ANGELMAN SYNDROME: IMPLICATIONS FOR PHYSICAL EDUCATION AND OTHER MOVEMENT SETTINGS

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Abstract

The primary purpose of this review of the literature is to improve the support given to those with Angelman syndrome in physical education and other movement settings. The additional purpose is to call attention to a rare neurodevelopment disorder that people are often unfamiliar with, by introducing you to some children with Angelman syndrome, explaining the genetic mechanisms in layman terms, and addressing concerns when learners with complex disabilities are included in physical education and other movement settings.

Keywords: Angelman syndrome, UBE3A gene, Chromosome 15, neurologic impairment, neurogenetic imprinting disorder, neurodevelopmental disorder, genetics, rare genetic disorder, Physical Education

My daughter, Maggie, was born 10 weeks premature after six weeks of a slow amniotic fluid leak. She spent the next six weeks in the NICU and two additional one-week stays in the PICU for feeding difficulties and complications coming off of anesthesia after a hernia repair, all before three months of age. Jamie, Maggie’s twin, was able to leave the NICU after only four weeks and never allowed her prematurity to slow her down. I was thankful to have twins to compare and highlight their differences. I knew in my gut that Maggie was not a typically developing child. Many would encourage me, saying, “Just give her time.” However, the old expression of “a mom knows” held true for me.

Based on my previous experiences as an Adapted Physical Education Specialist for a K-12 school district, and five summers at a camp for children with special needs, I suspected at 11 months of age that my daughter had Angelman syndrome due to her excessive laughter and increasingly evident developmental delay. She was seen by a developmental pediatrician at 13 months of age and given a diagnosis of cognitive impairment. The developmental pediatrician requested a brain MRI and a Chromosomal Microarray Analysis (CMA) to guide him in his diagnosis. Because this recommendation was made during cold and flu season, our family chose to follow the advice of our general pediatrician by waiting until the summer to do further testing as Maggie habitually turned routine outpatient hospital visits into five- to seven-day inpatient stays. When Maggie was 20 months old, the CMA and a methylation study were finally complete and provided the genetic diagnosis of Angelman syndrome with paternal uniparental disomy (UPD) as the genetic mechanism.

What is Angelman Syndrome?

Angelman syndrome (often abbreviated AS) is a neurodevelopmental disorder where neurons in the brain lack the expression of, or express a nonfunctional, UBE3A protein. Neurological disorders and diseases affect the brain, spinal cord, and/or the nerves that connect to these structures. In AS, there are general defects in all parts of the nervous system, which lead to clear differences in many areas because parts of the nervous system do not function as one would expect them to in typically developing children. All genotypes are characterized by a uniform phenotype marked by profound cognitive impairment, movement, or balance disorder (ataxia), speech impairment with minimal or no use of words, hyperactivity, and a friendly, happy demeanor. Often times, but not always present, are epilepsy, interrupted and short periods of sleep, and subtle dysmorphic facial features (Clayton-Smith & Laan, 2003; Jones, Jones, & Del Campo 2013; Kyllerman, 2013; Mayo Clinic, n.d.; & Williams et al., 2006). As a result of the friendly, happy demeanor and jerky movements, AS was historically referred to as “Happy Puppet syndrome” (Clayton-Smith & Laan, 2003, p. 87). However, this designation is derogatory; thus the suggestion to refer to this condition as “Angelman syndrome” after Harry Angelman, the first physician who described it in the literature, was proposed in 1982 (Williams & Frias). There are some general clinical (phenotypic) differences associated with the various genotypes1 (see Table 1). AS is often misdiagnosed at the clinical level as autism or cerebral palsy due to similarities in characteristics (PsychNet-UK, n.d. & Peters, Beaudet, Madduri, & Bacino, 2004).

