

Quadriplegic Cerebral Palsy with Infantile Spasm and Lissencephaly: An Unusual Case of Neurological Convergence

KRUPA BHANUSHALI¹, REVAT MESHARAM², AMAR TAKSANDE³, ADITYA JAIN⁴

ABSTRACT

Cerebral palsy is a clinically defined symptom complex applied to individuals with a static, non progressive motor impairment of early onset, cerebral in origin. It is an aetiologically heterogeneous condition with multiple causes that feature an aberration or injury to the maturing central nervous system. Lissencephaly is a rare congenital condition that causes the developing brain to appear smooth, rather than the normal sulci and gyri. Here is a case of a 7-month-old female child with features like microcephaly, hypertonia and atypical movements, suggestive of quadriplegic cerebral palsy. The child also had multiple infantile spasms and was managed for the same. On Magnetic Resonance Imaging (MRI) studies, features of lissencephaly with Dandy-Walker malformation were noted. While a child with cerebral palsy can be diagnosed with anatomical malformations of the brain as well as seizures, the convergence of lissencephaly and infantile spasms in a child with diagnosis of cerebral palsy is a rare phenomenon. Hence, the need to do a comprehensive workup of a child with cerebral palsy is critical to not miss the associated conditions. The present case highlights the importance of a comprehensive workup for a child with cerebral palsy to address correctable issues and improve quality of life.

Keywords: Dandy-Walker malformation, Developmental delay, Neuronal migration disorders, Salaam spells, Spastic cerebral palsy

CASE REPORT

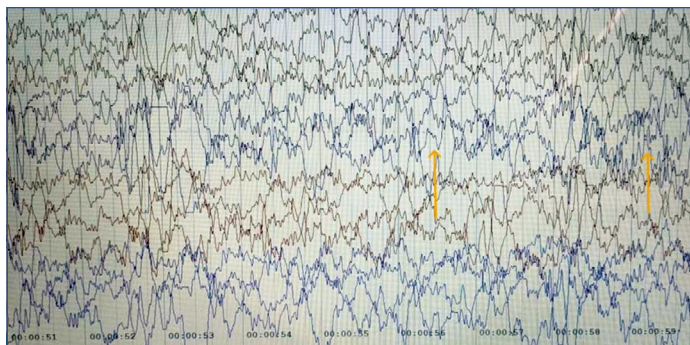
A 7-month-old female child was brought in the Department of Paediatrics with complaints of non attainment of developmental milestones and abnormal movements since birth. The child was born to a non consanguineous marriage via natural conception, first by birth order at 34 weeks of gestational age via normal vaginal delivery. The child had a weak cry after birth, was given oxygen support and was then admitted to the neonatal intensive care unit for 15 days. The child was started on Continuous Positive Airway Pressure (CPAP) support, continued for two days, oxygen, then tapered, and the child was then maintained at saturation on oxygen at room air with no signs of distress. Neurosonography was done on Day of Life (DOL) 8, suggestive of mildly dilated ventricles. The child did not have a history of ventilatory support, sepsis, convulsion or neonatal jaundice. The child was then discharged on DOL 15. Mother continued to breastfeed the child with no top feeds given. The child was taken to the same hospital for follow-up every month and was continued on vitamin D3 supplementation at 400 IU per day dose. Brainstem Evoked Response Audiometry (BERA) was performed at 2-months of age, which was suggestive of a 5th wave formation at 80 dB in both ears; no intervention was undertaken for this.

At the age of six months, the mother noticed that the child had excessive crying as compared to other infants and could not be consoled easily. The child also had a history of multiple episodes of sudden dropping of the neck with tightening and curling of all four limbs, lasting for around two minutes, occurring around 10-15 times in one day. These episodes would be preceded and followed by excessive crying. Upon eliciting a detailed history, the mother also reported that the child was unable to hold her neck or roll over. The child could not sit with support. The child also did not focus on colourful toys or light, and did not react to loud sounds around her. There was no history of fever, staring look, facial deviation, drooling of saliva, difficulty in breathing or fast breathing, or difficulty in feeding. The child was then brought to our centre for further evaluation.

