Obstructive Sleep Apnea in a Child with Aicardi Syndrome

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Citation

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Abstract

We report the first known case of Obstructive Sleep Apnea (OSA) in a five-year-old girl with Aicardi syndrome and severe developmental delay, hypotonia, and agenesis of corpus collosum. This case report emphasizes the importance of identifying sleep disordered breathing in all patients with hypotonia and developmental delay, regardless of etiology and level of functioning. The goal of treatment is a potential positive impact on the sleep and wake time quality of life.

ABBREVIATIONS:

(AHI) Apnea –hypopnea index
(REM) Rapid eye movement
(EEG) Electro encephalogram
(RDI) Respiratory disturbance index

INTRODUCTION

Aicardi syndrome is a rare, congenital X-linked disorder with central nervous system abnormalities, which produce severe disabilities. We present a child with Aicardi syndrome, who had symptoms of a sleep disturbance and was found to have obstructive sleep apnea on polysomnography. This is the first reported case of Aicardi syndrome and obstructive sleep apnea.

CASE REPORT

A five-year-old girl with Aicardi syndrome was referred to us for polysomnography because of snoring and witnessed apneic events. She had severe global developmental delay, hypotonia, visual impairment and agenesis of the corpus callosum. She had suffered from neonatal apneic spells, anal stenosis and epilepsy. She was born at 37-38 weeks gestation via induced vaginal delivery with a birth weight of 6 lbs. Immediately after birth she developed apneic episodes with desaturation, followed by cyanotic breath-holding spells. She is functionally blind due to bilateral optic nerve hypoplasia. She has a severe anal stenosis, which was treated with a colostomy, followed by a bowel reconnection procedure at 2 years of age. She developed a seizure consisting of head deviation to the left, followed by generalized clonic activity at 2 years of age. The seizure lasted for less than one minute and was associated with high fever. Subsequently, she had similar seizures with laughter at the end of the seizures, as well as episodes of unprovoked, paroxysmal laughter, myoclonic jerks, and staring spells with eye-rolling. All of these episodes occurred many times per day. The seizures were well controlled with levatiracetam and valproic acid. She has severe developmental delay and is functioning at a 12- month level in all areas. She has behavioral problems, including mouthing, biting, hyperactivity and a short attention span. There is no known family history of any neurological or psychiatric illness.

On physical examination, the child had bilaterally low-set ears, high arched palate, down- turning of the mouth, klinodactyly of the left 4th toe and overriding of the 2nd and 3rd toes. She had frequent retroflexion of the neck. She had atrophy below the knees bilaterally. There was no growth retardation. Her head circumference was between the 25-50th percentile for age. Her neurological examination was significant for poor eye contact, lack of pretend play and mouthing. She was non-verbal other than crying. She had severe, diffuse hypotonia with tight hip adductors and severe head-lag on traction. She had good strength of all extremities. She was able to sit independently and walked with a broad-based, unsteady gait, holding on to a walker with both hands. She had diffuse hyper-reflexia without clonus. Investigations revealed normal chromosomal pattern of 46, XX. Magnetic resonance imaging of the brain showed agenesis of corpus callosum, colpocephaly and high riding of the third ventricle. No neuronal migration defects were seen. Continuous video-electro encephalogram monitoring revealed a variant of hemihypsarrhythmia during wakefulness with a generalized hypsarrhythmic pattern during sleep. X- rays of the cervical spine showed C2-C3

(cervical) vertebral fusion. Ophthalmology evaluation showed bilateral small and pale optic nerves consistent with optic nerve hypoplasia. Polysomnography demonstrated a sleep efficiency of 58% with spontaneous electrocortical arousals occurring at the rate of eleven per hour. The study revealed obstructive sleep apnea characterized by thirty-four apneas and hypopneas with an apnea-hypopnea index of 10 per hour. These events were exacerbated during rapid eye movement sleep (apnea-hypopnea index during rapid eye movement sleep =20 per hour). Oxyhemoglobin desaturations were mild, dropping from a baseline of 98% to a nadir of 92%. The amount of sleep fragmentation with disordered breathing, including apneas, hypopneas, and disruptive snores was moderate with respiratory disturbance index of 16 per hour. Treatment attempts are ongoing and include caregiver education with techniques for continuous positive airway pressure mask desensitization and behavioral modification.

DISCUSSION

Aicardi syndrome is characterized by the triad of agenesis of the corpus callosum, infantile spasms and chorioretinal lacunae ([1,2]). Vertebral and rib abnormalities are commonly associated. The disorder was thought to have an extremely poor outcome with persistent epilepsy, high mortality rate and very high rate of disability in survivors. Recent publications have emphasized that the disorder can have more varied clinical features than initially proposed and that the outcome can be considerably more variable with higher survival rates and lower rates of disability in more mildly affected individuals ([3]). Other, more newly recognized features of the disorder include cortical polymicrogyria, periventricular heterotopia, and cysts of the choroid plexus and/or the pineal or periventricular interhemispheric regions ([4]). Typical electro-encephalogram findings are gross asymmetry and asynchrony of the hemispheres with hypsarrhythmic in one hemisphere and burst suppression in the other hemisphere ([4]). The asynchrony tends to disappear during sleep, but normal sleep architecture is not seen. Although electro-encephalogram findings are well described in the literature, there are no published reports regarding sleep disorders in individuals with Aicardi syndrome. The severity of neurological impairments and difficulty in controlling epileptic seizures has probably overshadowed complaints of sleep disturbances in affected children. Children with Aicardi syndrome are at high risk for sleep disorder breathing because of the hypotonia and cervical spine abnormalities that are typical of the syndrome. Apnea/hypopnea may contribute considerably to intellectual disability and behavioral problems, such as hyperactivity. Considering the neurological findings and craniofacial disproportion in these children, the positive yield of the sleep study is expected to be high. This association is seen in other inherited developmental conditions such as Down syndrome, where the incidence of sleep disorders approach 80% ([5]). Performing sleep studies and treating patients with significant cognitive and developmental delay may be challenging. It is possible that early recognition and treatment might improve the outcome in these children.

CONCLUSION

We believe it is important to consider sleep disorders in patients with Aicardi syndrome. Daytime symptoms related to sleep disordered breathing, seizures or other sleep disorders may overlap with wake Aicardi syndrome symptoms. In particular girls suffering from milder forms of the disease should be considered for polysomnography with an added seizure montage to assist in the treatment plan of improved sleep and daytime performance.

References

1. Glasmacher MAK, Sutton VR, Hopkins B, et al. Phenotype and Management of Aicardi Syndrome. J Child Neurol. 2007;22:176-184.
2. Smith CD, Ryan SJ, Hoover SL, Baumann RJ. Magnetic resonance imaging of the brain in Aicardi's syndrome: report of 20 patients. J Neuroimaging 1996;6 : 214-221.
3. Aicardi J. Aicardi Syndrome: Old and New Findings. International Pediatrics, 1999, 14(1): 5-8.
4. Aicardi J. Aicardi syndrome. Brain Dev 2005; 27: 164-71.
5. Shott SR, Amin R, Chini B, et al. Obstructive Sleep Apnea: Should All Children With Down Syndrome Be Tested? Arch Otolaryngol Head Neck Surg, Apr 2006; 132: 432 – 436.

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