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1 Phenotype is the face of the syndrome or what it looks like. Genotype is the genetic makeup that cannot be seen.
Table 1
Genetic Mechanisms Causing Angelman Syndrome and Associated Phenotypic Differences

Deletion – frequency ~70%
- Most severely involved in regard to microcephaly, seizures, relative hypopigmentation, motor difficulties (e.g., ataxia, muscular hypotonia, feeding difficulties), cognition, and language impairment

UPD – frequency 2-5%
- Better physical growth (e.g., less likely to have microcephaly)
- Lower prevalence of seizures
- Less movement and ataxia abnormalities
- Better cognition and less language impairment

ICD – frequency 3-5%
- Better physical growth (e.g., less likely to have microcephaly)
- Lower prevalence of seizures
- Less movement and ataxia abnormalities
- Highest cognitive, receptive language, fine motor, and gross motor abilities

UBE3A Mutation – frequency 5-10%
- Intermediate severity
  - Fall between the deletion and ICD classes in terms of microcephaly, seizures, motor difficulties, and language ability (more similar to the deletion group)
  - More closely resemble the UPD and ICD classes in terms of other clinical features

Other chromosome rearrangements – frequency 1-2%

Unknown – frequency 10-15%
- No evidence for deletion, imprinting defect, UPD, or UBE3A mutation was found in genetic testing and patients are given a clinical diagnosis

(Williams et al., 2006 & Thibert et al., 2013)

Analogy

There are several genetic mechanisms (or genotypes) that can disrupt, inactivate, or lead to the absence of the UBE3A gene on the maternally contributed chromosome 15, thereby causing AS. When explaining the four most common mechanisms that are known to cause AS, it is helpful to describe a more familiar situation in which compact discs with music are brought on a road trip. For the sake of the analogy, a male and a female are traveling, and both are to bring along 23 compact discs (which represent the 23 chromosomes). Because AS affects the UBE3A gene on the maternally contributed chromosome 15, that will be the “song” on the compact disc that the travelers will attempt to play. For the majority of travelers, the compact disc is inserted into the car’s device and it plays as expected. However, for 1 out of every 15,000 (Williams et al., 2006) discs, a problem will occur such that this particular song on the compact disc cannot play. This problem will either be due to a deletion (Del+), uniparental disomy (UPD), imprinting center defect (ICD), or a mutation in the UBE3A gene that results in AS.

Recall that the travelers in the analogy are male and female. This is important as AS and its chromosomal counterpart, Prader-Willi syndrome (PWS), provide the strongest evidence that in some instances the contributing parent of a chromosome matters greatly (Weiss, 1989). Weiss (1989) explains in a cleverly titled short article, “Prader Lacks Fader; Angelman Misses Mom?” how the sister disability (PWS) differs greatly in appearance while they are genetically so close. PWS is more like Down syndrome with the addition of an insatiable appetite than the two more common disabilities of CP and Autism for which AS is often mistaken. Typically, the gene for UBE3A on chromosome 15 is “turned on” when inherited from the mother (the song on the disc can be played) and “turned off” when inherited from the father (the song on the disc cannot be recognized) through a process called imprinting. When the paternally contributed chromosome 15 is affected, PWS results; however, when the maternally contributed chromosome 15 is affected, AS results.

Returning to the compact disc analogy, the male traveler may ask that his track (UBE3A) be played since the female’s is not working. This is a nice gesture. However, because the male’s UBE3A gene is “turned off” in the brain when inherited, the track cannot play; only the female’s song from the compact disc can play. With this understanding of genetic expression, the rest of this analogy will focus on only the 15th compact disc brought by the female traveler.

Types of Genetic Expression in AS

In the case of a deletion, the female traveler brought the compact disc, but the specific UBE3A song was deleted. The maternal copy of chromosome 15 that encompasses the UBE3A gene simply is not there. This group represents approximately 70% of the AS population, and patients with deletions are typically more severe in terms of phenotype (Williams et al., 2006).

In the case of a paternal UPD, the male traveler brought an extra copy of compact disc number 15. With three CDs and only room for two, the female’s CD is tossed out the window. This phenomenon is referred to as “trisomy rescue,” which allows the baby to survive, since three copies of chromosome 15 would otherwise result in early miscarriage (Williams et al., 2006). Even though there are two copies of compact disc number 15, both are from the male traveler and neither will play because the song cannot be recognized on either disc. Typically, a back-up copy is beneficial, but this is not the case when the region is imprinted. This group represents roughly 2-3% of the AS population and is typically milder in terms of phenotype (Williams et al., 2006).

