



Interview Briefing

<p>Key Messages:</p>	<p><u>Angelman syndrome</u></p> <p>What is Angelman syndrome (AS)?</p> <ul style="list-style-type: none"> ○ Angelman syndrome (AS) is a neurogenetic disorder similar to autism ○ AS is a rather severe disorder, as individuals with AS experience significant developmental delays. ○ AS is often misdiagnosed as autism, cerebral palsy or other neurodevelopmental disorders due to lack of awareness. <p>What are some of the characteristics of AS, and how do individuals with AS develop?</p> <ul style="list-style-type: none"> ○ Individuals with AS require life-long care. ○ Individuals with AS also typically exhibit a happy demeanor characterized by frequent smiling, laughter and excitability. ○ Symptoms of AS and their severity vary from individual to individual—however, individuals with Angelman syndrome generally experience developmental delay, lack of speech, seizures, sleep deprivation, and walking and balance disorders. ○ One of the more severe and concerning symptoms of AS is seizures, which 90 percent of individuals with AS experience, and which are extremely difficult to manage and treat in individuals with AS. ○ Seizures are a potentially life-threatening symptom and are very difficult to treat. Unfortunately, there have been individuals with AS who have passed away due to seizures. <p>Why should anyone outside of the AS community care about AS?</p> <ul style="list-style-type: none"> ○ AS is linked to autism, which affects millions of individuals throughout the world and is becoming a more commonly known and understood disorder. ○ Nearly 40 percent of individuals with AS have a correct dual diagnosis of AS and autism. ○ Autism can be caused by an excess of a particular gene or genes in the brain, whereas AS is caused by the loss of function of one specific gene in the brain. ○ Many in the scientific and medical fields believe that until a one-gene disorder, such as AS, can be understood and treated, multiple-gene disorders, such as autism, can not be fully understood or treated. ○ Research discoveries made in the field of AS, and many other research projects which are ongoing, have unveiled interesting findings that have proven useful in understanding and treating other neurodevelopmental disorders. <p><u>Angelman Syndrome Foundation</u></p> <p>Who is the Angelman Syndrome Foundation (ASF)?</p> <ul style="list-style-type: none"> ○ The Angelman Syndrome Foundation (ASF) is a national non-profit organization dedicated to advancing awareness and treatment of Angelman
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syndrome through education and information, research, and support for individuals with AS and their families.

- The ASF was founded by families and continues to be led by families and other individuals personally affected by AS.
- For more than 30 years, the ASF has:
 - Provided information, support and resource connections to more than 250,000 individuals.
 - **Funded research grants totaling more than \$10 million.**

How does the ASF support individuals with AS?

- **Research.** The ASF provides research grants to institutions dedicated to finding possible treatments for individuals with AS. To date, more than 117 research grants totaling more than \$12.4 million have been awarded by the ASF that have led to very important research discoveries.
- The ASF provides direct support by being the “**voice on the other end of the phone**” and providing newly diagnosed families, experienced AS families and other concerned individuals with first-hand information, and connecting them with the resources they need.
- **Angelman Syndrome Clinics.** The ASF has opened 24 AS clinics since 2012. At the AS clinics, individuals with AS can access all of the medical resources they need under one roof, rather than having to visit multiple sub-specialists in multiple locations to address all of the needs of individuals with AS.
- **Family Support Services.** The ASF provides a myriad of support services to our families across the globe.

National Walk

- Held in 51 cities across North America
- The ASF National Walk is the ASF’s biggest fundraiser – 70% of the funds raised from this event are invested directly to direct family support, AS clinics & research.
- Walk sites are coordinated by local volunteers, a majority of which are families of individuals with AS.
- The ASF has a goal to raise \$1.2 million in support of Angelman syndrome research AS Clinics and family support services.