THE SPATA FOUNDATION

Dedicated to families affected by SPATA5 and SPATA5L1 Genetic Disorders

www.SPATAfoundation.org | info@SPATAfoundation.org

Updated March 2026

We put together this packet to provide you with talking points that you can use to share about our organization and the genes.

About The SPATA Foundation

The SPATA Foundation is the first 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.

Our Mission

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

President and 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.

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.

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
- Microcephaly
- Spasticity
- Dystonia
- Chorea
- Ataxia
- Vision Impairment
- Abnormal MRI
- Neurostorming

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!

We post all our Annual Reports at www.SPATAfoundation.org to be transparent with where our funds are being allocated.

THE SPATA FOUNDATION

Advocacy and 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.

International Partners

GUERREROS SPATANOS
LUCHANDO CONTRA EL SPATAS

SPAIN GROUP

SPATA Australia

AUSTRALIA GROUP

SPATA cz

CZECH REPUBLIC GROUP

Partnerships

REN Rare Epilepsy Network

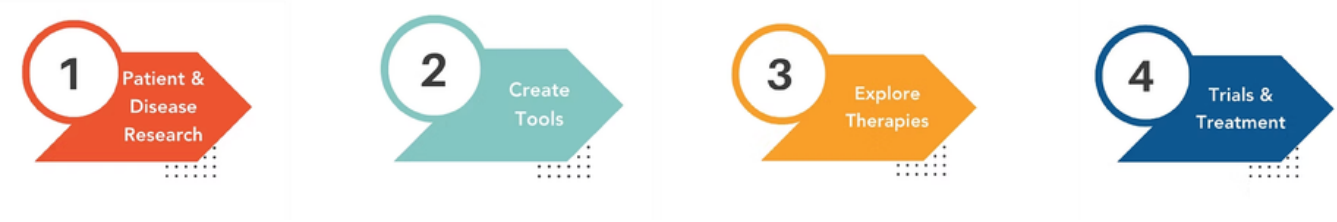
UNRAVEL BIOSCIENCES

Global Genes® Global Advocacy Alliance

COMBINED

Research

Our path to a cure recognizes that families need options now, not someday. We are advancing research into near-term, practical treatments that can improve quality of life and be accessible to the entire SPATA community, while also laying the scientific groundwork for transformative therapies such as gene therapy.



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

- Formed a Medical and Scientific Advisory Board and Made Strategic Relationships with researchers & other rare disease organizations
- Created a Patient Registry
- Secured development of mouse models through an NIH grant at Jax Labs
- Launched a Drug Repurposing Research Study
- Made relations with various gene therapy researchers
- Hosted our very first SPATA Foundation Family Conference

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

THE SPATA FOUNDATION

Medical and 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.

Michael Buszczak, PhD
Professor
—
Dept. of Molecular Biology
UT Southwestern

Michael Kruer, MD
Program Director Pediatric Movement Disorders
—
Barrow Neurological Institute
Phoenix Children's Hospital

Tapas Mukherjee, PhD
Postdoctoral Fellow
—
Dept. of Immunology
University of Toronto

Barbara Vona, PhD
Principle Investigator
—
Institute of Human Genetics, Institute for Auditory Neuroscience
University Medical Center Göttingen