In the case of an imprinting center defect (ICD), the compact disc brought by the female traveler was burned with the wrong format (e.g. MP3, data, etc.). In essence, the female traveler’s compact disc looks just like the one brought by the male traveler and thus will not play the song. From a genetic standpoint, the imprint was changed. Even though the baby inherits one copy of chromosome 15 from each parent, the female’s chromosome carries a male imprint and is “turned off.” This group represents roughly 3-5% of the AS population and is typically the mildest in terms of phenotype (Williams et al., 2006).

In the case of a UBE3A mutation, the compact disc brought by the female traveler was damaged. For example, the compact disc could have a scratch on it, preventing the UBE3A track from playing correctly. This group represents roughly 5-10% of the AS population and has the greatest variability in terms of phenotype (Williams et al., 2006; Thibert, Larson, Hsieh, Raby, & Thiele, 2013).
Medical, Motor, and Social Implications of Angelman Syndrome

The UBE3A gene is located in the q11-13 region of chromosome 15. As part of normal fetal development, the UBE3A gene is imprinted such that only the maternal contribution is available in neurons. Scientists know that the UBE3A gene “plays an integral role in the cellular ubiquitin-proteasome pathway, and is critical for synaptic development and neural plasticity” (Thibert et al., 2013, p. 271). The ubiquitin-proteasome pathway functions to tag and remove unneeded or damaged proteins from cells. The exact mechanism for how loss of UBE3A causes the brain dysfunction in AS is not well understood, but researchers are trying to identify therapeutics that may alleviate symptoms associated with AS, or reactivate the silenced paternal copy of UBE3A. Because their work has demonstrated that providing UBE3A into the adult mouse brain can reduce or eliminate AS symptomology (Daly et al., 2011), efforts to reactivate the paternal copy of UBE3A holds exciting promise. The goal is to make the UBE3A gene present on the paternal “compact disc” available so that the brain can access the information.

Medical Concerns and Management

Infants with AS often have difficulty with feeding, including problems with uncoordinated sucking, tongue thrusting, and poor breast attachment. Shortly after feeding, gastroesophageal reflux can occur (Williams et al., 2006). Frequent arching of the back after feeding can indicate a problem with reflux. Gastroesophageal reflux disease (GERD) is frequently controllable by the correct diagnosis and medication. Seizures are frequent, seen in roughly 80% of patients, and are often difficult to control with medication, although dietary interventions can be particularly effective (Thibert, Pfeifer, Larson, Raby, Reynolds, Morgan, & Thiele, 2012). Onset of seizures varies, but typically begins in early childhood (Clayton-Smith & Laan, 2003). It is helpful to take a short video clip of suspected seizures to show to a neurologist.

Dan and Chéron (2005) warn of intention tremors that appear when a child is concentrating on a motor task and note that there is a strong correlation between these tremors and bursts of electrical activity in the muscle. These bursts of electrical activity in the muscle are sometimes interpreted as potential epileptic activity; however, intention tremors are not of epileptic origin as there is not an accompanying burst of electrical activity in the brain (Dan & Chéron). It is more likely that it is the ataxia presenting itself and may be a way that the child’s muscles compensate for a lack of coordination. For example, when a child with AS is learning a new skill such as sitting, balance and smoothness of movement are initial concerns of therapists and parents alike. However, those aware of these intention tremors will rightly not be concerned, but instead provide ample movement opportunities and time for the child to refine the balance and learn to coordinate the movements smoothly.