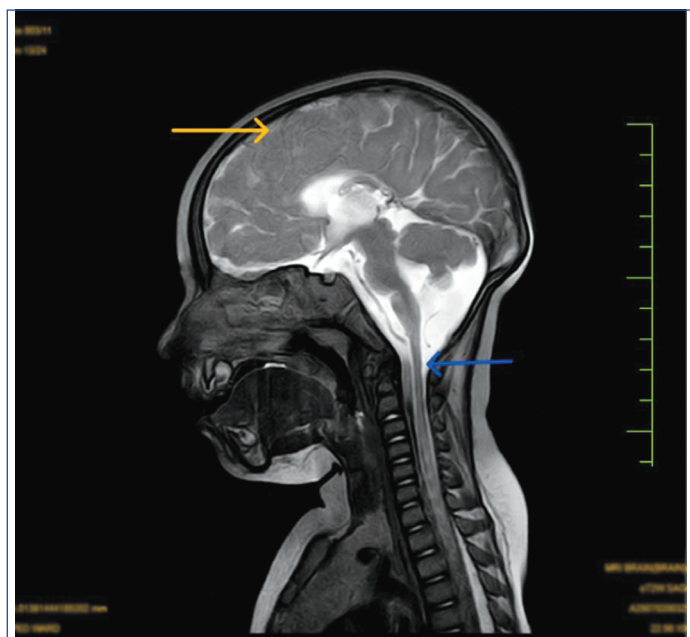
Upon admission, the child was vital and stable. Head circumference of the child was 34 cm, significantly less for the age suggestive of microcephaly with an anterior fontanelle open only to one finger's breadth. The child had grade 3, acute on severe malnutrition. The child is extremely irritable with a stiff posture. On ventral suspension, head lag was noticed. Scissoring of lower limbs noted. On motor examination, tone of the muscle increased in all four limbs with decreased power and plantar extensors. Moro's reflex and the asymmetric tonic neck reflex are absent. The child did not turn their head to sound or follow the light.

The child had multiple episodes of abnormal movements during hospital stay, characterised by sudden, jerky, downward drop of the neck with tightening of all four limbs, lasting for around two minutes, self-aborting, no altered sensorium, diagnosed to be infantile spasms.

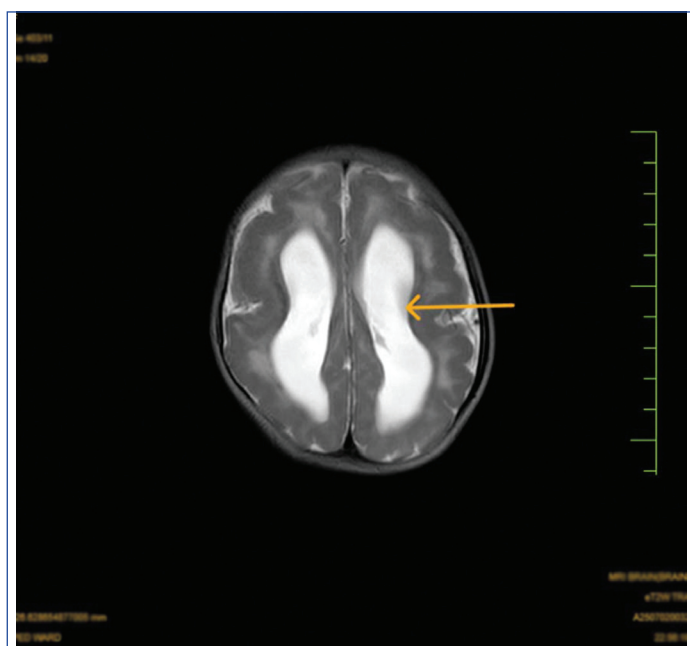
Upon further investigation, the thyroid profile was found to be normal. BERA done- suggestive of 5th wave formation at 80 dB in bilateral ears. Visual assessment done did not follow light, retina normal. Electroencephalogram (EEG) done with record showing intermittent sharp and high amplitude waves in centro-parieto-temporal and occipital region with background record of rhythmic synchronous symmetrical theta 4-7 Hz waves [Table/Fig-1]. MRI brain done, reported to have evidence of abnormal cortical development with few poorly formed gyri with smooth outer surface, pachygyria. Features suggestive of lissencephaly [Table/Fig-2]. Bilateral lateral and 3rd ventricles dilated with an Evans index of 0.49, ventriculomegaly [Table/Fig-3]. Narrowing was also noted at the level of the aqueduct, evidence of aqueduct stenosis. Bilateral cerebellar and inferior vermis hypoplasia with cobblestone appearance. Upward rotation of the vermis remnant with torcula seen lying above the lambdoid suture with an enlarged posterior fossa, features suggestive of Dandy-Walker malformation. The significant birth history and the examination findings are suggestive of cerebral palsy, other differentials like metabolic disorders which would show deranged metabolic profile, atypical facies or episodes of hypoglycaemia. Neuromuscular disorders would ideally present



