

# Cognitive Issues, Sensory Impairments in Individuals with Angelman Syndrome

## Cognition, Social Cognition, and Adaptive Behavior Defined

**What is cognition?** Definition of cognition - "Cognition" or cognitive development refers to mental processes that include language, memory, attention, pattern recognition, perception, motor skills, and problem-solving. A person may have strengths and/or weaknesses across various aspects of cognition/processes such that they may excel in one domain but have considerable difficulty in another. The motor aspects of cognition involves mental processing in which the motor system draws on stored information to plan and produce actions, as well as to anticipate, predict, and interpret the actions of others. This involves motor planning, and execution of actions. Cognitive processes are integrated into most of what we do on a daily basis and form the basis for learning/acquiring skills. Social cognition extends these processes to social interactions and involves understanding another person's perspective/thoughts/feelings, and social norms.

**What is adaptive behavior?** Cognition can also influence "adaptive behavior." Adaptive behavior refers to independent performance of activities of daily living. This refers to broad-based skills (e.g. feeding, dressing, talking, reading, sharing, understanding feelings and emotions, coloring, walking) that must be acquired through development in order to function in and access a variety of settings.

Various aspects of cognition and adaptive behavior are often measured for children/adults to receive special services. These evaluations are typically conducted individually by a psychologist. Teachers and parents typically act as informants for adaptive behavior. Taken together, an evaluation of cognition and adaptive behavior provides an estimate of an individual's level of understanding/performance and how they may function in school/work settings.

## Cognitive Processes in Angelman Syndrome

**How do cognitive process work in individuals with Angelman syndrome?** Cognitive processes in AS - Research has consistently demonstrated that individuals with AS have difficulties with all aspects of cognition. Our experience is that individuals with AS perform better on cognitive tasks that are less abstract, and that involve minimal use of language. Many individuals have difficulties with the motor aspects of cognition, and it may take them longer to demonstrate a skill. Depending upon the nature of the cognitive task, and how demanding it may be, individuals with AS may exhibit aggressive and/or challenging behaviors. These may include throwing objects, mouthing objects, hitting others, spitting, pinching, kicking, biting, hair pulling, and/or attempts to leave the room/run. We have also experienced/observed tight hugs, flopping on the floor, and having an individual put their head down on the table when presented with harder tasks. These behaviors may serve to avoid the difficult task, and/or convey frustration. We have also found that an increase in challenging behaviors may occur as individuals begin to master new skills (i.e. as their cognitive abilities improve), and when they are with a new therapist/teacher.

**How can I help decrease aggressive behaviors in individuals with Angelman syndrome in situations with high cognitive demands?** To counteract aggressive behaviors in these situations, we suggest starting with easier tasks so that the individual can feel successful and have some positive interactions. If the individual with AS seems to have an understanding of cause/effect (i.e. if I do this, then I can have \_\_\_\_ or \_\_\_\_ will occur), then we would also suggest using tangible reinforcers/motivators (e.g. verbal praise, screen time, stickers, listening to music, small bites of food, etc.) to help the child. Some individuals with AS seem to be more successful with working in shorter blocks of time (e.g. 20 minute blocks) with breaks that are integrated. For individuals who are attentive to pictures/photographs, we suggest incorporating visual schedules to prepare them for what may be coming next. A "first-then" board is one example of a visual schedule. The board shows that a less preferred activity must happen before a preferred activity can occur. Photographs, picture symbols, and/or real objects can be incorporated as part of a first-then board.

We would also suggest some type of baseline evaluation to determine strengths and weaknesses, and how areas of strength can be used to compensate for tasks that are more challenging. Regardless of the strategy or approach that is utilized, we would recommend that individuals with AS be given extra time to process instructions and carry out a task (due to potential difficulties with motor planning).

## Defining Sensory Issues

We are going to shift directions right now to discussing sensory processing and how this impacts behaviors and interactions in AS.

