

# Angelman Syndrome Clinic at Kennedy Krieger Institute

## Who We Are

The Angelman Syndrome Clinic at Kennedy Krieger Institute is a comprehensive, interdisciplinary clinical program focused on treating people with a genetically confirmed diagnosis of either Angelman syndrome (AS) or maternal 15q duplication (dup15q) syndrome.

We provide comprehensive medical evaluations, treatment, clinical care and genetic counseling. Our expertise in these rare disorders, combined with our vast resources, ensure that our patients' needs can be met at one location. It is our goal to help our patients and their families gain a better understanding of their condition and access appropriate medical care to improve their overall health and quality of life.

AS is a rare neurodevelopmental genetic disorder that affects approximately one in 15,000 people. AS primarily impacts the nervous system, and people with AS typically have recurrent seizures, balance and coordination issues, developmental delays, intellectual disability, and sleep disturbances. AS is commonly diagnosed in the first year of life. Developmental delays—usually the first sign of AS—are often present in affected individuals by the time they are 6 months old. Seizures typically begin before the age of 3 years.

Dup15q syndrome is also a rare neurodevelopmental genetic disorder. While its prevalence is unknown, it is estimated to affect approximately one in 5,000 people. It is characterized by hypotonia, motor delays, intellectual disability, autism spectrum disorder (also known as ASD) and epilepsy, including infantile spasms.

The Angelman Syndrome Clinic, in partnership with the Angelman Syndrome Foundation and the Dup15q Alliance, is the only clinic dedicated to the care of this patient population in the Mid-Atlantic region.



## Who We Serve

The clinic serves children, adolescents and adults with either AS or dup15q syndrome. These two rare disorders can have a variety of symptoms, including:

- Delayed development
- Intellectual disability
- Gastrointestinal issues and feeding challenges
- Challenges with movement and balance
- Jerky body movements
- Severe speech impairments, with little to no expressive speech
- Hyperactivity
- Seizures
- Difficulty sleeping
- Happy, excitable personalities



## Our Team

Our interdisciplinary team is made up of experts from various clinical specialties. Clinicians, research scientists and rehabilitation professionals work together to provide optimal interdisciplinary care for our patients and their families. Because AS and dup15q syndrome can affect many different systems in the body, treating a patient with one of these disorders often requires the collaboration of a wide range of medical specialists. Our team includes the following:

- Neurodevelopmental disabilities specialist
- Genetic counselor
- Behavioral psychologist
- Social worker
- Physical therapist
- Occupational therapist
- Speech-language pathologist

## Our Approach

### Evaluation

AS and dup15q syndrome are diagnosed via a blood test and/or genetic testing. Individuals who come to the clinic should already have a genetically confirmed diagnosis of either AS or dup15q syndrome. A clinical evaluation will determine how the diagnosis has impacted the individual, and the clinic team will recommend treatments and medical care, as well as further testing and genetic counseling, if needed.

### Treatment

While there are no specific treatments that can reverse the symptoms of AS or dup15q syndrome, there are treatments that can improve patients' quality of life. We use evidence-based guidelines and standard-of-care practices to address symptoms such as intractable seizures, movement and balance issues, sleep disturbances, and gastrointestinal difficulties.

Additionally, the clinic serves patients and their families across the lifespan, connects families with national and local resources, and assists with school support, as needed.

## Contact Information

### Angelman Syndrome Clinic at Kennedy Krieger Institute

For more information or to schedule an appointment, please call **443-923-9400**, or call toll-free at **888-554-2080**. TTY users, please contact us at **443-923-2645**, or dial **711** to make a Maryland Relay call.

### KennedyKrieger.org

### Physicians and Healthcare Professionals

To make a referral, email us at **FindASpecialist@KennedyKrieger.org** or call our Physician Referral Line at **443-923-9403**.

### Clinic Location

801 North Broadway  
Baltimore, MD 21205

### Mailing Address

707 North Broadway  
Baltimore, MD 21205

*The photograph used is for illustrative purposes only, and any person shown is a model.*

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