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Gait Impediment: A Rare Ca(U)Se

Dr. Thakur Keerthi[1], Dr. K Smeeta Merlin[2], Dr. Ravanangomagan[3*]

[1]Post Graduate, Department of Paediatrics, Sree Balaji Medical College and Hospital
[2]Post Graduate, Department of Paediatrics, Sree Balaji Medical College and Hospital
[3*]Associate Professor, Department of Paediatrics, Sree Balaji Medical College and Hospital

Corresponding Author: [3*]Ravanagomagan
Chennai-600044

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ABSTRACT:

12year old girl, second born of second degree consanguineous marriage, with normal birth history presented with gradual onset of difficulty in walking since two years, currently being unable to walk without support. No other domains of development were affected. She was on antiepilectics from 3 to 6 years of age. On examination, B/L Ptosis was present with hypertelorism, tower skull, strabismus, microcephaly and generalized muscle wasting. CNS examination revealed hypertonia of both upper and lower limbs with brisk deep tendon jerks, right sided ankle clonus and extensor plantars. MRI Brain revealed large posterior fossa cyst communicating with slit like opening with fourth ventricle and hypoplasia of left cerebellar hemisphere. MRI C-Spine demonstrated anterior displacement of tip of odontoid process and severe compression of spinal cord, suggestive of Dandy Walker Malformation with CV Junction anomaly.

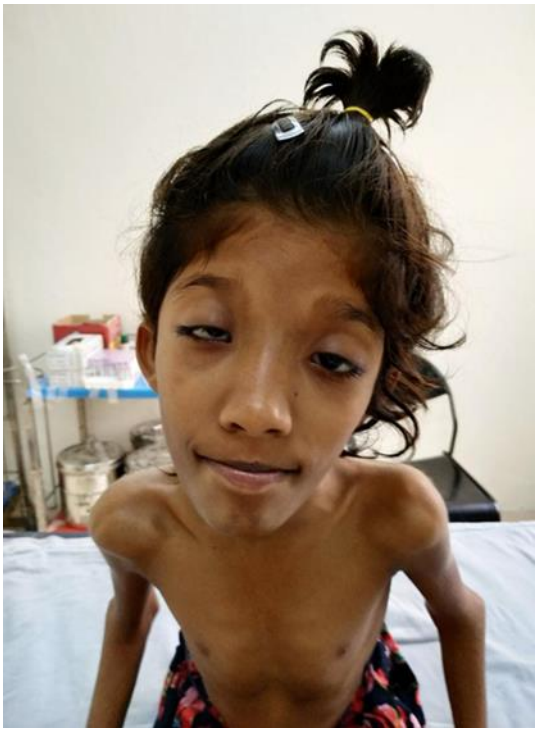
Keywords: Posterior Fossa Cyst, Left Cerebellar Hypoplasia, CV junction anomaly

1. Case Report:

12 year old girl, second born of second degree consanguineous marriage, presented with gradual onset of difficulty in walking since 10 years of age, currently being unable to walk without support.No other domains of development were affected. She was currently studying in Class 7 and there was no history poor scholastic performance.Significant past history included use of antiepilectics (Tab. Phenytoin) from 3 - 6 years of age, given for Seizure disorder. Last known seizure was at 4 years of age.She was delivered by normal vaginal delivery, cried immediately at birth and there was no history of NICU admission. Her mother had a history of one previous IUD.

Her general examination revealed presence of bilateralptosis, hypertelorism, tower skull strabismus,microcephaly, glossitis and pallor. It was observed that her mother had strabismus (Esotropia) too since childhood. The child's CNS examination demonstrated no abnormalities with regards to the higher functions. Her motor examination revealed hypertonia of the upper and lower limbs. Brisk deep tendon jerks and right sided ankle clonus were elicited along with upgoing plantar reflexbilaterally. The child was currently unable to walk without support. Her sensations were intact.

Primary work-up included complete blood profile and urine routine, which revealed no abnormalities. Her CPK levels were in the normal range. Opthalmologist consultation was sought in view of ptosis and strabismus and the patient was advised spectacle usage.



Bilateral Ptosis and Strabismus

Examination of mother revealed Strabismus (Esotropia).

