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CASE SERIES

Congenital Perisylvian Syndrome: Magnetic Resonance Imaging Findings in Six Cases

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Abstract

Background: Congenital perisylvian syndrome is an extremely rare congenital neurological disorder associated with distinctive imaging findings. Herein we describe the clinical features and imaging appearance of congenital perisylvian syndrome on Magnetic Resonance Imaging (MRI). **Material and Methods:** Six patients with clinical history, neurological examination and MRI findings suggestive of perisylvian syndrome were included in this review. **Results:** Six patients were found to have perisylvian syndrome on MRI findings. Five patients presented with pseudobulbar palsy while seizures were present in 3 patients. The most common MRI findings were vertically oriented sylvian fissures that were continuous with the central or post-central gyrus and perisylvian polymicrogyria.The findings were bilateral in 4 cases and unilateral in 2 cases. **Conclusion:** Perisylvian syndrome is a rare neurological disorder which can be diagnosed with certainty on MRI findings along with relevant clinical history.

Keywords

Perisylvian syndrome, Magnetic Resonance Imaging, Pseudobulbar palsy, Polymicrogyria, Seizures

Introduction

Perisylvian syndrome is an extremely rare developmental neurological disorder characterized by abnormal development involving a particular area of the brain (perisylvian region); the underlying abnormality being polymicrogyria.[1] The term polymicrogyria refers to abnormal appearance of the cerebral cortex with numerous abnormally small convolutions and reduced sulcation. Exact incidence and prevalence of polymicrogyria and perisylvian syndrome is still unknown due to heterogeneity of the disease. In a first of its kind population-based cohort study in Stockholm from 2004 to 2021, overall polymicrogyria prevalence was 2.3 per 10000 and perisylvian polymicrogyria accounted for 21% of these.[2] Perisylvian syndrome may be unilateral or bilateral and bilateral perisylvian syndrome is commoner than its unilateral counter part.Clinically, the patients primarily present with pseudobulbar palsy with dysarthria. Severe restriction of tongue movements is also common. Majority of these patients have seizures, as well as delayed motor and language milestones.[3,4]

Magnetic Resonance Imaging (MRI) of brain is the modality of choice in the diagnosis of perisylvian syndrome. Careful evaluation of distinctive MRI findings in collaboration with detailed clinical history and examination helps in clinching the diagnosis with a high degree of certainty.

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Material and Methods

Six patients with characteristic MRI findings of perisylvian syndrome formed the material of this review. Detailed clinical history including perinatal, developmental, family, drug and past history and findings of neurological examination were obtained in all the patients. In patients with history of seizures, careful attention was given to the type of seizures,their onset and frequency. An EEG (electroencephalogram) was also obtained in all thepatients. MRI of brain was performed with 1.5 Tesla MRI scanner (Siemens Magnetom Symphony). The imaging protocol included Axial T1Weighted (T1W), T2Weighted (T2W) and Fluid Attenuated Inversion Recovery (FLAIR) sequences, coronal T2W sequence, coronal and sagittal T1W sequences. Brain morphology was evaluated on MRI sequences and findings were recorded in detail.

Clinical Details

Age of the patients varied from 2 to 22 years with a mean age of 10.5 years. Out of 6 patients,4 were males with male to female ratio of 2:1. No Family history of consanguinity was elicited, however history of similar complaints in the sibling was noted in one patient.

One patient had history of delayed cry but rest of the patients had unremarkable perinatal and antenatal history. Physical examination showed varying degrees of pseudobulbar symptoms in 5 patients. The most common clinical feature was dysarthria noted in 5 patients, with severity varying from mild to severe and frequently associated with restricted/abnormal tongue movements in 4 patients. History of seizures were noted in 3 patients with one of the patient having first seizure episode at the age of 22 years. Two patients had complex partial seizures while one of them had atypical absent seizures. Hemiparesis affecting the opposite side of the body was present in one patient. One patient had associated bilateral club foot also. The clinical features are summarized in Table 1.

Imaging Findings

MR Imaging findings revealed vertically oriented and widened sylvian fissures continuous with central/ postcentral sulcus. The findings were bilateral (*Fig1*) in 4 cases and unilateral (*Fig 2*) in 2 cases. Associated findingsinclude thickening of the involved cortex with blurring of grey white matter interface along with shallow sulci and broad gyri (polymicrogyria) in perisylvian regions. In bilateral cases the findings were nearly symmetrical in 3 patients while asymmetric involvement was noted in one patient (*Fig 3*). In one of these patients polymicrogyria was also noted in the parietal and sub-frontal regions.

