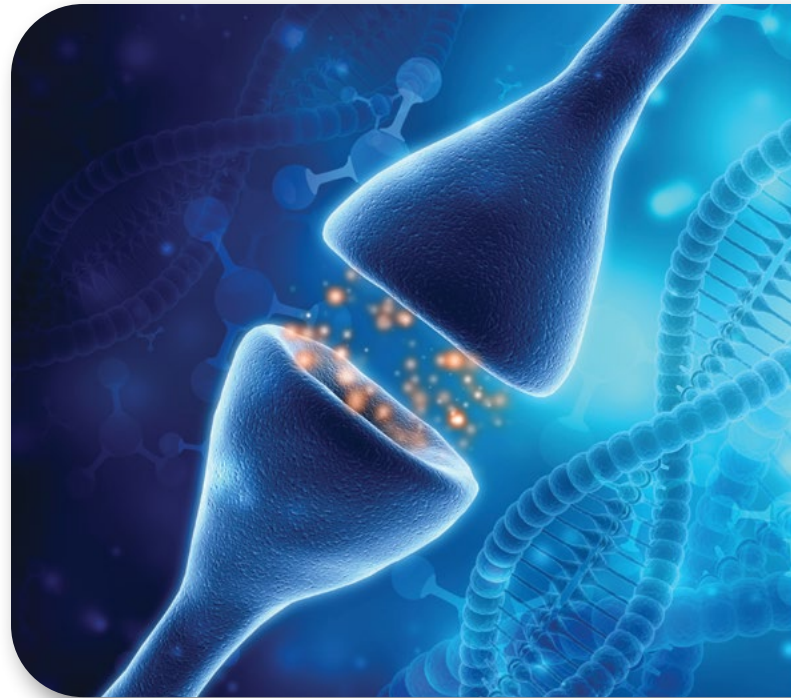
Who We Are

At Kennedy Krieger Institute’s Synaptopathies Clinic, an interdisciplinary team provides comprehensive evaluations and clinical care. It is our goal to help each of our patients, and their family members, gain a better understanding of their condition and access appropriate medical and therapeutic interventions to improve their overall health and quality of life.

Who We Serve

Kennedy Krieger provides expert care for children with developmental challenges related to changes in several synaptic genes (SYNGAP1, SHANK 3, GABRB, STXBP1, GRIN, GRIA, KCN, SCN and the SLC family of genes). Symptoms may include global developmental delays, intellectual disability, autism spectrum disorder, sensory processing difficulties, disruptive behaviors and epilepsy. The severity and onset of symptoms can vary among individuals, but symptoms appear by a child’s second birthday.

Symptoms alone cannot be used to diagnose synaptopathies. Genetic testing, which can be done at Kennedy Krieger, is required for diagnosis.



Our Team

Kennedy Krieger is home to a unique interdisciplinary collaboration of experts in synaptopathies. Our clinicians, research scientists and rehabilitation professionals work together to provide optimal interdisciplinary care for patients. Because synaptic dysfunction can affect many different systems in the body, treating a patient with one of these disorders often requires the collaboration of a wide range of medical specialists. Key specialists who see our patients include those working in the following fields:

- Genetics
- Epilepsy
- Neurology
- Neuropsychology
- Behavioral psychology
- Social work



Treatment Approach

Treatment is individualized and can include:

- Management of epilepsy, sleep, and gastrointestinal and feeding issues
- Support from a neuropsychologist or behavioral psychologist to manage cognitive delays and behavioral challenges
- Physical, occupational and speech therapies to manage developmental delays

Resources for Families

SynGAP Research Fund

syngapresearchfund.org/families/support/welcome-pack

SYNGAP1 Foundation

syngap1foundation.org

Phelan-McDermid Syndrome Foundation

pmsf.org/resource-library/

SCN Portal

scn-portal.broadinstitute.org

Contact Information

Synaptopathies Clinic at Kennedy Krieger Institute

For more information, contact the clinic at **CFSD@KennedyKrieger.org** or **443-923-2778**. To schedule an appointment, please call **443-923-9400**, or call toll-free at **888-554-2080**. TTY users, please contact us at **443-923-2645**, or dial **711** to make a Maryland Relay call.

KennedyKrieger.org

Physicians and Healthcare Professionals

To make a referral, email us at **CFSD@KennedyKrieger.org** or call our Physician Referral Line at **443-923-9403**.

Clinic Location

801 North Broadway
Baltimore, MD 21205

Mailing Address

707 North Broadway
Baltimore, MD 21205

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