Autism Spectrum Disorder and Chiari 1 Malformation Co-occurring in a Child

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Very few studies have shown associations between autism spectrum disorder, attention deficit hyperactivity disorder and Chiari 1 malformation. Here, we report an 10-year-old male that presented after having seizures with a history of Chiari 1 malformation, autism spectrum disorder and ADHD with moderate mental retardation and speech delay. This case highlights the fact that autism spectrum disorder as biologically based neurodevelopmental disorder with altered brain growth may be associated with Chiari 1 malformation and ADHD.

Key words: Chiari 1 malformation, ADHD, Autism

INTRODUCTION

Autism spectrum disorder (ASD) is a disorder characterized by impairments in social communication, social interaction, restricted, repetitive patterns of behavior, interests, or activities [1]. Early presentation of the symptoms during early development is the norm with occasional case where symptoms may not become manifest until social demands exceed limited capacities.

Chiari 1 malformation (CM-1) is characterized by posterior cranial fossa disproportion leading to overcrowding and the descent of the cerebellar tonsils [2]. The commonest symptoms manifested by patients with CM-1 include occipital headache, neck pain due to raised intracranial pressure, cranial neuropathies, and dizziness when bending the head due to brainstem compression.

CM-1 malformation manifesting with ASD is a rare occurrence. We present a case report of a patient with CM-1 malformation presenting with ASD and we prefer a hypothesis to explain this unusual occurrence.

CASE DESCRIPTION

Patient D, W who is a 10-year-old African-American male was noted from early childhood to be having impaired communication, mild intellectual delay, limited range of interests, lack of interests in engaging in group activities with his peers, hyperactivity, poor attention span, impulsivity, aggressiveness and seizure disorder. Patient prenatal history is a single surviving twin that was vaginally delivered spontaneously at 35 weeks weighing 3.062 g. He had pyloromyotomy at age 5 for pyloric stenosis. Other medical issues include mild intermittent asthma. Patient was seen after an episode of seizure and mother described patient seizures as that he stared into space, fell backwards and his whole body started jerking. The seizure duration was approximately 5 minutes long and when he recovered he was confused and had decreased responsiveness for approximately 30 minutes. She also reported that the patient had urinary incontinence after his fall.

On physical examination vitals were within normal limits. His mental status is awake, alert and appropriate responses for age. Pediatric Glasgow Coma Scale noted spontaneous eye opening, spontaneous movements and following objects and interactions with physician. Examinations of cranial nerves (CNII-XII) were grossly normal with normal motor coordination and sensation (to light touch and pain) in all four extremities. Laboratory results were within normal limits except for mildly elevated potassium at 5.3 mEq/L, blood urine nitrogen to creatinine of 25. Electroencephalogram showed findings of normal awake and drowsy individual consistent with normal findings for patient’s age while MRI was revealed cerebellar tonsillar downward herniation of 7 mm.

An assessment of ASD, attention deficit hyperactive disorder (ADHD), seizure disorder and CM-1 was made. Neurosurgical evaluation suggested that patient be followed up conservatively, while neurological evaluation suggested anti seizure medications and patient received rectal diazepam 2.5 mg. Patient also received Language, speech therapy in addition to parent training, social skills training and behavioral interventions. Patient was also commenced on risperidone 0.5 mg HS to help curtail his aggression while extended release clonidine 0.1 mg daily was added as an adjunct to behavioral/psychologic interventions for his ADHD due to poor response to trial of stimulants.
DISCUSSION

ASD is still poorly understood due to the diverse and complex nature of its clinical manifestations. There is male predominance just as was seen in our patient and there is suggestion that it has a genetic etiology which impairs brain development, causing poor social and communication skills as was seen in our patient [3, 6]. Intellectual disability, seizures are common in ASD just as was seen in our patient however there are few cases in the literature documenting ASD, ADHD, seizure disorder and CM-1 occurring together.

Out of the myriad clinical manifestations associated with CM-1 malformation, the commonest ones include headache, which is typically occipital, neck pain, dizzy feeling that is exacerbated by physical activity or valsalva maneuver and seizures. The exact mechanism for the development of CM-1 is still subject of debate, however there is genetic postulation that there may be defects in brain growth, associated bony structures leading to posterior cranial fossa disproportion, overcrowding, compression of neural tissues and the descent of the cerebellar tonsils [2, 4].

The cephalocranial disproportion in neural tissue in CM-1 patient has been described by Osuagwu et al., 2006 [2]. This disproportion in neural tissue is also observed in patients with ASD as neuroimaging and histological evaluations has shown abnormalities in brain volumes and anatomy in ASD patients leading to impaired cognitive processing. Brain abnormalities documented include enlargements in cerebral hemispheres, cerebellum and caudate nucleus [7]. Altered cranial growth with enlargements in brain size during childhood with abnormal neural networks has also been described in patients with ASD suggesting that our patients cramped posterior cranial fossa might have been due a similar mechanism that was responsible for his ASD [5]. Structural and functional imaging studies have shown evidence of altered brain volumes in ADHD leading to dysfunction in fronto-subcortical pathways manifesting in restlessness and inattention as was seen in our patient [9].

Our patient was born at 35 weeks of gestation and there is suggestion that preterm birth might be a risk factor for ASD. Furthermore, our patient was diagnosed with asthma and association of asthma, beta 2 agonist’s exposure and ASD has been reported [10].

These may have lead to eventual neural dysregulation leading to complex neuropsychological and functional abnormalities causing social, communication impairment with restrictive interests and repetitive behavior.

CM-1 and ASD have associations with altered brain volume suggesting the possibility that the structural abnormalities due to the CM-1 might have an effect on patients ASD symptoms, however more work need to be done regarding this.

CONCLUSION

The presence of these constellations of symptoms makes its pertinent for more studies to elucidate genetic factors that might lead to cephalocranial disproportion with altered brain volumes that modifies brain development causing social, communication impairment with restrictive interests and repetitive behavior.

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CONFLICTS OF INTEREST DISCLOSURE

The authors do not have any conflict of Interest to Disclose.

REFERENCES:


Figure Sagittal view MRI showing cerebellar tonsillar herniation.