Physical and Motor Abilities and Accommodations

Balance and smoothness of movement are difficulties that nearly all individuals with AS experience. Many have an ataxic gait, spasticity of the distal lower limbs, and mild tremors (Thibert et al., 2013). Appropriate therapies are often needed to address physical and neurological concerns, and special education services are often required for educational needs (Clayton-Smith & Laan, 2003). High muscle tone contributes to stiffness and poor balance. OTs and PTs should be consulted to stretch affected muscles and strengthen opposing muscles. Low muscle tone often indicates a deficit in the proprioceptive system and causes fatigue, poor coordination, and clumsiness. OTs and PTs should be consulted to strengthen muscle groups.

The most significant and challenging physical limitation of those with AS is their characteristic gait: wide base of support and feet pointed outward with arms up and out to the side. This adaptive gait is the natural solution to stabilize posture (Dan & Chérion, 2005) in the face of ataxia, motor planning, and balance disorders common to AS. While balance-improving strategies do work for those who learn to walk, it is important to remember that each step is a conscious balancing act that requires a great amount of cognition. The effort put into a cognitive task reduces the available cognitive capacity to attend to other skills (see Figure 1). Automaticity of gross motor skills should remain a focus with the help of professionals such as a podiatrist.

Muscle tone affects the gait of those with AS. Half of all infants with AS have reduced muscle tone, and about one-quarter continue to experience moderate to severe global hypotonia throughout childhood (Thibert et al., 2013). About one-third of students with AS have mixed muscle tone disorder (hypertonic spastic lower limbs and truncal hypotonia), which results in a stiff gait and jerky arm movements (Clayton-Smith, 2010; Thibert et al., 2013). Like walking, sitting can require great amounts of cognition due to low muscle tone. In this case, supportive seating that is not too restrictive (remember our muscles were made to move; not be restrained) should be considered in conjunction with physical therapists (see Figure 2). Table 2 organizes considerations and modifications to common activities in the physical education setting by learning characteristics of those with AS.
<table>
<thead>
<tr>
<th>Learning Characteristics</th>
<th>Considerations</th>
<th>Modification</th>
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<tr>
<td><strong>Communication</strong></td>
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<td>Profound expressive language disability</td>
<td>Do not mistake what a student with AS can express to be the same as what that student can understand. Students with AS have limited cognitive ability and do not know how to interact purposefully with peers or equipment. For this reason, teacher-directed teaching styles should be used over guided discovery or cooperative learning. According to Pangrazi and Beighle (2013), mastery learning “is well suited for students with disabilities” (p. 40).</td>
<td>Individuals using the most effective communication systems have the least challenging behaviors (Clayton-Smith, 2001). To modify for limited speech and communication skills, use smaller groups to increase engagement in the activity; allow them to watch another student perform the skill first; use demonstrations (physical assistance as needed), pictures, simple terminology, or fewer words; and check for understanding in a safe setting. Use cues with strong visual, tactile (manual guidance), and auditory stimuli (Auxter et al., 2010), active demonstration, parallel talk (Pangrazi &amp; Beighle, 2013, p. 131). “Keep verbal directions to a minimum. They are often ineffective when teaching children who are more severely disabled... The less ability the child has to communicate verbally, the more manual guidance should be considered as a tool for instruction (Auxter et al., 2010, p. 373). Metzler describes active demonstration as the students following “along as the teacher talks and demonstrates (p. 85). This demonstration can also be done in slow motion. When using parallel talk, “the teacher says, “Tyler is throwing the yarn ball.”” (Pangrazi &amp; Beighle, 2015, p. 131) as the student throws the ball.</td>
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<td>Cognition</td>
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<td>Intellectual disability</td>
<td>Disability is specifically related to challenges in memory formation. Individuals with AS have a strong memory for social interactions. There are different approaches to teaching. Provide structure, high levels of organization/daily routine, and learning centers/stations.</td>
<td>Focus on supports for memory formation such as repeated sequencing, the use of strong rhythms, and teaching cues. Activate background knowledge to ensure that students are building on pre-existing schemes rather than constructing new schemes for each new experience. When applying the bottom-up approach, the teacher will “select activities that appeal to the child and use those until the deficits are eliminated” (Auxter et al., 2010, p. 163). When applying the top-down approach, the teacher will “program activities at the highest level of dysfunction... and probe down into contributing components for deficits” (Auxter et al., 2010, p. 163). Metzler (2011) suggests posting “an overview of the lesson plan in the locker room or entrance to the gym so that students know what to expect in the lesson” (p. 79). Visual/pictorial schedules help children to know what to expect and minimize distractions. Those with AS often have a shorter attention span so less time should be spent on more activities during class time.</td>
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<td>Global dyspraxia</td>
<td>Students may find it difficult to imitate familiar gestures on command (Penner, Johnston, Faircloth, Irish, &amp; Williams, 1993).</td>
<td>Use a visual schedule to plan what activity is coming next. Natural contexts/authentic assessment is generally the best way to demonstrate knowledge.</td>
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<td>Physical abilities</td>
<td>Fine motor skills tend to be more difficult for those with AS than gross motor movements (Williams, Driscoll, &amp; Dagli, 2010). Those with AS generally walk with a distinctive broad-based gait with upraised arms and flexed wrists. This gait is a strategic, adaptive response to stabilize posture (Dan &amp; Chéron, 2005).</td>
<td>Focus on increasing the object size when fine motor skills are required.</td>
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| Ataxia/Unintentional movements | Children with AS generally have ataxia (difficulty coordinating muscular movements resulting in jerky movements such as an unsteady gait). | Modifications such as allowing physical supports, increasing the width of balance beams, or using tape on the floor instead of balance beams will help (Block, 2007).

