

Barth Syndrome Clinic at Kennedy Krieger Institute

Who We Are

The Barth Syndrome Clinic at Kennedy Krieger Institute is an interdisciplinary clinic dedicated to the diagnosis and treatment of Barth syndrome. Barth syndrome is a rare X-linked genetic disorder caused by the deficiency of a complex lipid called cardiolipin. Because cardiolipin is the major phospholipid of mitochondria, the elements of cells that make energy, many systems in the body can be affected.

For more than 75 years, Kennedy Krieger has been helping patients with physical and cognitive disabilities. This expertise, combined with our newly expanded, modern facilities, offers our patients access to top experts in their fields and innovative rehabilitative services, all under one roof.

Who We Serve

We provide expert care for children and adults who have, or are suspected of having, Barth syndrome. Affecting only males, Barth syndrome can cause significant problems, including:

- Cardiomyopathy (abnormal heart muscle)
- Muscle weakness
- Chronic fatigue
- Growth delay and delayed puberty
- Neutropenia (low white blood cell count)

Our Team

Our clinic features a unique collaboration among experts in Barth syndrome from various disciplines. Our clinicians, research scientists and rehabilitation professionals work together to provide optimal interdisciplinary care for patients. Because Barth syndrome can affect many different systems in the body, treating a patient with Barth syndrome often requires the collaboration of a wide range of medical specialists. Key specialists who see our patients with Barth syndrome include those in the following fields:

- Metabolism
- Cardiology
- Hematology
- Genetic counseling
- Physical therapy
- Nutrition



Our Approach

Evaluation and Diagnosis

As a first step, we discuss the onset and variation in symptoms of Barth syndrome and review any previous genetic and metabolic testing. If a genetic diagnosis has not yet been made, a genetic counselor guides patients and their families through the diagnostic process.

We are all born with great potential.
Shouldn't we all have the chance to achieve it?

Treatment and Care

For patients with a confirmed diagnosis of Barth syndrome, we provide expertise on the various clinical concerns of the disorder and tailor a treatment plan to each patient's metabolic and physical needs. Patients will have the opportunity to meet with both a hematologist to address protection against infection, and a cardiologist to discuss cardiac management. An echocardiogram will also be performed to correlate current biochemical status with a patient's cardiac function. Our clinic can develop individualized nutrition and physical therapy plans to be carried out within the patient's home healthcare system. In addition, we can also facilitate access to Kennedy Krieger's comprehensive team of specialists to assist with specific educational, behavioral or other therapy needs.

Our Research Initiatives

Our laboratory and clinical research programs play an important role in the Barth Syndrome Clinic. Dedicated to finding and developing new treatments for this disorder, we led the first therapeutic clinical trial for Barth syndrome. We maintain strong collaborations with other academic centers interested in Barth syndrome, and with parent and patient advocacy groups throughout the U.S., the United Kingdom and Europe. We will continue to dedicate our research efforts toward these goals.

Contact Information

Barth Syndrome Clinic at Kennedy Krieger Institute

For more information or to schedule an appointment, please call **443-923-2783**, 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/BarthSyndrome

Physicians & Healthcare Professionals

To make a referral, please call our Physician Referral Line at **443-923-9403**.

Clinic Location

801 North Broadway
Baltimore, Maryland 21205
443-923-2783 (phone)
443-923-2781 (fax)

Mailing Address

707 North Broadway
Baltimore, Maryland 21205

The full interdisciplinary clinic is held on a quarterly basis. However, individual appointments with clinic physicians can be scheduled as needed.



“Without Kennedy Krieger and the Barth Syndrome Clinic medical team, I would be lost as to how to care for my son and his rare disorder. It is so important for me to understand how this illness affects my son, and the team takes the time to explain it to me in as much detail as I need.”

– Erin M., parent

Kennedy Krieger Institute recognizes and respects the rights of patients and their families and treats them with courtesy and dignity. Kennedy Krieger Institute provides care that preserves cultural, psychosocial, spiritual and personal values, beliefs and preferences. Care is free from discrimination based on age, race, ethnicity, religion, culture, language, physical or mental disability, socioeconomic status, sex, sexual orientation, and gender identity or expression, including transgender. We encourage patients and families to become active partners in their care by asking questions, seeking resources and advocating for the services and support they need. © 2019 Kennedy Krieger Institute 02/2019



Learn More. Get Involved. Stay Connected. Visit KennedyKrieger.org/Connect

