angelman
SYNDROME FOUNDATION

FAMILIES. RESEARCH. CLINICS. COMMUNITY.

WITH YOU FOR THE JOURNEY.

> *"Those of you who will share your experiences will do it because in the Angelman family, that is what we do. Whatever road you have traveled, others have traveled before you, so there is no need to travel alone."*
>
> – Audrey Angelman

Your support helps families navigate every new and challenging step.

MASON (DEL+)

From initial diagnosis, first time parents Krista and Stephen followed a journey shared by so many of our families. When their son, Mason was slow to reach developmental milestones, doctors struggled to find a diagnosis. A second round of genetic testing led them to an answer they had never heard of – Angelman syndrome.

WHAT IS ANGELMAN SYNDROME?

Initially presumed to be rare, it is now believed that thousands of cases of Angelman syndrome (AS) have gone undiagnosed or misdiagnosed as cerebral palsy, autism or other disorders.

The following are clinical characteristics of Angelman syndrome:

- Developmental delay – functionally severe
- Speech impairment – minimal or no use of words
- Movement and balance disorder
- Seizures – sometimes severe – the onset usually occurs between 18-36 months of age
- Sleep disturbances – AS individuals typically require very little sleep
- Unusual attraction and love of water
- Any combination of frequent laughter/smiling; apparent happy demeanor; easily excitable; hand flapping movements (hypermotoric behavior)
- Children with AS will require a lifetime of constant, 24/7 care and multiple specialists

Today, we know that Angelman syndrome is a genetic disorder caused by abnormal function of the UBE3A gene, located on the 15th chromosome.

MASON'S JOURNEY

It began with getting much-needed specialized care. He was having problems eating and was not gaining weight. His doctor felt a feeding tube was the only answer. This was a big decision, so Stephen and Krista decided to have Mason seen at an ASF Clinic. The team of doctors as the ASF Clinic found that a feeding tube was not necessary and came up with an alternative plan. Working with the clinic's G.I. specialist and dietitian, they were able to get Mason eating, giving him proper nutrition without a surgical procedure.

Today, Mason is thriving and his parents are thrilled with his progress and feel confident that they can get the support they need.

> *"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 developing treatments for AS. It gives us confidence that we are not alone in this."*
>
> – Stephen M.

YOUR DONATION COUNTS!

The Angelman Syndrome Foundation is dedicated to funding research geared toward treatments and an eventual cure. At the same time, we are dedicated to supporting our families by:

- Funding clinics with specialized doctors
- Providing services, resources, supports and events
- Providing education via conferences, webinars, online and e-training (communications, behaviors, IEP bank)

Your generosity enables AS families to receive the support and resources they need. Your financial gift funds research, makes every day a little easier and opens the doors to more ASF Angelman Clinics with specialized care.

> *"The ASF is committed to walking each step with our families on this journey. It is often terrifying, exhausting, joyful and uncertain; but having an organization like the ASF makes the journey a little less frightening. It is because of donors like you, the ASF can commit to giving families the smiles they deserve."*
>
> – Amanda Moore, ASF CEO

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