Teach students how to improve their balance by lowering their center of gravity, widening their base of support, using eyes optimally by focusing on one spot (generally on the floor a few feet away), and keeping as much of the body in contact with the surface as possible (Block, 2007). |
<p>| Apraxia (Disorder of Motor Planning in which individuals cannot perform a motor skill although they are willing, able, and understand the task.) | Motor apraxia results in difficulty planning and executing a movement pattern and may cause individuals with AS to move slower than their peers. | Allow the student with AS to perform a simpler skill while their peers perform a more complex skill to even the competition (Block, 2007). |
| Dyspraxia (Disorder of Motor Planning in which individuals have immature motor skills and difficulty organizing their movement.) | Students with AS demonstrate a “central dyscoordination resulting in difficulties in positioning the body and interacting with the environment” (Beckung &amp; Kyllerman, 2005, p. 143). Ceaseless movement is a common symptom of dyspraxia. It is as though they are gearing up for their next move. Dyspraxia explains why individuals with AS can perform a skill one day and then not perform it another day. | Students who experience dyspraxia may benefit from approaches such as backward chaining (Bauman &amp; Kemper, 1994) such as standing at the foul line and rolling a bowling ball before learning to add successive steps. Backward chaining is excellent for any skill that is rewarded at the end of the sequence – knocking down pins. Students benefit from explicit supports to imagine and then plan their physical actions. For some students, physical touch on the body part that needs to move will help the student organize the related movement, such as touching the ankle before lifting the foot to climb stairs or touching the shoulder before writing. To accommodate for limited coordination and accuracy, use larger, lighter, softer balls and larger striking implements; decrease distance the ball is thrown/kicked or increase size of target; and in bowling-type games use lighter, less stable pins (Block, 2007). |
| Abnormal/Disrupted muscle tone | Students who experience dyspraxia may benefit from approaches such as backward chaining (Bauman &amp; Kemper, 1994) such as standing at the foul line and rolling a bowling ball before learning to add successive steps. Backward chaining is excellent for any skill that is rewarded at the end of the sequence, knocking down pins. Students benefit from explicit supports to imagine and then plan their physical actions. For some students, physical touch on the body part that needs to move will help the student organize the related movement, such as touching the ankle before lifting the foot to climb stairs or touching the shoulder before writing. To accommodate for limited coordination and accuracy, use larger, lighter, softer balls and larger striking implements; decrease distance the ball is thrown/kicked or increase size of target; and in bowling-type games use lighter, less stable pins (Block, 2007). | Shorten the distances in running games (Block, 2007). Additional rest breaks between turns can help. Additionally, longer distances for those who excel can help to even the competition (Block, 2007). Challenge by choice is a good way to accomplish this. |</p>
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<tr>
<th>Senses</th>
<th>Central auditory processing disorder (Hearing impairments)</th>
<th>Vestibular</th>
<th>Proprioceptive</th>
<th>Affect</th>
<th>Attention</th>
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<td>Cortical vision impairment (visual impairments)</td>
<td>Sensory integration dysfunction may cause those with AS to struggle with distinguishing the sounds of oral speech from background noise in the gym.</td>
<td>Vestibular input provides information about our bodies in space as one moves and maintains balance, which is important for organizing and maintaining attention.</td>
<td>Proprioceptive input is received through the joints as one engages in weight-bearing activities or receives deep pressure, which is important for organizing and maintaining attention.</td>
<td>Students tend to have strong behavioral flexibility that allows them to adapt to changes (Didden et al., 2008) and are prone to overexcitement.</td>
<td>Attention skills are consistent with overall development. Challenges can arise from distractibility due to difficulty filtering out competing stimuli.</td>
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<td>Use high contrasting colors for equipment (Block, 2007).</td>
<td>Sensory integration dysfunction may cause those with AS to struggle with distinguishing the sounds of oral speech from background noise in the gym.</td>
<td>Jumping, swinging (including swinging side-by-side as well as front-to-back), and rocking are movements that provide vestibular input.</td>
<td>Create heavy work (such as carrying objects or wheelbarrow walking) to provide proprioceptive input (Block, 2007).</td>
<td>Students tend to have strong behavioral flexibility that allows them to adapt to changes (Didden et al., 2008) and are prone to overexcitement.</td>
<td>Video-based instruction (or a show-and-tell approach) and hands-on learning activities such as the use of manipulatives, tangible objects, and materials with appealing sensory properties (i.e., shiny or crinkly textures) includes multiple senses and helps with distractibility.</td>
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<td>Control lighting and prevent glare (Block, 2007).</td>
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<td>Know what motivates each child. Social time with peers, electronic time, water-play, or play with crinkled paper (cellophane) are common fascinations for those with AS (Williams et al., 2006). Use motivators to decrease frustration or disinterest when activities are challenging for them. Create ways for students to have a sense of mastery, community among classmates, personal value, influence, and hopefulness for future classes/activities.</td>
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<td>Provide verbal or auditory cues in lieu of visual or physical cues such as body language (Block, 2007).</td>
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<td>Create break stations for students to escape from sensory input (Block, 2007).</td>
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<td>Realize students with AS may be paying close attention even if they are not visually attending.</td>
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**Figure 1.** Maggie (22 months of age, UPD) is all smiles as she learns to use a walker to assist her mobility with Sheri Baker, PT. In this photo, Maggie is working on transitioning from a slower paced carpet surface to a faster paced hard surface.  

