

Angelman syndrome (AS)

Related Conditions & Complications

Not all conditions listed below will be applicable to every individual who has Angelman syndrome (AS). Some don't have epilepsy, some might sleep well, many are placid and have no behaviours that require management, and some might only have mild problems with reflux and constipation. However, when the following conditions *are* present, they need to be carefully monitored and **managed**. Some of these conditions can cause pain, stress, violent behaviours, and major disruptions to family life - and can even become life threatening and fatal, if not treated appropriately.

1. EPILEPSY

More on our website [here](#)

2. SLEEP DISORDER

- 2024: **The Type, Severity, and Impact of Sleep Problems in Children With Angelman Syndrome and Parental Help-seeking Patterns**
- 2020: **Angelman syndrome and melatonin: what can they teach us about sleep regulation.**
- 2015: **Circadian clock linked to Angelman syndrome.** *"...Serious sleep disorders are one of the symptoms of AS. These can be so serious that the*

children must sleep in special crib-like beds to keep them from wandering or harming themselves at night.”

- 2004: **Sleep disturbances in Angelman syndrome: a questionnaire study** *“...A significantly high frequency of disorders of initiating and maintaining sleep, prolonged sleep latency, prolonged wakefulness after sleep onset, high number of night awakenings and reduced total sleep time were found in our AS patients, as compared to age-matched controls. We also found other types of sleep disorders, never reported before, such as enuresis, bruxism, sleep terrors, somnambulism, nocturnal hyperkinesia, and snoring.”*
- 2018: **Why My Child’s Sleep Problems Just Can’t Be ‘Put to Bed’** *“...According to Angelman-related literature, sleep problems with our children can’t just be put to bed. The degree to which sleep evades children with AS is enormous, and can include everything from difficulties with falling asleep to reduced sleep time, bedwetting, waking during the night, terrors, and even sleep walking. Like typical children, youngsters with AS also don’t like to stay in bed and often try to escape their beds or rooms to seek stimulation via toys, electronics, water, and even outside spaces. My son, as well as many other children with AS, sleeps in a specialized safety bed that keeps him enclosed and safe during the night.”*

3. GASTROINTESTINAL (GI) SYMPTOMS

- 2022: **Association between early and current gastro-intestinal symptoms and co-morbidities in children and adolescents with Angelman syndrome**
- 2017 study: **Prevalence of gastrointestinal symptoms in Angelman syndrome** *“... The majority of patients' medical records indicated at least one symptom of gastrointestinal dysfunction, with constipation and gastroesophageal reflux disease (GERD) the most common. Other gastrointestinal issues reported were cyclic vomiting episodes, difficulty swallowing, excessive swallowing, and eosinophilic esophagitis. Upper gastrointestinal symptoms such as GERD, swallowing difficulties, cyclic vomiting, and eosinophilic esophagitis were more*

common in those with deletions and uniparental disomy, likely related to the involvement of multiple genes and subsequent hypotonia. The frequency of constipation is consistent among all genetic subtypes while early feeding issues appear to mainly affect those with deletions. Caregivers and healthcare providers should be aware of the high prevalence of these issues, as proper treatment may improve not only gastrointestinal dysfunction but also sleep and behavioral issues.”

- **2017: Angelman Patients Frequently Have Gastrointestinal Problems**
- **Dysphagia (swallowing problems):** Some adults with Angelman syndrome experience problems swallowing certain foods or liquids, while others can't swallow at all. Signs include coughing or choking when eating or drinking, bringing food back up, sometimes through the nose, a sensation that food is stuck in your throat or chest, persistent drooling of saliva. Over time, dysphagia can also cause symptoms such as weight loss and repeated chest infections.
- **Eosinophilic Esophagitis (EE):** Eosinophilic esophagitis is a chronic allergic inflammatory condition of the oesophagus, characterized by symptoms such as solid-food dysphagia (difficulty swallowing) and food impaction in adults, and feeding intolerance and GERD symptoms in children. An EGD (Esophagogastroduodenoscopy) is a procedure that examines the oesophagus, stomach and first portion of the duodenum (small intestine) using a long flexible tube with a camera at the end of it. The scope is inserted into the mouth and advanced to the small intestine.
- **Elevated ammonia levels:** Constipation and certain Epilepsy medications, eg. Epilim/Depakote (also known as valproic acid) can be causes of elevated ammonia levels, which can cause a variety of problems. Ammonia is a nitrogen waste compound that is normally excreted in the urine. Elevated blood ammonia levels can affect a person at any age and happens for a variety of reasons.
- **Reflux and constipation** are common and can become serious if left untreated during adulthood. Ideally GI issues should be treated holistically with an appropriate diet plan, correct positioning during and after meals, physical activity, as well as medical intervention when necessary.