**What are sensations?** Defining Sensory Issues - "Sensations" involve the sight, feel, taste, sound, and smell of things. In order to function effectively, we integrate the information we receive from our senses. This information is then sent to the brain via neural pathways, and then it can be acted upon. When sensory functions are working correctly, they keep us from touching things we should not touch, and eating things we should not eat for example. Disruptions to these processes can result in being hyper-responsive (i.e. over-responsive) to sensory input, or in being hypo-responsive (under-responsive) to sensory input. These disruptions can also result in challenging behaviors across a variety of situations, including situations where cognitive demands are high.

- **What is hyper-responsiveness?** Hyper-responsiveness - When a person is over-responsive to sensory inputs, they tend to overreact to one or all of the sensory inputs (e.g. touch, smell, movement, vision, hearing, taste, and body position). This is also termed "sensory aversions." If a person is sensitive to touch, they may dislike the feel of certain types of clothing, not want to wear shoes, be sensitive to lights, and/or dislike the feel/temperature of certain foods. They may also have a very difficult time with medical and/or dental procedures, having their hair brushed or cut, and having their teeth brushed. Some individuals with sensory aversions may hit others who get too close to them, may throw objects that they do not like the "feel" or taste of, and may scream/yell, and/or cover their ears when overwhelmed by sensory inputs. As applied to classroom based situations, individuals who are over-responsive to touch may not respond well to

"hand-over-hand" physical prompts to teach them new tasks. It may take certain individuals some time to calm down after being on "sensory overload."

- **What is hypo-responsiveness?** Hypo-responsiveness - When a person is under-responsive to sensory inputs, they may seek out stimulation by mouthing/biting objects, seek out deep pressure by giving tight hugs and/or engaging in more physical play with others, seek movement (e.g. spinning), and it may seem as though they have trouble hearing. They may, for example, hit another person to get them to play and/or greet them. These behaviors can also be called "sensory-seeking" behaviors. Individuals who are hypo-responsive may not respond to pain/differences in temperature, and they may be highly attracted to lights/shiny objects and may examine things from close visual range (e.g. putting their nose/face right up to the television screen).

## Sensory Issues in Angelman Syndrome

**What do we know about sensory issues in Angelman syndrome?** Sensory issues in individuals with AS - We know from studies of basic neuroscience that sensory processes are disrupted in AS. The Ube3a protein is necessary for maintaining the brain's response to sensory experiences during development. Clinical studies of individuals with AS have demonstrated that sensory-seeking behaviors (e.g. biting, mouthing, fascination with water, examining shiny objects, high interest in plastic) are common in AS. Different individuals may have varying degrees of sensory-seeking behaviors. The severity of these behaviors may also differ on a day to day basis. A smaller percentage of individuals with AS also exhibit sensory aversions (sensitivity to noise, sensitivity to certain foods, clothing, etc.). In these cases, a child may retreat from social interactions or loud situations.

**What can I do about treating sensory issues?** Teachers, occupational therapists, and some behavioral therapists are accustomed to treating and/or accommodating sensory issues. An individual who commonly mouths/bites objects may, for example, be given a "chew tube" to wear around his neck accompanied by the instructions: "bite this, not this." Individuals who are easily overwhelmed in crowded/loud settings may benefit from a smaller group setting or a quiet space where they can retreat to when they are overwhelmed. Some individuals may be calmed when wearing a weighted vest and/or blanket. Music may be calming and/or reinforcing to some children who have sensory issues. Therapy may also incorporate movement (e.g. swinging, spinning, etc.). In addition, limiting choices to no more than 2-3 options may be helpful to children who have more sensory issues (i.e. they are less likely to become overwhelmed).

Those who have extreme sensitivity to textures, including food textures, may be gradually introduced to a variety of textures within a therapy context (e.g. grass, shaving cream, crunchy chips, etc.). It is important to note that most "sensory-integration therapy" is not an evidenced-based treatment. This means that there is not a scientific basis for these forms of intervention, and they cannot be guaranteed to produce improvements in an individual's behavior. Nonetheless, parents of individuals with AS have anecdotally reported improvements for their children as a result of various forms of sensory-integration therapy. In addition, some parents have noted improvements in the severity of sensory issues as individuals become older (i.e. extreme responses to sensory stimuli become less severe into adolescence and adulthood). This has been demonstrated through research in children with autism spectrum disorders.

