THE SPATA FOUNDATION

DEDICATED TO THOSE AFFECTED BY SPATA5
AND SPATA5L1 RELATED GENETIC DISORDERS

SPATAfoundation.org

APRIL 2024

Thank you for your interest in fundraising for The SPATA Foundation! We put together this packet to provide you with talking points that you can use to share about our organization and the genes. We rely on donations to further our mission.

Your efforts are so important and we are appreciative! Thank you for all you do!

ABOUT THE SPATA FOUNDATION

The SPATA Foundation is the first and only patient advocacy group founded for SPATA5 and SPATA5L1 Related Genetic Disorders. We are a 501c3 organization. Our EIN is 93-3768682.

The SPATA Foundation was founded in October 2023 by Mariah and Nicholas George of Palm Coast, FL. Their son, Luca, was diagnosed with SPATA5L1 Disorder in February 2023. The Georges looked for other families and were lucky to find a group of families affected by SPATA5 Related Disorder.

With no patient organization and very little research, the Georges knew they needed to take action and The SPATA Foundation was born. They are actively educating the public, advocating for research, and raising funds for research.

THE SPATA FOUNDATION

OUR MISSION

The SPATA Foundation is dedicated to advocating, educating, and driving research for SPATA5(AFG2A) and SPATA5L1(AFG2B) Related Disorders.

PRESIDENT & DIRECTOR

Mariah George, founder, sits as the President and Director of The SPATA Foundation. Mrs. George is the primary contact for anything associated with The SPATA Foundation. She is passionate about advocating for her son, Luca, and all others affected.

ABOUT SPATA5 AND SPATA5L1

SPATA5 (also called AFG2A) and SPATA5L1 (also called AFG2B) are two genes found in the body. When these genes become mutated, it results in SPATA5 and SPATA5L1 Related Disorders. These disorders result in the same characteristics of hearing loss, epilepsy, physical and intellectual disability, and more. There are approximately 150 diagnosed world-wide with these conditions.

The function of these genes is not fully understood and no treatment currently exists. The SPATA Foundation plans to change that.

THE SPATA FOUNDATION

SYMPTOMS

It's important to note that every case is different, and not everyone affected will experience all symptoms, and symptoms may be experienced at various degrees of severity. There may be other symptoms present related to these disorders, but these are the most common.

- Hearing Loss
- Developmental Delay
- Physical/Intellectual Disability
- Epilepsy
- Feeding Difficulties
- Hypotonia
- Spasticity
- Dystonia
- Chorea
- Ataxia
- Vision Impairment
- Abnormal MRI
- Microcephaly
- Neurostorming

PATIENTS

We are currently aware of approximately 80-100 SPATA5 families and 40-60 SPATA5L1 families worldwide. The oldest living patient known is 31 years old.

OUR BUDGET

Everything we do is primarily funded by generous donations from our SPATA Families and their networks. We cannot fulfill our mission without monetary support!

THE SPATA FOUNDATION

ADVOCACY & PARTNERSHIPS

We believe that collaboration between patients, families, and researchers is essential to furthering our mission. Our families deserve to be heard.

The SPATA Foundation is involved in multiple collaborative efforts. Though our rare disease community is small, the general rare disease community is huge. We partner with other rare disease organization & advocacy groups with similar goals in order to achieve our goals.

RESEARCH

The SPATA Foundation is dedicated to furthering research of SPATA5 and SPATA5L1 Related Disorders. We consistenly communicate with other rare disease organizations and researchers to determine our best path forward towards the best treatment options.

We are following a 4-step strategic approach in order to prioritize tasks & funding

THE SPATA FOUNDATION

RESEARCH

We have made significant strides since our foundation in October 2023:

- Made Strategic Relationships with researchers & other rare disease organizations
- Began the process of creating a Patient Registry
- Secured development of mouse models through an NIH grant at Jax Labs
- Advanced towards our first research opportunity: Drug Repurposig
- Made relations with various gene therapy researchers
- Researched and opened the idea for a designated clinic

Our top research priorities are:

- Understanding the function(s) of SPATA5 and SPATA5L1
- Creating a viable treatment option quickly to reduce symptoms
- Developing and funding a long-term plan towards gene therapy

MEDICAL & SCIENTIFIC ADVISORY BOARD

The purpose of the Medical and Scientific Advisory Board (MSAB) is to support the mission of The SPATA Foundation by providing guidance and direction in regards to translational and clinical research, therapeutics, and relevant medical topics. The MSAB will be composed of appointed Advisors who have credentials, training and/or expertise, and interest in SPATA5 and SPATA5L1 Related Disorders that qualifies them to provide medical and scientific guidance and assistance to The SPATA Foundation.

Current Members:

Michael Kruer, MD

Pediatric Movement Disorders and Neurogenetics

Phoenix Children's Hospital

Tapas Mukherjee, PhD

Postdoctoral Fellow - Immunology

University of Toronto