[Table/Fig-1]: Electric encephalography showing intermittent sharp and high amplitude waves in centro-parieto-temporal and occipital region with background record of rhythmic synchronous symmetrical theta 4-7 Hz waves.



[Table/Fig-2]: MRI brain T2W section showing abnormal cortical development with few poorly formed gyri with smooth outer surface, pachygyria. Features suggestive of lissencephaly (yellow arrow) and aqueduct stenosis (blue arrow).



[Table/Fig-3]: MRI brain, T2W section showing bilateral lateral and 3rd ventricles dilated with Evans index of 0.49 suggestive of ventriculomegaly (arrow).

with lower motor neuron features and any spinal deformities like tethered cord would have cutaneous markers. Hence, other differentials were ruled out.

The child was started on topiramate at 2 mg/kg/day, clobazam 2.5 mg (0.5 mg/kg/day), Pyridoxine 50 mg/day and oral baclofen at a minimal

dose for spasticity. The child was also started on physiotherapy which included limb exercises to maintain the tone, help in increasing the range of movement and also occupational therapy was done. Physiotherapy was given three times daily for the duration of the hospital stay, totalling around 20 sessions.

Oral corticosteroid-prednisolone, 20 mg tablet, was started at 2 mg/kg/day with gradual tapering over one month for infantile spasm due to financial constraints for Adrenocorticotrophic Hormone (ACTH) injections. Child planned for hearing aid trial after one year of age, reassessment of vision and regular physiotherapy with occupational therapy. During course of stay in the hospital, significant improvement was noted in the infantile spasms, with reduction noted in the frequency of the spasms, however child was not brought back for follow-up hence long-term impact was not documented.

DISCUSSION

Cerebral palsy is a group of permanent disorders of movement and posture that cause activity limitation and are due to non progressive insults to the developing foetal or immature brain [1]. While the aetiology of cerebral palsy is generally attributed to perinatal asphyxia and other such causes, neuronal migration disorders like lissencephaly, more specifically pachygyria, can be a cause for cerebral palsy. It is a failure of the cerebral cortex to form convolutions because of defective neuroblast migration in early gestation, which leads to a smooth cerebral surface without sulci or gyri [2].

Several reports describe the association between neuronal migration disorders and epileptic spasms. Eng NY, Nie DA reported infantile epileptic spasms in a child with lissencephaly associated with a Platelet-Activating Factor Acetylhydrolase 1b Regulatory Subunit 1 (PAFAH1B1) mutation, highlighting the strong epileptogenic potential of cortical malformations [3]. Similarly, Tian G et al., described infantile spasms in a patient with lissencephaly due to KIF2A mutation and global developmental delay [4]. Posterior fossa malformations such as Dandy-Walker malformation have also been reported with seizures and developmental delay in isolated case reports. However, the coexistence of quadriplegic cerebral palsy phenotype with lissencephaly, infantile spasms, and Dandy-Walker malformation together remains rarely described in literature, making the present case noteworthy. A study conducted by Truwit CL et al., reported that nearly one-third of the cases of cerebral palsy in infants born at term had cortical dysgenesis secondary to disordered neuronal migration [5].

