Drowning as a Cause of Death in Angelman Syndrome

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Abstract

Angelman syndrome is characterized by mental retardation, seizures, ataxia, inappropriate laughter, lack of speech, a particular facial appearance, and generally a chromosome 15q11-q13 deletion. Recently, a fascination with water and water-related activities has been reported in individuals with the syndrome. We report on a 9.6-year-old male previously diagnosed with Angelman syndrome who died unexpectedly by drowning in a shallow backyard wading pool. This case further illustrates the fascination with water by individuals with Angelman syndrome and highlights that this fascination may lead to death. We wish to alert careproviders that this fascination with water and water-related activities may contribute to death and that these individuals should be closely supervised when in the presence of water.

We read with interest the article by Clarke and Marston (2000) reporting the problem behaviors associated with Angelman syndrome. This syndrome is characterized by mental retardation, seizures, ataxia, inappropriate laughter, lack of speech, a particular facial appearance, and, generally, a chromosome 15q11-q13 deletion (Williams et al., 1995). Additional associated problems include overactivity, restlessness, eating disturbances, and sleep problems. Williams et al. also reported a fascination with water in 68% of the Angelman syndrome subjects reviewed in their survey. Because the characteristics of this syndrome include ataxia and poor coordination, seizures, lack of speech, and a fascination with water, caregivers should be alerted to the dangers raised by water and water-related activities for individuals with Angelman syndrome.

In this study, we report on a child with Angelman syndrome, confirmed cytogenetically with a chromosome 15q11-q13 deletion, who died at 9.6 years of age due to drowning. This young boy, whose mother had an unremarkable pregnancy, had a history of growth delays and developmental problems noted during the first 6 months of age. Hypotonia and a poor suck were noted at birth. He weighed 2.87 kg, was 50.8 cm long, and had a head circumference at the 3rd percentile at birth. At 9 months of age, gastroesophageal reflux was diagnosed along with failure to thrive. At 2.5 years of age, he developed seizures and a grossly abnormal EEG was reported. He was placed on an antiepileptic medication. He developed an ataxic gait with poor coordination and had severe developmental delays. His head circumference was less than the 3rd percentile at the time of his genetic evaluation at 6.7 years of age, and his weight was at the 3rd percentile and height at the 25th percentile. His hair was very blonde, and he had a fair complexion. He had not developed speech. His mother noted a fascination with water and sleep disturbances. Cytogenetic testing showed a chromosome 15q11-q13 deletion, which is seen in 70% of individuals with Angelman syndrome.

Unfortunately, at 9.6 years of age, this child died from drowning in a wading pool. His mother reported that the young boy left the house unnoticed and entered a 1.22× 1.83 m inflatable wading pool, which was 55.9 cm deep and located in the back yard. The pool contained between

27.9 and 38.1 cm of water at the time of the drowning. There was a flexible plastic top covering the pool. He was unattended for approximately 15 minutes when his mother noticed that he was not in the house and saw him floating in the pool, partially concealed by the pool cover. He was immediately removed from the pool but could not be resuscitated. Due to his history of seizures (although he had recently been under good control), a possible seizure may have occurred while he was in the wading pool. His mother noted that he had enjoyed the use of the wading pool many times previously while in the presence of others.

This case further illustrates the fascination with water by individuals with Angelman syndrome and draws attention to the fact that this fascination may lead to death. These individuals have poor coordination, ataxia, low muscle tone, and mental retardation, making it difficult for them to swim unassisted. Their high rate of seizures and nonverbal communication further increase the likelihood of water-related accidents. We wish to share this information with careproviders of individuals with Angelman syndrome in order to alert them of the fascination with water and health issues that may contribute to death during water-related activities. Persons with Angelman syndrome should be watched closely in the presence of water (even inflatable wading pools or bath tubs). Drowning may occur with increased frequency in this patient population.

We extend this warning regarding the possibility of submersion injury and/or death to children with epilepsy. The relative risk of drowning reported by Diekema, Quan, and Holt (1993) for children with epilepsy was 96 (95% CI 33 to 275) in the bathtub and 23.4 (95% CI 7.1 to 77.1) in the pool. In addition, patients with Romano–Ward syndrome, a form of congenital long QT syndrome, present with syncopal episodes and are at risk for sudden death. The diagnosis of Romano–Ward syndrome has been made in patients who present with near-drowning and drowning episodes (Ackerman, Tester, Porter, & Edwards, 1999; Harris, Knapp, & Sharma, 1992). People with other disorders associated with physical impairments, seizures, impulsive behavior, lack of speech, autism, and/or a fascination with water may also have an increased risk for drowning. For example, children with 5p- syndrome have a fascination with water and are impulsive and, as such, may also require close supervision around backyard pools and bathtubs.

The report of our patient should caution caretakers and alert healthcare professionals to warn families about the risk of accidental drowning by individuals with other types of mental retardation syndromes that share these risk factors (primarily seizures, fascination with water, and impulsivity).

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