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Case Report

Congenital Bilateral Perisylvian Syndrome: A Rare Cause of Refractory Seizures in Infancy

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ABSTRACT

Congenital bilateral perisylvian syndrome (CBPS) is an extremely rare, congenital neuronal migration disorder characterized by refractory seizures, pseudobulbar palsy, delayed milestones, cognitive deficits and bilateral perisylvian polymicrogyria on brain imaging. We report a case of one and half year old girl with intractable seizures, spastic quadriparesis and feeding problems since birth with magnetic resonance imaging findings of CBPS.
This case report emphasizes the importance of detailed investigation in patients with epilepsy, especially patients with refractory seizures for the possibility of identifying a specific syndrome.

Keywords: Intractable seizures, Global developmental delay, Polymicrogyria.

INTRODUCTION

Congenital bilateral perisylvian syndrome (CBPS) is a migration disorder of the brain associated with distinctive clinical and imaging features. [1] The clinical spectrum may vary from mild speech difficulties to severe disability, intractable seizures, cognitive and behavioural problems. Seizures are difficult to treat in the majority and are resistant to antiepileptic medications. CBPS is more common than previously thought, is diagnosed by MRI brain and should be suspected clinically in any infant or child presenting with oromotor dysfunction, pseudobulbar signs, developmental delay and intractable seizures. MRIof the brain plays an important role in the diagnosis of this syndrome.

CASE REPORT

A one and a half year old girl presented in emergency with history of fever, cough and fast breathing for 5 days. She was referred from a peripheral hospital where she had received 5 days of Ceftriaxone and Amikacin therapy. She was a product of non-consanguineous marriage, born to 30 years Para two, unbooked mother with uneventful antenatal period. Baby was born by full term vaginal delivery at home with history of delayed cry at birth. There was history of drooling of saliva, feeding difficulties since birth, delayed milestones and episodes of intermittent stiffening of all four limbs since six months of age along with seizures in the form of twitching of facial muscles, up rolling of eyeballs and frothing from mouth. These episodes had a frequency of 3-4 per month but no medical advice was sought by the parents due to ignorance. There was no family history of seizures, epilepsy or progressive neurological disorders.

At admission to our hospital the child was critically sick with impending respiratory failure, generalised tonic clonic seizure and shock. On examination breath sounds were inaudible over right side of chest. There were no meningeal signs, dysmorphism or congenital anomalies on examination. The possibility of right pneumothorax with impending respiratory failure was kept. After stabilising the patient urgent portable x-ray was done which confirmed our diagnosis of pneumothorax. This was followed by mechanical ventilation and right intercostal tube insertion. Keeping the possibility of Staph Pneumonia injection Meropenem and vancomycin were started along with iv fluids, orogastric feeds, antiepileptics and ionotropic support. Serial X-rays weredone which showed improvement. Routine laboratory blood and urine tests were normal. After 5days patient was gradually weaned off ventilatory support and on 7th day post hospitalisation right intercostal tube was also removed.

After extubation a detailed neurological re-evaluation was done. Weight, length and head circumference was <3rd centile. There was global developmental delay, drop attacks, spastic quadriparesis, exaggerated deep tendon reflexes and extensor plantar response. She also had restricted tongue movements, drooling of saliva, feeding and swallowing problems along with lack of speech and language development. At 14th day of hospitalisation patient again had generalise tonic clonic seizures which was managed with injection Levetiracetam.