**Figure 2.** Sophie S. (14 months of age, del+) using pool noodles, towels, and a laundry basket fashioned together as a supportive “seat” because her tone was too low to sit without assistance. This allowed her to reach forward and play with objects. It also increased her opportunities and ability to cross midline when reaching for toys.
Social Concerns and Motivation

Verbal communication skills are very limited to absent in those with AS. Individuals with AS are more successful with alternative and augmentative communication techniques. Clayton-Smith and Laan (2003) suggest that “acquisition of communication skills is often easier as patients get older and concentration span improves” (p. 88); however, language and literacy instruction should be a focus from the start. While individuals with AS typically have little to no spoken language, receptive language is often much stronger. Individuals with AS are generally very social (Heald, Allen, Villa, & Oliver, 2013), which can help motivate their learning of multiple methods of unaided as well as aided communication. Calculator (2013) describes unaided communication as “nonspeech vocalizations, natural gestures, and pulling a listener in the direction of a desired object that is out of reach as well as behaviors such as signs and enhanced natural gestures, which must be taught” (p. 557). He describes aided communication as nonelectronic, such as communication boards, or electronic aids, such as low-tech voice output communication aids (VOCA) and high-tech “systems which provide access to often unlimited numbers of spoken messages” (p. 558). Sign language is often difficult due to the impaired fine motor skills, but can be learned with additional practice and patience. Using natural gestures side steps the need for practice with the motor skills and allows for immediate teaching of the communicative meaning.

