

angelman
SYNDROME FOUNDATION

Clinics



**For Patients.
For Providers.
Together
Toward
The Cure.**

Part of the

LADDER

Learning Network



A DIAGNOSIS

Hearing that a child has been diagnosed with Angelman syndrome, a rare neurogenetic disorder for which there is not yet a cure, is initially a difficult concept for parents to grasp. What can be expected for their child in the areas of development, intellectual and emotional progress? What resources and services are available for their child's care and upbringing?

Caring for a loved one with Angelman syndrome can be overwhelming. At every turn, parents find themselves managing new concerns and new issues that put tenacity, patience, perseverance, ingenuity and will to the test. Parents deal with financial, legal, educational, vocational, behavioral and social matters that require guidance, support and assistance beyond anything they can manage on their own. Angelman Syndrome Foundation joins hands with these families and walks with them through this journey as we work to support the needs of their loved one with AS.



Meet SOPHIE

“Sophie’s seizures have been the most difficult part of AS for us as a family. Aspects of Sophie’s epilepsy have baffled other child neurologists we have worked with, and the only recourse offered to us was to increase medication or add additional medications—both of which impacted her quality of life. Dr. Thibert and his team at the ASF Clinic in Boston have a wealth of experience with seizures in AS and with treatments that work. Now that she

sees this team regularly, we have a seizure treatment plan that emphasizes no seizures and medication side effects. Knowing this team is just a phone call away between visits has helped us treat seizure outbreaks without having to constantly increase her medications. Her quality of life has improved significantly, and we feel supported in our quest to keep her seizure free.”

JACOB'S story

Fred, Renee and Jacob Pritzker



“Renee and I are Jacob’s parents. He is a kind, joyful and profoundly decent young man whose life we treasure more than words can describe. But like most people with Angelman syndrome (AS), he suffers from anxiety, which affects the quality of his life (and ours). We don’t always know what triggers it, but we know too well its manifestations: cyclic vomiting, incessant swallowing, self-injurious behavior and obvious sadness and fear.

AS is a rare, relatively recently identified and far from fully described disorder. Simply put, most practitioners see too few AS patients to fully understand and effectively treat their symptoms. That was certainly true in Jacob’s case. Jacob was worked up with invasive and uncomfortable GI procedures and treated with a number of medications that never re-mediated his symptoms.

There is nothing worse than seeing someone you love suffer and be powerless to stop it. We decided to make the trip with Jacob to the ASF Clinic at Mass General Hospital in Boston where he was first seen by Ron Thibert, and later by Chris Keary, two fine and compassionate physicians who have devoted their professional careers to improving the lives of people like Jacob. They were able to properly diagnose

and treat him with new medication that markedly improved the quality of his life.

When problems arise, medication needs to be adjusted, or when Jacob’s local doctors need support for their clinical decisions, his MGH team offers timely and reassuring assistance. This is comforting in a way that only the parents of a child with severe disabilities can fully appreciate. Because of our experience at MGH and because there is no other comparable resource, we are committed to contributing to and raising money for these vitally important ASF Clinics.

We are doing so for another reason. Advances in research will at some point cure AS. This should continue. But cures and improvements in quality of life for people with AS are not mutually exclusive. To invest in the future, we must support (and fund) treatments and therapeutic modalities for patients with AS who are suffering now. There is no better way to do that than funding ASF Clinics. Please join us.”

— Fred Pritzker



Get to Know **MASON**

“In the past, I had taken Mason to about 30 different doctors. It was painstaking. Mason was having problems eating and was not gaining weight. His doctor at home was adamant that a feeding tube was the only answer. We knew this was an invasive and last resort procedure and decided to have Mason seen at an ASF Clinic. The team of doctors at the ASF Clinic found that a feeding tube was not necessary and together came up with a

plan to help Mason. Working with the clinic’s GI specialist and dietitian, they were able to get Mason eating and receiving proper nutrition without an invasive procedure.

“ *It gives us confidence that*
WE’RE NOT ALONE
IN THIS.” – Steve M.

It gives us confidence that our son Mason is getting the best care possible. It gives us confidence that there is a really knowledgeable, caring and powerful organization out there pushing forward the research and the development of treatments for AS. It gives us confidence that we’re not alone in this.”

—Steve M.

THE FOUNDATION

Angelman Syndrome Foundation is the voice on the other end of the phone. It's the voice that is available to interact with any and all affected by Angelman syndrome and provides information and direction as families begin their journey with AS. Angelman Syndrome Foundation was formally incorporated in 1992, but it has origins dating back to the pioneering work, passion and vision of Dr. Harry S. Angelman, after whom the syndrome is named.

Today's Angelman Syndrome Foundation has grown from a parent-led organization serving the needs of just a few families to a professionally led organization serving thousands of AS families every year. This growth has allowed Angelman Syndrome Foundation to accelerate funding of basic- and clinical-science research by more than \$14 million in research-grants, which in turn, has resulted in \$150 million in new peer-reviewed research grants at prestigious and notable academic medical institutions such as University of North Carolina and University of Connecticut. We also took an ambitious step and launched ASF Clinics, a network of more than 20 clinics across the globe to provide much needed clinical care, like that received by Jacob, for children, young adults and adults with AS.