Neurologist was consulted and further evaluation in terms of MRI Brain and MRI Cervical Spine were done. MRI Brain revealed large posterior fossa cyst communicating with slit like opening with fourth ventricle and hypoplasia of left cerebellar hemisphere.MRI C-Spine demonstrated anterior displacement of tip of odontoid process and severe compression of spinal cord with compressive myelopathy changes within the cord, suggestive of Dandy Walker Complex with CV Junction anomaly.
Neurosurgeon was consulted in view of craniovertebral junction anomaly, causing compressive myelopathy. The patient was advised decompression and stabilization surgery.However, patient's parents were not willing for surgery when the risk of quadriplegia was explained to them. The patient was advised physiotherapy and occupational therapy.She still continues to go to school and is performing well academically. She was advised to come for regular follow up and advised to use a cervical collar.

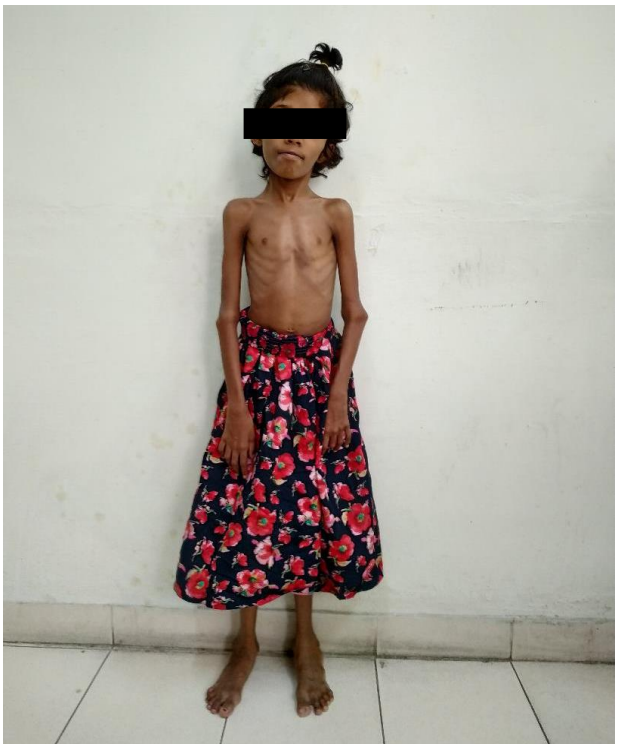
2. Discussion:

Dandy-Walker complex is a congenital group of disorders that have overlapping symptoms including Dandy-Walker malformation having a small cerebellar vermis, large fourth ventricle, and enlarged posterior fossa Isolated cerebellar vermis hypoplasia, Mega-cisterna magna, Posterior fossa arachnoid cyst. It is more common in females than males. The incidence is 1 in every 30,000 births. The longterm outcome and prognosis depend on the underlying malformation and symptoms it causes.Most of the cases of Dandy Walker are associated with hydrocephalus. However, this case was not associated with hydrocephalus. Mental retardation occurs in fewer than half, most often in those with severe hydrocephalus, chromosome abnormalities or other birth defects. Seizures occur in 15-30% of those affected. DWM results from defects in early embryonic development of the cerebellum and surrounding structures. A few patients have chromosome abnormalities including deletion of chromosome 3q24.3 (the location of the first DWM genes, known as ZIC1 and ZIC4), 6p25 or 13q32.2-q33.2, or duplication of 9p. In the remainder, it is probably due to other more complex genetic and perhaps environmental factors (teratogens) as the recurrence risk in siblings less than 5%. DWM may also occur as part of a genetic syndrome that includes

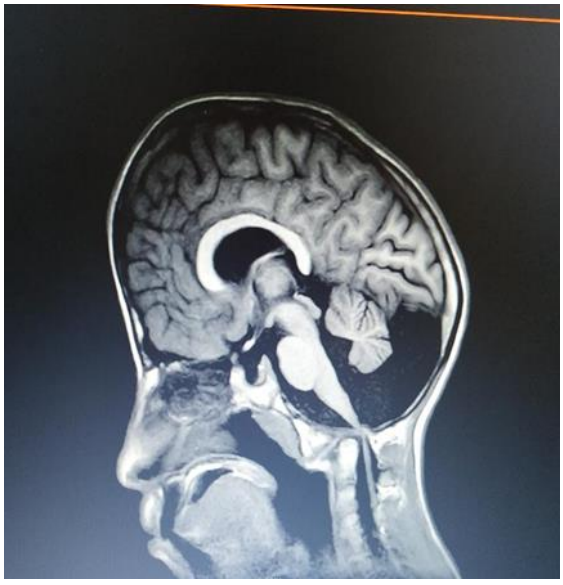
multiple birth defects, such as the PHACES syndrome of facial hemangioma, heart and sternal defects and DWM. Many other syndromes and chromosome abnormalities have been reported with DWM, but most of these appear to have CVH rather than typical DWM.