As we noted, difficulties with sensory processing are a common part of AS. We have also found, over time, that some individuals with AS exhibit features of autism spectrum disorders. We are now going to discuss autism and its relevance to AS.

## **Autism Spectrum Disorders**

**What is Autism?** Autism - Autism spectrum disorders (ASD) (use autism/ASD interchangeably) are complex, heterogeneous disorders that likely result from a variety of complex combinations of genetic and environmental factors. ASD is characterized by deficits in social affect accompanied by restricted and repetitive/stereotyped behaviors.

**Question - how is autism diagnosed?** It is a behavioral diagnosis that is made based on direct observations of an individual's behaviors. There is currently no blood test or medical test (e.g. X-Ray, MRI) that determines whether or not an individual has autism. Physicians may order genetic testing to look for a genetic cause, and they may recommend other tests or studies to determine if other medical conditions are present (e.g. seizures). Often, behavioral observations are done through a formal evaluation with a psychologist. Information about behaviors is also gathered from parents, teachers, and/or therapists in addition to direct observation in order to make a formal "diagnosis" of autism.

**Question - what is social affect?** Social affect refers to the appropriate use of eye gaze to regulate social interactions, using a facial expressions, shared enjoyment in interactions, use of nonverbal gestures, and the quality of a social approach. For example, an individual may approach someone else to obtain an object, but not look directly at them, instead being focused primarily on the object of interest. They may hit the other person in greeting, may not use any facial expressions or nonverbal gestures (e.g. pointing, signing), or may even use their hand as a tool to pull the object toward them. Even though this individual approached another person, they clearly have deficits in their social affect skills since the "quality" of this interaction is impaired.

**Question - what are repetitive/stereotyped behaviors?** Repetitive/stereotyped behaviors may include hand-flapping, finger-flicking, lining up of objects, and/or spinning of objects. The term "restricted interests" refers to a strong, narrow interest in certain toys or topics (e.g. trains, dinosaurs, birds, cars, plastic bottles) that impair the quality of social interactions with others. An individual may seem "obsessed" with these toys/topics and may have a difficult time with making a transition when these objects are taken away. During times of transition, they may exhibit challenging behaviors. There may be varying degrees of impairments in social affect and restricted/repetitive behaviors, and thus there are varying degrees of "autism."

## **Autism in Angelman Syndrome**

**How does autism apply to Angelman syndrome?** Autism in AS - there is a subgroup of individuals with AS who exhibit enough features of autism such that this warrants an additional diagnosis. Our research has shown that those individuals with AS who tend to have autism exhibit larger deletions

(Class 1 deletions). Individuals who have both AS and autism do not differ from their peers with regard to cognition or adaptive behaviors, but they do have more impairments in their social affect and have more repetitive behaviors than their peers with AS alone. They also seem to have more sensory aversions (i.e. a hyper-responsiveness to sensory inputs). It is important to note that having a low cognitive level alone does not drive an additional diagnosis of autism in AS; careful observation of the behaviors previously described is essential in making this determination. Thus far, our longitudinal follow-up of features of autism in AS reveals that in most cases, the ASD features persist over time. Regression (or loss of skills) is not typically associated with a diagnosis of autism in AS.

**How do you treat autism in Angelman syndrome?** To treat features of autism in AS, we typically recommend applied behavioral analysis (ABA). ABA is a scientifically based (i.e. evidence-based) treatment where the overall goal is to modify behaviors through changing the relationship between the behavior and the environment. Individuals with autism tend to be more responsive to visual cues, preparation for transitions (by using visual schedules, timers, etc.), and immediate/tangible rewards that are associated with performing desired actions. The details of ABA are discussed in another module.

## Case Study: Bethany

### Case Study: Bethany

Bethany is a 6-year-old girl with Angelman Syndrome (deletion positive). She lives with both of her parents and her typically developing sister. Over the past year, her parents noted that she has made several improvements with regard to her attention span, and her ability to sit still. She also seems to understand more. She has also started to become more independent with her toileting. She currently receives speech therapy school support services, where they are working on using pictures. She receives occupational therapy twice per week, and also receives physical therapy. She uses a friend's iPad and may use an iPad at school, although this is not always integrated throughout her day. In spite of these clear improvements in cognition, language, and adaptive behavior skills, her parents have become increasingly concerned about her hitting and pinching behavior both at home and at school. This behavior has worsened significantly in the last few weeks. Her parents are unsure of whether changes to her medication regimen could be contributing to these behavioral difficulties. Due to her exhibiting these behaviors in school, they will have a behavior consultant attend the classroom and track her behaviors. In general, she gets along with her peers. She becomes anxious when in medical settings. Recently, a family member was hospitalized and she became very upset when seeing this individual in his hospital bed.