Suggestions for Physical Education and Movement Settings

Teachers in physical education or any other movement setting must consider their approach to teaching motor skills. One can either teach from a bottom-up (forward chaining) or a top-down (backward chaining) approach. It is generally accepted that for younger children with disabilities that affect learning, the bottom-up approach is preferred; and for older children, especially those with dyspraxia (complex motor planning difficulties) as is common in AS, the top-down approach is preferred (Bauman & Kemper, 1994). Using a bottom-up approach early will “develop their sensory-motor and perceptual motor systems, as well as help them learn the basic elements of fundamental movement skills” (Aaxter, Pyfer, Zittel, Roth, & Huettig, 2010, p. 372). Those who implement the top-down approach place the highest emphasis on the end of the skill sequence rather than what is to be taught next (Aaxter et al., 2010).

The bottom-up or developmental approach is based on hierarchical theories of motor control involving sensory-integration and perceptual motor training (Barnhart, Davenport, Epps, & Nordquist, 2003). The instruction is focused on the lowest level of motor function before specific skills. The goal is “studying the underlying biological processes governing maturation” (Gallahue, Ozmun, & Goodway, 2012, p. 7) known as the process, rather than “describing the mechanisms of various stages of movement skill acquisition, and developing normative criteria for a variety of motor performance measures” (Gallahue et al., 2012, p. 7) known as the product. For example, learners would engage in activities that build their balance, visual tracking, and cross-lateral integration. One might observe young children playing a game of tug-of-war with a blanket while sitting on the floor, slightly older children sitting on mats pulling a rope, and finally, mature children standing while pulling on a large rope. The key to success for the bottom-up approach is preparing individuals for fundamental movement skill readiness through a hierarchical progression of motor functions, which is why the bottom-up approach is preferred for younger children with disabilities.

The top-down or task-specific approach is influenced by the dynamic systems approach to motor learning (Barnhart et al., 2003). According to Gallahue, the “word dynamic conveys the concept that developmental change is nonlinear” (2012, p. 28) and is “seen as a discontinuous process” (p. 28) in which new patterns of movement replace old ones. This is quite the opposite of the hierarchical bottom-up approach that views motor development as smooth and “moving toward ever higher levels of complexity and competence in the motor system” (Gallahue et al., 2012, p. 28). The instruction is focused on direct teaching of a skill with lots of practice and repetition. The goal is the product, rather than the process.

One should not be confused with the difference between process (as defined when discussing the bottom-up approach) and task-specific intervention, which involves the breaking down of a skill into its component parts to be linked together as the product. There are several well-established special education strategies such as backward chaining, naturalistic predictable scripted sequences, and system of least to most prompts, which teaches the final step as part of the whole activity rather than a sudden performance demand (Collins, 2012; Donnellan, Hill, & Leary, 2013; Kliweer, 2008). For example, learners would engage in activities to improve each component part of a fundamental movement skill such as throwing. One might observe young adolescents practicing stepping with the foot opposite of their throwing arm (balance), a backswing and trunk rotation that begins with a movement away from the intended line of flight and then a forward movement toward the target (visual tracking), and finally a follow through of the arm that crosses the center of the body (cross-lateral integration). The key to success for the top-down approach is repetition and practice of the fundamental movement skill resulting in a refined end product, which is why it is preferred for older children, with or without disabilities.

