

HOW DO I MAKE AN APPOINTMENT?

In order to maximize your benefits from visiting the Johns Hopkins Center for Hypotonia we kindly request that you or your referring physician forward the following information to us prior to scheduling an appointment.

- any reports of laboratory tests and/or muscle biopsies
- any reports from other physicians
- reports of any MRI testing and other radiology testing
- a list of medications
- a referral from your primary care physician
- any other significant documents

Once you have been able to gather all the information, please contact our genetic counselor, **Emily Lisi**, who will assist in scheduling your appointment.



CONTACT INFORMATION

For more information, please contact:

Emily Lisi, MS CGC

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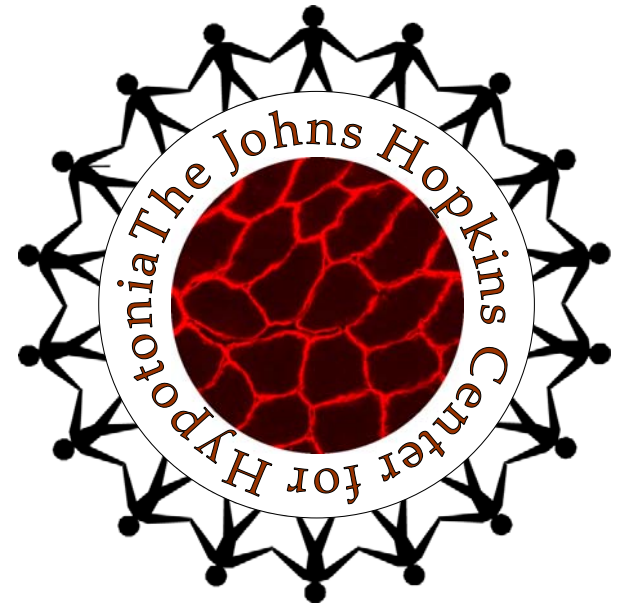


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THE JOHNS HOPKINS CENTER FOR HYPOTONIA

McKUSICK-NATHANS INSTITUTE
OF GENETIC MEDICINE

JOHNS HOPKINS CHILDREN'S
CENTER



Director:
Ronald D. Cohn, MD

Genetic Counselor:
Emily Lisi, MS CGC

OUR MISSION

The Johns Hopkins Center for Hypotonia

is the only clinical and research center of its kind in the world. We focus on identifying, supporting and treating patients with various conditions associated with hypotonia. We strive to ensure the clinical approach to and therapy for the patient will be tailored to his/her individual needs.

Our ultimate goal is to gain a deeper understanding of the natural course of hypotonia and various associated conditions. Subsequently, these insights will be used to develop novel therapeutic strategies which will help the individual patient to achieve their maximum cognitive and physical performance.

WHAT IS HYPOTONIA?

Hypotonia is **not** a diagnosis, rather it is a symptom of diminished tone of skeletal muscle associated with decreased resistance of muscles to passive stretching. Although we are not always aware of it, our muscles receive continuously information and instructions from our brain, whether to relax or to contract and build up tension. The precise combination of contraction in some muscles and relaxation in others enables us to keep in a certain position or posture.

In general, muscles of children with low tone are slower to respond to the contraction stimuli they receive from the brain. In addition, they are often not able to maintain the intensity of the contraction for an extended time period. Together, this causes children to not be able to sit independently, crawl or even stand and walk. Later in life, this may lead to various motor dysfunctions such as an unusual body habitus, inability to run and/or the inability to take stairs independently.

It is important to note that low muscle tone does not necessarily equal decreased muscle strength. However, often, hypotonia and muscle weakness go together and it is clinically very important to distinguish these two from each other as different diagnostic and therapeutic strategies are required for disorders associated with hypotonia and muscle weakness.

We are therefore striving to gain insights into the natural course of hypotonia in various disorders in order to select the appropriate diagnostic and therapeutic path beneficial to the individual patient.

WHAT DISORDERS WOULD BE APPROPRIATE FOR OUR CLINIC?

Each year, hundreds of patients are told to have some form of hypotonia. However, most often, the underlying cause remains

unidentified. Hypotonia can be a symptom of over 500 different disorders. In addition, many other conditions are yet waiting to be identified.

Our center serves as coordinator of multidisciplinary services to evaluate and support patients and their families, in particular:

- **Any hypotonia syndrome associated with or without global developmental delay**
- **Joint hypermobility associated with muscle hypotonia**
- **Arthrogryposis**
- **Congenital disorders of glycosylation**
- **Mitochondrial encephalomyopathies**
- **Metabolic myopathies**
- **Recurrent Rhabdomyolysis**
- **Muscular dystrophies**
- **Pompe's disease**
- **Congenital myopathies**