The cerebral cortex shows poorly organised cortical lamination, loss of radial orientation of pyramidal tracts and blurring of the white and grey matter junction in cases of pachygyria [6]. There is also evidence of disruption of neuronal polarisation. While children with lissencephaly are more susceptible to developing seizures, cases with infantile spasms occurring in children with lissencephaly have also been documented. KIF2A gene mutation has also been occasionally reported to cause infantile spasm and global developmental delay in children with lissencephaly [4]. Infantile spasms are the most common severe epilepsy in infants with sudden, brief, bilateral and symmetric contraction of muscles of the neck, trunk and extremities that occur in clusters with a peak age of onset between 3-7 months [7]. These brief spasms are missed frequently if not for keen observation. As noted in this case, the child had a history of brief jerky movements; however, the finding was ignored, presumed to be normal and only diagnosed later to be infantile spasm. While infantile spasms notably occur in the background of West syndrome, characterised by hypsarrhythmia and developmental delay, they can occur in other conditions [8].

A case of cerebral palsy requires a multidisciplinary approach and evaluation of multiple systems to improve the quality of life. MRI brain, although not a necessity, is often done to rule out any correctable underlying diseases. As noted in this case, MRI revealed

lissencephaly, more specifically pachygyria and thorough history and examination revealed the occurrence of infantile spasms in the child, confirmed by abnormal EEG. Incidental diagnosis of lissencephaly and infantile spasms in a case of cerebral palsy highlights the need for thorough evaluation of children suffering from cerebral palsy and the necessity to take corrective measures to improve the standard of living. Ophthalmic evaluation, including Visually Evoked Potential (VEP), fundus examination, squint correction, hearing evaluation with BERA, EEG for seizure evaluation, and sonography for reproductive organ assessment, are all conducted and necessary corrective steps are taken for the same. Genetic workup is recommended for those children with dysmorphic brain features or brain malformations [9]. Metabolic workup for inborn errors of metabolism is considered in cases with a progressive course [9].

For evaluation of infantile spasms, an EEG suggestive of hypsarrhythmia is classical. Hypsarrhythmia is characterised by high voltage, asynchronous slow waves with multifocal spikes that vary in duration and location [10]. The epilepsy panel can be considered in cases with a debatable diagnosis. Prenatal diagnosis of lissencephaly is possible with recent advances in foetal sonographies and foetal MRI, where findings of absent parieto-occipital fissure, calcarine fissure and abnormal appearance of sylvian fissure and insula are highly suggestive of lissencephaly. This requires advanced expertise and facilitates early obstetric interventions if demanded [11].

While limited interventions are available for children diagnosed with lissencephaly and cerebral palsy, the diagnosis of infantile spasms warrants immediate treatment. First-line treatment for infantile spasm is hormonal therapy with ACTH [8]. Two dosing regimens, high or low dose, have been cited; the low dose regimen consists of ACTH 20-30 units per day intramuscularly for two weeks, the high dose regimen consists of ACTH 75 units/m² intramuscularly twice daily for two weeks and tapering for two weeks [8]. Alternatively, corticosteroids can be considered at 2 mg/kg/day for a 6-week course with gradual tapering [12]. Evidence supporting the efficacy of levetiracetam and topiramate at ameliorating spasms and halting hypsarrhythmia has also been noted [12].

Lissencephaly has no cure; however, management of seizures with antiepileptic medications, physical and occupational therapy is the mainstay of management.

Children with quadriplegic cerebral palsy have a poor quality of life with developmental delay, physical disabilities due to spasticity and contractures. The target of management of CP focuses

upon maximum functioning ability, which includes aggressive physiotherapy, occupational therapy, drugs like baclofen, dantrolene, benzodiazepines, and botulinum toxin. Surgical options that can be explored are baclofen pump, selective dorsal rhizotomy, tendon release, spinal fusions or deep brain stimulation [13].

CONCLUSION(S)

While in the current scenario, though evaluation of children with cerebral palsy is not performed, the present case brings forth the need to determine co-existing co-morbidities that are commonly encountered in the disease spectrum and can be correctable with either drugs or surgical intervention. It also sheds a light on recognition and management of seizures in children with cerebral palsy to improve their quality of life.

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