WHAT IS ANGELMAN SYNDROME

Angelman syndrome is a complex neurogenetic disorder that crosses all socio-economic and ethnic lines. It is estimated to occur in one out of every 12,000 to 15,000 live births. The characteristics of Angelman syndrome include severe developmental delay, speech and communication difficulties, seizure disorders and difficulties with movement and balance. The hallmark features of Angelman syndrome are a happy and excitable demeanor and the absence of speech. For those it touches, AS changes lives forever.

Seizures in individuals with Angelman syndrome can be life threatening—unfortunately claiming the lives of too many. Perhaps the most severe of all symptoms, seizures that go untreated due to improper or delayed diagnosis can cause even further damage to the already fragile development of individuals with Angelman syndrome.

The earlier the diagnosis of Angelman syndrome, the sooner therapeutic and rehabilitative measures can begin to help individuals reach their maximum potential and optimum quality of life. Treatment is a life-long process that involves multiple disciplines and approaches. That is where ASF Clinics come into play!

Each ASF Clinic offers the following clinical specialists to each patient:

- + Clinical Geneticist
- + Neurologist
- + Psychiatrist
- + Psychologist
- + Speech and Language Therapist
- + Physical and Occupational Therapist
- + Genetic Counselor
- + Social Worker
- + Nutritionist

ASF provides necessary funding for these Centers of Excellence so that our loved ones with AS can receive the care they need when they need it. ASF Clinics currently have 20 locations across the globe. However, several of these clinics are relatively new and are still not fully staffed because more funding is needed.

ASF Clinics currently serve approximately 2,500 AS patients—a tiny fraction of the AS community members who desperately need access to this comprehensive care.



“ASF Clinics offer families an opportunity to get comprehensive evaluations and discuss their concerns with professionals knowledgeable about Angelman syndrome. The clinics allow families to access expert care directed at all aspects of the disease, as well as critical clinical research and trials of cutting-edge treatments targeted to the underlying disorder. This kind of care is tremendously important for any rare disease and specialized clinics will be key to the delivery of the treatments of the future”

Elizabeth Berry-Kravis MD PhD

*Departments of Pediatrics, Neurological Sciences, Biochemistry
Rush University Medical Center*

Our Vision IS BOLD

We want to provide the best clinical care possible and support for every individual with AS. In order to do that, we need you. We have positioned Angelman Syndrome Foundation to enhance and expand its excellent clinical research network. Now, it is time to grow and insure sustainability for years to come. Now is the time to lead.

With You For the Journey



Working TOGETHER *Toward The Cure*

While Angelman syndrome has been a known and identifiable rare disease since the 1980s, AS is still considered to be a relatively new disorder, and therefore is not always familiar to most physicians. Because most physicians do not encounter patients with AS regularly, when confronted with an AS patient and their often complex medical needs, these physicians are not always familiar with the best practices used to treat AS patients. When a loved one with AS does not get the appropriate treatment needed, it only adds to the concern and frustration of parents and family members.

To address the ever-increasing need for more physicians who are well versed in best practices for treating individuals with Angelman syndrome, Angelman Syndrome Foundation has partnered with leading research medical institutions across the country in collaboration with Dup15q Alliance to form the LADDER Learning Network. Inclusive of ASF Clinics, this specialty network is designed to provide individuals with AS the comprehensive medical care needed, all available in one clinical visit in one single location. Prior to this, families would search for multiple clinics and multiple specialists to receive the necessary care for their loved one with AS.

YOUR IMPACT

For these initiatives, we look to patients, friends, and foundations who share our commitment to partner with us as we take a huge step in our journey to cure AS together. Your support can immediately make an impact on our boldest ideas and can ultimately change the lives of those living with AS all over the world. The funding opportunities in this brochure represent extraordinary chances to help bring clinical care to those in need and to support a network of clinicians committed to working toward the cure.

Specifically, we are seeking support for people, clinical care, research and discovery. Our campaign objectives include approximately \$6 million in new funding to advance our vision. There will be many ways for you to engage with us and take this journey together to bring so much hope to those all over the world.



OUR VISION

Looking into the future, there are critical steps that we must take to sustain, grow and change the landscape of clinical care for those with Angelman syndrome. To do so, we are launching an ambitious campaign to ensure the critical funding to expand and sustain these life-changing clinics and possible therapeutic treatments. Through this comprehensive campaign, our objective is to provide:



Support For People

Support for ASF Clinics to bring on multi-disciplinary experts and support for the ASF Family Fund, which was created to help families offset travel costs.

Total Support Needed: \$1M



Support For Clinic Discovery

Creation of a fund for Discovery Grants that will allow current AS clinicians and researchers to apply for funding to expand their services to the AS community. The advances made possible by innovative research will provide critical new knowledge for ASF Clinics.

Total Support Needed: \$2M

+ Discovery Grants: \$1M per year for 5 years to support clinical research



Support For Research

Sustain current clinical research and clinical trials support by providing a yearly stipend. Therapeutic discovery is happening daily in ASF Clinics which must be supported and sustained to get closer to treatments.

Total Support Needed: \$2M



Support For Future Clinical Care

Expansion of other multi-disciplinary clinics to support more individuals with AS across the world.

Total Support Needed: \$1M

+ Development of heat map study to determine need of new clinics and supporting the start up of new clinics at \$50,000 for the first year.

Total Needed to Jump Start Our Vision for the Future: \$6M



The mission of Angelman Syndrome Foundation is to advance the awareness and treatment of Angelman syndrome through education and information, research, and support for individuals with Angelman syndrome, their families and other concerned parties. We exist to give all of them a reason to smile, with the ultimate goal of finding a cure.

Angelman.Org

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