Table 1: Clinical Features in Perisylvian Syndrome (n=6)

Clinical features	No. of patients affected	% age of patients affected
Dysarthria	5	83.3%
Restricted tongue movements	4	66.7%
Delayed language milestones	4	66.7%
Difficulty in swallowing	4	66.7%
Cognitive deficit	4	66.7%
Seizures	3	50%
Delayed motor milestones	3	50%
Saliva drooling	1	16.7%
Hemiparesis	1	16.7%

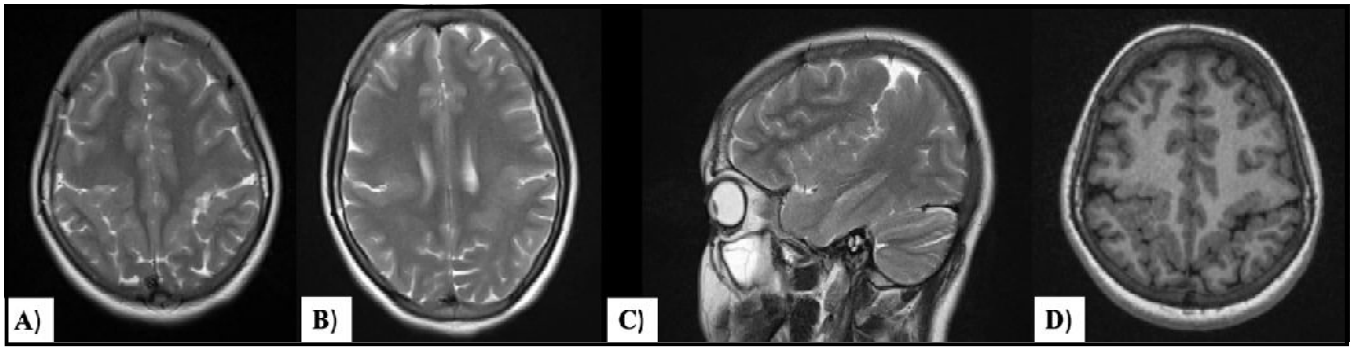

Figure 1. Bilateral Perisylvian Syndrome; Axial T2W (A,B), Sagittal T2W and Axial T1W MRI images reveal vertically oriented and widened sylvian fissures continuous with central/postcentral sulcus and presence of perisylvian Polymicrogyria.

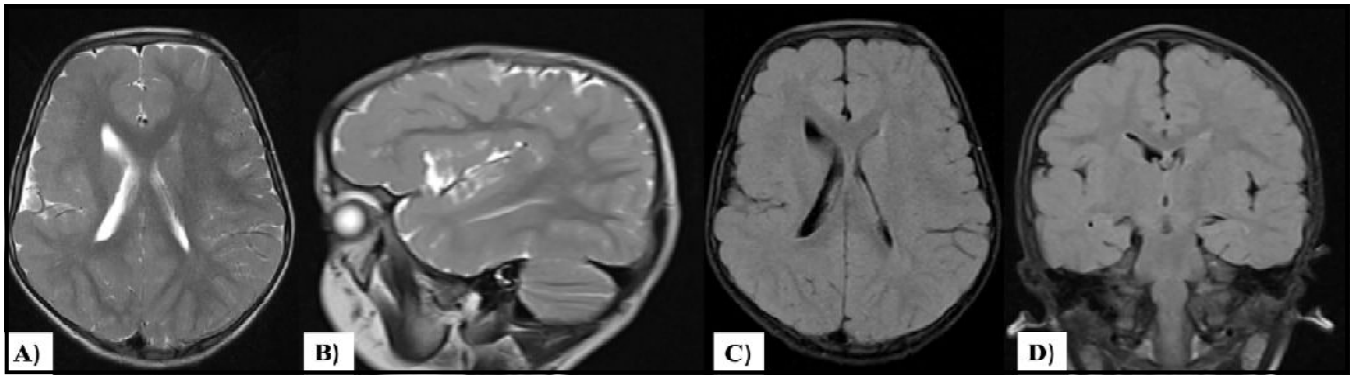

Fig 2. Unilateral Perisylvian Syndrome; Axial and Sagittal T2W (A, B) and Axial and Coronal FLAIR (C, D) images reveal right perisylvian polymicrogyria with widened sylvian fissure

