

Comprehensive Cardiac Care In **Friedreich's Ataxia**



The Children's Hospital of Philadelphia®



Penn Medicine

Founded in 1855, The Children's Hospital of Philadelphia is the birthplace of pediatric medicine in America. Throughout its history, a passionate spirit of innovation has driven this renowned institution to pursue scientific discovery, establish the highest standards of patient care, train future leaders in pediatrics, and advocate for children's health. A haven of hope for children and families worldwide, CHOP is a nonprofit charitable organization that relies on the generous support of its donors to continue to set the global standard for pediatric care.

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Friedreich's Ataxia Program

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The Penn Medicine/CHOP Friedreich's Ataxia Center of Excellence is supported by the Friedreich's Ataxia Research Alliance (FARA), in partnership with the Hamilton and Finneran families.

Discovering that you or a family member has received a diagnosis of FA can be frightening. Taking the appropriate steps to receive care is one of the best things you can do. The Children's Hospital of Philadelphia is part of a network of centers around the world devoted to clinical research in FA. Many new therapeutic approaches to FA are in various stages of development.

RIGHT PLACE TO BE



COMPREHENSIVE CARDIAC CARE IN FRIEDREICH'S ATAXIA

Friedreich's ataxia (FA) is a progressive neurogenetic condition found in approximately 1 in 50,000 people. While it is relatively rare, it is the most common form of inherited ataxia, a condition characterized by progressive lack of coordinated movement and loss of balance. FA also involves degeneration of heart muscle and nerve cells. Cardiac involvement in FA can include hypertrophic cardiomyopathy, where the heart muscle is abnormally thick, and heart rhythm abnormalities.

The Children's Hospital of Philadelphia (CHOP) and Penn Medicine have a special program for patients with FA. Through the Penn Medicine/CHOP Friedreich's Ataxia Center of Excellence, our teams work together to ensure that our FA patients have access to top-notch clinical care as well as cutting-edge research opportunities.

At CHOP, we follow more than 150 patients a year with FA. Our patients come here from all over the world for comprehensive care, from initial diagnosis

and testing through long-term disease management. Although there is currently no cure for FA, we utilize the latest research findings and approaches, including medications for managing symptoms and improving quality of life. We also keep our patients updated on the latest research so they can better understand the natural progression of FA and identify new therapeutic approaches. Cardiac care for FA patients at CHOP includes dedicated support from our team of cardiomyopathy specialists, nurse practitioners, study coordinators, and clinical researchers.

CONTACT INFORMATION

To schedule an appointment with the cardiologist, call Kimberly Lin, M.D., at 267-426-2882.

To schedule an appointment with the neurologist, call David Lynch, M.D., at 267-426-9738.

To inquire about research studies related to FA cardiomyopathy, call Felice Wilson at 267-426-2995.

State-of-the-Art Cardiac Services

An outpatient cardiomyopathy evaluation for a typical FA patient may include:

- **Echocardiogram:** An ultrasound of the heart, which can tell us about heart function and thickness.
- **ECG and Holter monitor:** Gives us a snapshot of the heart's rhythm.
- **Laboratory studies:** Certain blood tests may give us a sense of how well your heart is doing.
- **Cardiac MRI:** A noninvasive picture of your heart, which can tell us about heart function, thickness and heart muscle properties such as scar formation.
- **Exercise stress testing:** We have special adaptive equipment that allows us to measure cardiometabolic performance in patients with a wide range of physical abilities.
- **Clinic evaluation:** Our team of physicians and nurse practitioners specialize in cardiomyopathy and advanced heart failure therapies and work with our partners in neurology and other fields to better understand how FA affects the heart.

■ **Coordination of care:** We will communicate directly with your primary care physician and (if you have a cardiologist closer to home) your local adult or pediatric cardiologist to ensure that your cardiac care is seamless and well-coordinated.

Additional consultations may be scheduled as necessary with other subspecialty services such as endocrinology, orthopedic surgery, pediatric electrophysiology, and adult cardiology. We refer patients to them as needed based upon our evaluation findings, and work together with other specialists in a collaborative manner to optimize patient care.

The FA cardiac team meets weekly with the FA neurology team including Dr. David Lynch, Co-Director of the FA Center of Excellence, to provide additional coordination of care within CHOP.

■ **Research participation:** If you are interested in participating in clinical trials, our FA study coordinator will work with you to determine what trials you may qualify for and how to best coordinate any research visits with your clinical evaluation.