4. CYCLIC VOMITING

- 2025: **Cyclic vomiting (CVS)**
- 2019: '**Guidelines on management of cyclic vomiting syndrome in adults by the American Neurogastroenterology and Motility Society and the Cyclic Vomiting Syndrome Association**'
- 2017: '**Treatment of Cyclic Vomiting Syndrome**'

Cyclic vomiting syndrome (CVS) can cause dehydration, injury to the food tube (sometimes the oesophagus becomes so irritated it bleeds), and tooth decay. The acid in vomit can corrode tooth enamel. If episodes occur more than once a month or require hospitalization, your doctor may recommend preventive medicine, such as amitriptyline, propranolol (Inderal), cyproheptadine and topiramate. Read more [here](#).

5. ANXIETY AND AGGRESSIVE BEHAVIOURS

- 2022 - **Anxiety in Angelman Syndrome**
- 2019 **Anxiety-associated and separation distress-associated behaviours in Angelman syndrome** - *This study used a combination of standardised and clinician-derived survey items to assess the frequency, nature and severity of behaviours associated with anxiety and separation distress in 100 individuals with Angelman syndrome.*
- **Aggressive Behaviour in Angelman Syndrome**
- **Behaviours that Challenge Associated with Frustration Related to Communication Difficulties**
- **Behaviours that Challenge Related to Anxiety**

6. ORTHOPEDIC CONDITIONS

Scoliosis & Skeletal - see **Management of Angelman Syndrome - A Clinical Guideline**

Gait and movement disorders: **Physical therapy** is essential to help improve ambulation. Sometimes bracing (AFOs) or surgical intervention may be needed to properly align the ankles, feet and knees.

Osteopenia - [Bone mineral density in Angelman syndrome](#)

7. VISION - STRABISMUS AND CVI:

2020: [Will Angelman Syndrome Affect My Child's Vision?](#)

Management of strabismus requires evaluation by an ophthalmologist who will advise on a course of action. See [Cortical Visual Impairment Symptoms and Causes](#).

8. PICA

Pica is the repeated or chronic ingestion of non-nutrient substances. In children with developmental delay, pica occurs in 10–33%. Although many people with AS are picky eaters, they might still be impulsive when it comes to chewing, biting and swallowing objects that are not food, eg. *Foam stuffing, small objects, plastics, small toys, gravel, balloons, etc.* This can cause choking and/or obstructions in the throat and digestive system that can become serious and at times, lethal. People with Pica require 24/7 supervision.

9. ANAESTHETICS AND SURGERY RECOVERY:

Read more here: [FAQs About Anesthesia in Angelman Syndrome](#)

Aspiration and respiratory distress: Though most people with AS are very resilient and seem to withstand anaesthetics and extensive surgeries well, many can struggle with recovery in the first few days after surgery. It is most important to achieve good pain control while closely monitoring for **aspiration** and **respiratory distress**.

10. BODY TEMPERATURE REGULATION

by *Brian Murphy, Ph.D*, 2020 [Temperature Sensitivity in Angelman Syndrome](#)

- Increased sensitivity to heat - Since **temperature control may be poor** in Angelman patients, caregivers should avoid overdressing them and ensure adequate hydration, especially in warmer areas.