On formal evaluation, she walked with her parents toward the evaluation room but became very anxious upon seeing the room and examiners. She clung to her parents and vocalized. Upon entering the room, she sat on her mother's lap. She warmed up when shown an iPhone application that played music, though at times she covered her ears (seeming sensitive to the sensory inputs). She made good eye gaze, coordinated with vocalizations and facial expressions to display her like or dislike of the music. It was quite clear that she does not have autism. She was able to transition to testing at the table though needed her mother to sit next to her. She showed good attention for tasks that were easy for her. However, once tasks became difficult, she threw the test materials or hit the examiners. She vocalized and reached for her mother to try to avoid testing. She was responsive to "If-Then" statements paired with visual cues (real objects), re-direction, ignoring of inappropriate behaviors, and some use of visual cues when paired

with verbal prompts. She was somewhat tactile defensive when she was upset, and did not like the use of hand-over-hand physical prompts. She frequently gave objects to her mother or the examiner when she was done with them. She mouthed several objects during the assessment. She flapped her arms when excited.

In addition to some of the strategies that have been discussed throughout this module, we recommended that the child's parents/teachers/therapists work to track her behaviors within the context of any changes to her medications. Specifically, we suggested the use of a functional behavioral analysis, with careful attention to when her behaviors seem to be better and what is occurring during that time. Within the context of the evaluation, her behaviors were quite manageable when she was being presented with tasks that were within her developmental level, but she exhibited several challenging behaviors as tasks became harder. Once the more detailed information has been gathered, we suggested that her parents share this information with the physicians who are monitoring her medications so that any necessary adjustments can be made as needed. They should also monitor any medical issues within the context of significant changes to her behavior.

## Summary

### Frequently Asked Questions (FAQ)

**Q. How do you determine "cognition" in a nonverbal individual?**

**A.** We administer tests that are most suitable to developmental level as opposed to chronological age. There are certain tests that are geared toward individuals who are nonverbal. It is very important that standardized tests are used, because those are given the same way by people around the world, and the scores were therefore developed using populations of individuals around the world. We do encourage parents to bring any assistive technology device or communication aide (e.g. PECS, visual schedules, etc.) and where permissible, we provide ample time for responses (given motor planning issues).

**Q. How would knowing my child/adult's cognitive level help? They are much smarter than what any "test" says.**

**A.** It may help select activities that are developmentally appropriate for them, and may allow all involved in their care to know how best to teach/build new skills. We consider this a formal manner by which to assess a "baseline" level for beginning intervention. For example, if a child who developmentally does not yet attend to real photographs is introduced to PECS, they may become extremely frustrated and may be averse to learning that skill. This is considered as a "snapshot" of functioning within a standardized setting, and may also assist in acquiring appropriate, intensive therapy services.

**Q. How do I get sensory integration therapy?**

**A.** Sensory integration therapy is typically provided by occupational therapists. A physician or psychologist may recommend a formal evaluation for occupational therapy. Also sensory-seeking behaviors and/or aversions may be noted by a teacher or therapist. Depending upon the outcome of that formal evaluation, sensory integration therapy may be warranted.

**Q. Does it really matter whether autism is formally diagnosed in my child with AS? Shouldn't they get the services they need anyway?**

**A.** In many states within the United States and countries around the world, if an additional diagnosis of autism is warranted, it opens the door to more intensive therapeutic services. In some cases, individuals are able to receive applied behavioral analysis services and this may be covered by insurance if there is additionally a diagnosis of autism. Individuals with AS benefit from applied behavioral analysis regardless of a diagnosis of autism, but it may be easier to acquire services if autism is formally diagnosed (if warranted).