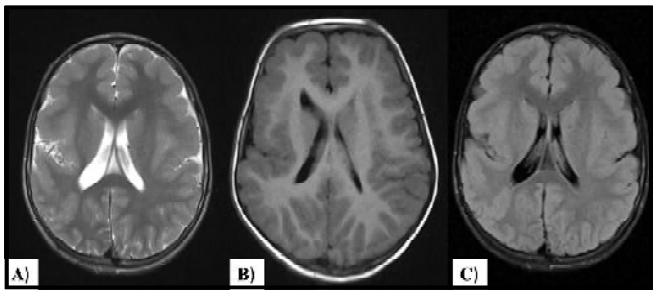

Fig 3. *Asymmetrical Perisylvian Syndrome; Axial T2W (A), Axial T1W (B) and Coronal FLAIR (C) images reveal bilateral asymmetrical perisylvian polymicrogyria with widened sylvian fissures.*

Discussion

Perisylvian syndrome is a congenital neurological disorder which is primarily diagnosed on the basis of neuroimaging and speech abnormalities.[3] The clinical manifestations of the syndrome include orofacial diplegia, markedly restricted/abnormal movements of the tongue, difficulty in swallowing, variable degrees of mental retardation and seizures.[5] Pseudobulbar palsy is the usual presenting complaint in bilateral perisylvian syndrome while unilateral cases commonly present with contralateral hemiparesis. Seizures and cognitive impairment may occur in unilateral as well as bilateral forms of the disease.[2,3] Different modes of inheritance have been reported including X-linked, autosomal dominant and autosomal recessiveinheritance.[1] The principal underlying abnormality in perisylvian syndrome is polymicrogyria which affects the perisylvian region of the brain.[1] Polymicrogyria, which is a malformation of cortical development, demonstrates multiple abnormally small convolutions with shallow sulci and broad gyriand thickening of the cortex. Polymicrogyria is manifested by a disturbance in the normal cerebral cortical development in the stage of neuronal migration in the later part or early in the stage of cortical organization and thus, it is presumed to be a disorder of neuronal organization.[6] On histology, a range of appearances is observed in polymicrogyria with derangement of the normal six-layered lamination of the cortex, associated derangement of sulcation, and fusion of the molecular layer across sulci.[7,8]

All six patients had relatively homogenous clinico-radiological findings comprising of pseudobulbar symptoms, delayed language and motor milestones, cognitive impairment, seizures and evidence of perisylvian cortical malformations on imaging. Dysarthria wasthe most prominent clinical symptom and the severity of dysarthria in these patients may vary from mild to severe. The characteristic clinical features are due to bilateral involvement of theoro-facial region of the motor cortex that results in the abolition of the compensatory mechanism contralateral to the lesion site.[5]

MRI is essential for complete delineation and detection of subtle changes in brain. Characteristic MRI findings include polymicrogyria with focal cortical thickening and loss of grey-white matter differentiation in the perisylvian region.[9] The cortical surface can have multiple small gyri or appear thick and irregularly bumpy or can be paradoxically smooth because the outer cortical (molecular) layer fuses over the microsulci. These alterations in the appearance of polymicrogyric cortexmay be easily missed on 5-mm thick routine images and so thin sections images with optimal grey-white matter differentiation should always be acquired.[4] In few cases, T2W hyperintensity may be noted in the affected region.[9] The opercula are dysplastic and incomplete and the sylvian fissures are wide and underdeveloped and

vertically oriented. Severity of polymicrogyria can also be graded on MRI (Grade 1 being the most severe and Grade 4 the mildest): Grade 1, perisylvian microgyria extending to the frontal or occipital lobe; Grade 2, polymicrogyria extending beyond the perisylvian region but not to any other lobe; Grade 3, polymicrogyria of the perisylvian region only, and Grade 4, polymicrogyria restricted to posterior perisylvian regions only.[4] The affected polymicrogyric cortex usually displayssimilar signal characteristics to normal grey matter. In occasional cases (<5%), the abnormal cortex may demonstrate foci of calcifications, often in patients with congenital infection.[9]

Association of Perisylvian syndromeincludessepto-optic dysplasia, which consists of absent septum pellucidum, hypoplastic pituitary stalk and hypoplastic optic chiasma/ nerves and globes.[10] Bilateral perisylvian syndrome may also be associated with chromosomal abnormalities and malformations such as arthrogryposis, clubfeet, micrognathia, polydactyly and constriction band syndrome.[11,12] Bilateral club foot was seen in one of our cases.

Treatment of perisylvian syndrome is mostly symptomatic and particularly aimed at controlling seizures (a common clinical manifestation). Rehabilitative measures like speech therapy, training in oral motor skills and swallowing as well as tongue muscle exercises may also help these patients.[9]

Conclusion

Perisylvian Syndrome has characteristic MRI findings and is the modality of choice in the diagnosis of perisylvian syndrome. MRI findings along with distinctive clinical findings can help in making ade finite diagnosis in majority of the cases.

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Conflicts of Interest

There are no conflicts of interest.

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