Case Report

Rare Presentation of Schmahmann's Syndrome in Dandy-Walkers Malformation - A Case Report

Abstract

The aim of this case report is to highlight the varied presentation of neurological disorders and the need for detailed evalution of the acute manisfestations of psychiatric symptoms. We report the case of a 17 year old boy presenting with complaints of acute onset of behavioural symptoms. We have briefly reviewed and discussed the clinical , diagnostic aspects of schmahmanns syndrome and therapeutic aspects of behavioural symptoms in dandy walkers malformation.

Keywords: Dandy Walker Malformation, Schmahmann's Syndrome, Disinhibition, Repetition, CCAS scale.

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Introduction:

Dandy Walker Malformation is a posterior fossa anomaly of the cranium, characterised by agenesis or hypoplasia of vermis; cystic enlargement of fourth ventricle with communication to a large cystic dilated posterior fossa; upward displacement of tentorium and torcula; and an enlarged posterior fossa. [1] First described by Dandy and Blackfan (1914), supplemented by Taggart and Walker (1942), it was introduced as its current description by Bender (1954).[2] Dandy Walker Complex (DWC) is a group of neurodevelopmental anomalies believed to occur between week 7-10 of gestation.[3] and comprise of Dandy-Walker Malformation(DWM), Dandy-Walker variant(DWV), mega-cisterna magna and posterior fossa arachnoid cyst.[4]

Clinical presentation of patients depends upon multiple factors, including severity of hydrocephalus, intracranial hypertension and underlying comorbidities. Symptoms including but not limited to developmental delays, macrocephaly, cognitive impairment, ataxia, hypotonia, occulomotor abnormalities, epilepsy and equilibrium disturbances may be seen in this condition. Association of psychosis and DMV, though rare, was reported by Sasaki et al. in pediatric patients.[5] Correlation between new onset of psychosis and cerebellar abnormalities in an adolescent patient have been hypothesised by Ryan et al. [6] Although the relationship between the two is still unclear due to lack of abundant data.

Another rare form of presentation of DWM is Schmahmann's syndrome which we will discuss in this case report.

Case Report:

Mr. ABC, 17 year old male, presented to the Psychiatry out-patient department of rural tertiary hospital with complaints of acute onset of repetitive and disinhibitory behaviour. He also had episodic outbursts of anger and irritability.

Clinical history elicited from his family members included repetitive movements of tying shoe-laces, latching and unlatching the door. They also reported that he was undressing publicly, in front of the parents and other family members at times, which was not his usual behaviour. On further enquiry, there was history of delayed developmental milestones, memory disturbances and poor scholastic performance.

The patient was not a known case of any psychiatric disorders or medical diseases. No past history of similar episodes was noted. Family history yielded insignificant in this case.

On examimation mental status examination was within normal range and no focal significant neurological deficits were elicited. And it occurred in clear sensorium. Patient was conscious, co-operative and oriented in time, place and person.

On investigation, there were no signs of raised intracranial pressure now or evidences of macrocephaly in infancy. Due to its acute nature and episodic presentation, patient was evaluated further and neuroimaging studies were done, during which the following positive findings were seen on the Magnetic Resonance Imaging (MRI) scan of the brain - Hypoplasia of inferior cerebellar vermis was noted with prominent IVth ventricle. It was thus confirmed as Dandy Walker Malformation.[1] Subsequently, based on clinical history, laboratory investigations and neuroimaging studies, a diagnosis of Cerebellar Cognitive Affective Syndrome in a case of Dandy Walker Malformation was made.

Administration of the Cerebellar Cognitive Affective Scale (CCAS) by Hoche [7] confirmed the diagnosis of Schahmann's syndrome.

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He was treated with Injection Haloperidol and Promethazine hydrochloride I.M. stat. Then started on Tablet Risperidone, Tablet Lorazepam and 4 hourly Injection Normal Saline with Multivitamin concentrate infusion(MVI). Regular follow up with visits in case of fresh complaints, similar such episodes or any emergencies was advised.

Discussion:

In the current case scenario, acute onset repetitive and disinhibitory behaviours were the core symptoms of the patient on presentation. The most notable pathway out of the many that govern repetitive behaviours is the cortico-basal ganglia-thalamic pathway, which is also involved in the motor activities.[8]

The patient also was reported to have delayed developmental milestones and had poor academic performance along with episodes of anger outbursts and There was no past history of similar complaints. No history of fever, trauma was found. The differential diagnoses of Autism Spectrum Disorder (ASD), Obsessive Compulsive Disorder (OCD), Cerebellar Cognitive Affective Syndrome (CCAS) were made.

The patient had no prior complaints of deranged behaviour patterns, difficulty understanding social cues, impaired communication and lack of empathy which helped rule out ASD. The Y-BOCS questionnaire, along with clinical history helped rule out OCD as the patient did not have tics, obsessive thoughts and compulsions in the past.

On administration of the CCAS/ Schmahmann's scale, postive findings were noted for the same and diagnosis thus confirmed. Schmahmann's syndrome is characterized by four clusters of symptoms including: (a) impairment of executive functions such as planning, set-shifting, verbal fluency, abstract reasoning and working memory, (b) impaired visuo-spatial cognition, (c) personality changes with blunting of affect or abnormal behaviour, and (d) language deficits including agrammatism, wordfinding disturbances, disruption of language dynamics and dysprosodia.[9]

Neuroimaging done due to acute presentation of disinhibitory symptoms, involved an MRI scan of the brain which showed the findings of - Hypoplasia of inferior cerebellar vermis was noted with prominent IVth ventricle; with normal brain stem and cerebellum, normal cisterns, sulci and sellar/ parasellar structures. No evidence of hemorrhage or midline shift or mass lesions was seen. Intracranial vessels and dural venous sinuses displayed normal flow voids. Midbrain, pons, medulla, orbits, paranasal sinsues and calvarum appeared normal suggestive of Dandy-Walker Malformation.

Surprisingly, the only symptoms the patient had were of cerebellar and cognitive impairment. Classical findings of raised ICT were absent, and no reports of macrocephaly during infancy was made either.

Management included treating the patient with Injection Haloperidol 2.5 mg and Injection Promethazine hydrochloride 50 mg I.M. stat. He was then started on Tablet Risperidone 1 mg twice daily, Tablet Lorazepam 2 mg once daily. Patient was stabilised and monitored. There were improvement in the behavioural symptoms. The dose of resperidone was reduced to once daily and Tab Lorezepam was stopped on subsequent follow up after 20 days.

Conclusion:

Dandy Walker Malformation can also present with symptoms of Cerebellar cognitive affective syndrome. This case report highlights the importance of complete physical and neurological including imaging studies in a child or adolescent presenting with psychotic or behavioural symptoms. Awareness of psychiatric manifestation of congenital malformations will impact the diagnosis, treatment and prognosis of the individual.

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Patient informed consent was obtained.

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Sonali Gogate: (30%): Contributed with comments on manuscript organization and write up.

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