Students who experience kinesthetic dilemmas (Bauman & Kemper, 1994), such as dyspraxia (complex motor planning difficulties), may benefit from well-established special education strategies designed to reduce the performance demand on the last part of a movement or step in a skill. Backward chaining, system of least to most prompts, and naturalistic predictable scripted sequences are examples of strategies that string a series of steps together while the focus remains on the task as a whole (Collins, 2012; Donnellan et al., 2013; Kliweer, 2008).
A Note about Inclusion with Peers

Pangrazi and Beighle (2013) stated it best when they described quality physical education as adapted physical education “because quality physical educators are constantly adapting lessons, equipment, and activities to meet the needs of all students, with or without disabilities” (p. 128). Physical education teachers experience additional difficulties when including students with disabilities into their program; they must give equal consideration to cognitive, physical, and social abilities. It is the combination of these three domains that draws many into the field of physical education or other movement settings as opposed to other instructional programs. With six reminders (see Table 3) from Pengrazi and Beighle (2013), everyone can experience successful inclusion/quality physical education (recall that this goes equally for those with and without disabilities).

Table 1
Reminders for Successful Inclusion

1. Help students with disabilities meet target goals specified in the IEP and participate in the general program of activities. This may call for resources beyond the physical education class, including special work and homework.
2. Stress what the child is capable of doing and focus on accomplishments. Eliminate established practices that unwittingly contribute to embarrassment and failure.
3. Foster peer acceptance by treating each student as a functioning, participating member of the class.
4. Concentrate on the student’s physical education needs and not on the disability. Stress fundamental skills and physical fitness qualities.
5. Provide continual monitoring and periodically assess the student’s target goals. Anecdotal and periodic record keeping are implicit in this guideline.
6. Be constantly aware of students’ feelings and anxieties concerning their progress and integration via consistent communication. Provide positive feedback as a basic practice.

(Pengrazi & Beighle, 2013, pp. 134–135)

Educational Services and Outcomes

Educational teams must always consider the whole child when planning their goals. Neither the development of functional motor skills nor academic learning should be prioritized over the other. With the use of low and high-tech Augmentative Alternative Communication (AAC) devices, all students should be given the opportunity to participate in academic instruction. Strengths in cognitive skills can be masked by developmental motor delays, which give reason to abandon the “readiness” approach. Presumed competence should be the motto even in the absence of expressive communication. This approach never results in denying students with greater dyspraxia access to academic instruction.

Additionally, it is important to remember that when changes are made to the environment, the child may no longer view the task as the same. The adult could interpret the behavior as the child willfully choosing not to perform the skill. However, based on brain behavior, it is more likely that the child does not understand what is expected of him/her. For example, a proud grandmother may say with great excitement to the mom, “Maggie just wiped her mouth!” and then turn to Maggie to request her to perform the task again. When executive functioning is low, the ability to override the natural instinct to avoid unpleasant tasks is decreased. Most children find that wiping their mouth is unpleasant, and is certainly the case for those with tactile defensiveness. Therefore, the child does not understand the need to comply with the request to perform a task that has just been completed because it is unpleasant and does not need to be repeated. A change to the environment, such as making another mess on the child’s face, could possibly lead to a more favorable response. Adults need to consider why the child is behaving as such and make every effort to alter the environment to illicit the desired response. Those with typical executive brain functioning will make the decision to perform for the sake of the performance and the praise (Buron & Wolfberg, 2008).

Conclusion

It is important to remember that while those with AS do have similar limitations, they are not all the same; some may have low tone, and others may have high tone. Those responsible for physical education and other movement settings should enjoy getting to know the intricacies of all people they come in contact with and challenge each to expand on their present skills. What we do know is that those with AS do learn. It just takes longer and requires more repetition. Thus, sustained therapies even in the absence of progress may be necessary. The author of this article hopes to inspire those working in physical education and other movement settings to be creative in their approach to educating children with any disability.

Aya Lorenzana (14 years old, clinical diagnosis) on boogie board at Best Day Foundation’s Beach event.
Author’s Note

Maggie is learning so much every day. My husband and I marvel at the accomplishments that she makes. At 22 months of age, she figured out how to get back down to the floor after pulling-to-stand at the couch. We have been working on this for nearly 6 months! As I recall, her twin, Jamie, learned to pull to stand and get down almost in the same day. Maggie watches Jamie and tries to mimic everything she does. Developmental milestones come very slowly, but they do come! There are so many research opportunities in the area of movement settings to help children with Angelman syndrome continue to succeed.

Selected References


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