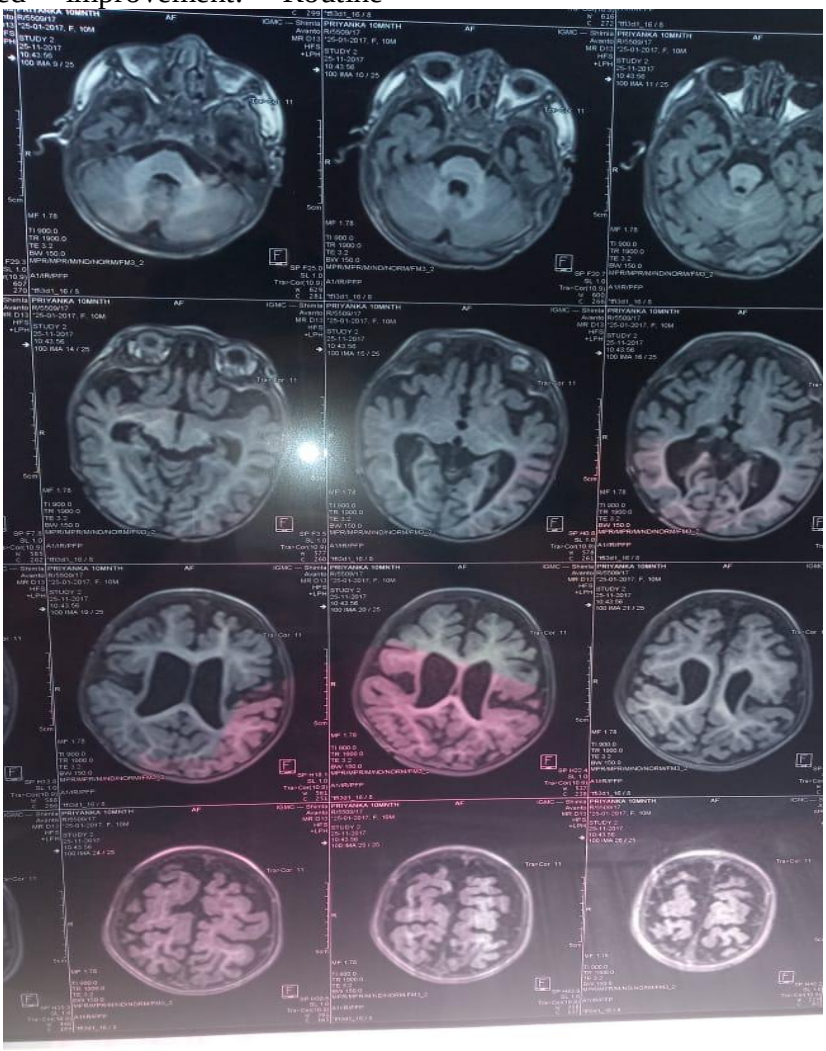

Figure 1.Magnetic resonance imaging (MRI) brain Axial image, showing bilateral perisylvian polymicrogyriawith irregular cortex and widening of the fissures.

Her chromosomal analysis, electromyography (EMG), nerve conduction study (NCS) and muscle biopsy were normal indicating a central cause. She had generalised epileptic form discharge on EEG. MRI Brain was done which was suggestive of cortical thickening along the gyri in bilateral sylvian fissure region and deep symmetrical cleft in bilateral sylvian fissure region extending till Pre – Rolandic region and lateral ventricles showed inverted appearance(Figure 1.). These findings were consistent with the diagnosis of CBPS. Therefore, on the basis of the typical clinical and neuroimaging findings diagnosis of CBPS was made.

Now the patient is on regular follow up with us (Figure 2.)She is two years old now and has shown some improvement in her oro-motor skills. She continues to have paucity of voluntary facial movements, drooling of saliva and marked global developmental delay. Her head circumference continues to grow at the 50th centile. She has been receiving physiotherapy and antiepileptic medications.

Figure 2. Patient in follow up phase with spastic quadriparesis and scissoring of lower limbs.

DISCUSSION

Perisylvian Syndrome refers to a neurological disorder in which the perisylvian region develops abnormally. The underlying developmental abnormality is polymicrogyria which is excessive number of small convolutions (gyri) on the surface of the brain; it can be generalized or focal. [2] The postulated mechanisms for this syndrome include cerebral hypoxia/ischemia, injury during neuronal migration and gene mutation, however the exact cause of this syndrome or the timing of the development of the malformation remains unknown. [1-4] Different modes of inheritance including X-Linked, autosomal dominant and autosomal recessive from different families have been reported and the mode of transmission remains unknown. [5-7] Relatively few pediatric cases of congenital bilateral perisylvian syndrome have been reported.

Multiple syndromes of region specific bilateral symmetrical polymicrogyria have been reported. [7] Bilateral Frontal Polymicrogyria typically results in developmental delay, mild spastic quadriparesis, variably impaired language development and epilepsy. Essential criteria(present in 100% of the cases) for diagnosis of this syndrome are oropharyngoglossal dysfunction, moderate to severe dysarthria and bilateral perisylvian malformations on imaging. [4] Additional criteria (present in more than 85% of the cases) include delayed milestones, epilepsy with onset usually between the age of 4 and 12,mental retardation and abnormal EEG. [1,5] Other criteria (< 50% of the cases) for diagnosis are arthrogryposis multiplex, infantile spasms, polydactyly, constriction band syndrome and pituitary hypoplasia. [8,9] Kuzniecky et al. reported that the mostfrequent seizure types included atypical absence, atonic/tonic seizures or drop attacks progressing to Lennox-Gastaut syndrome, tonic clonic seizures and partial seizures (26%). Seizures are poorly controlled in about 60% of the patients. Similarly in our case the patient had repeated episodes of pooling of saliva, drooling and difficulty in swallowing along with spastic quadriparesis and drop attacks. She had epileptiform discharge on ictal EEG and refractory seizures.

CBPS is identifiable on MRI as thickened cortex, poorly developed sulci and an irregular margin at the cortical white matter junction. The sylvian fissures are wide and underdeveloped. The bodies of the lateral ventricles show inverted appearance, typical of this disorder as was in our case.

Patients with refractory seizures should undergo detailed investigation, including brain imaging for the possibility of having a specific syndrome. Brain MRI plays an important role in the diagnosis of this syndrome and treatment mainly involves control of seizures with anti-epileptic drugs for long term. Antenatal diagnosis using ultrasound can be difficult as the regions of the brain that are involved in this malformation may not have reached their final folding until birth. Prognosis for epilepsy cannot be predicted based on the early response to treatment. [10] Corpus callosotomy has been reported to be useful treatment in patients with disabling seizures, especially with intractable drop attacks. [1,2,5]

CONCLUSION

Congenital bilateral perisylvian syndrome should be suspected in a patient presenting with refractory epilepsy, feeding difficulties, mental retardation and cognitive impairment. Since this syndrome carries a poor prognosis, early MRI brain is the gold standard for its diagnosis.

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