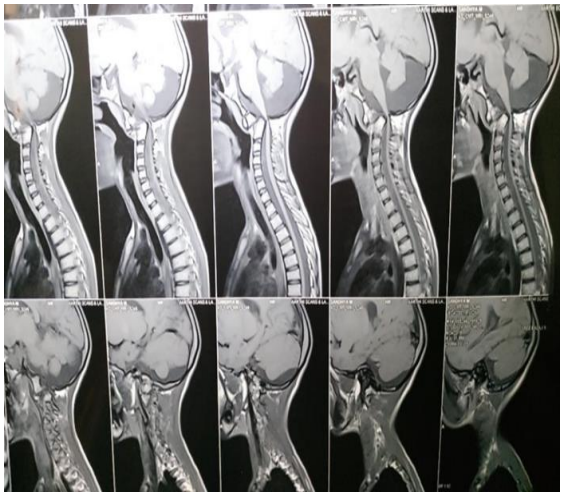
3. Conclusion:

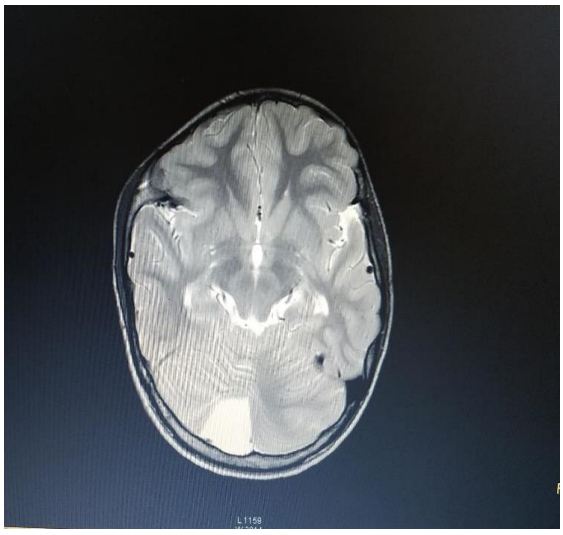
A supportive team approach for children with Dandy-Waller malformation is often warranted and may include special education, physical therapy and other medical, social or vocational services.Genetic counselling is recommended for families that have a child with Dandy Walker malformation.

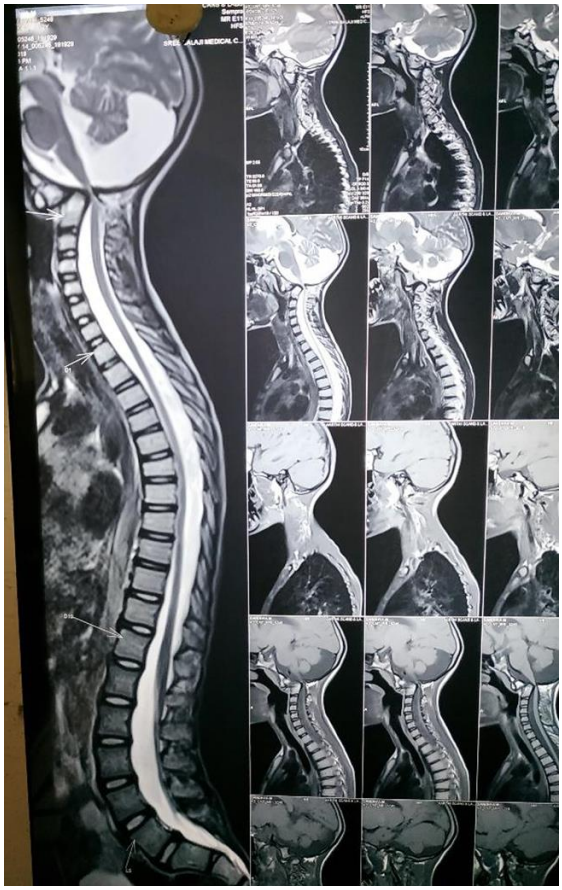


Patient standing supported against the wall









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