Parry Romberg’s syndrome is an uncommon disorder characterized by atrophy of skin and subcutaneous tissue of one side of face. It has neurologic sequel. The commonest of which is epilepsy. Here, we present a 17-year old girl with features of Parry Romberg’s disease with intractable epilepsy. Her seizures have stopped with systemic corticosteroids. This treatment response, together with previous reports is suggestive of an autoimmune basis to this disorder. Thus the epilepsy in some such cases may be steroid responsive.

**Key words:** Autoimmune, Parry Romberg’s syndrome, partial epilepsy, steroid responsiveness

**Introduction**

Progressive facial hemiatrophy, otherwise known as Parry-Romberg syndrome (PRS) is an uncommon disorder characterized by progressive atrophy of skin, soft tissue, muscle, and underlying bone of one side of face. Rarely, ipsilateral or contralateral limbs may be affected. The disease usually starts in the first two decade of life with skin changes resembling localized scleroderma. Various neurological complications including migraine, facial pain, seizure and radiological changes have been described in PRS. Of these, partial seizure appears to be the commonest. In a world wide survey about 11% of Parry Romberg’s disease patients had coexistent epilepsy.

The basis for this epilepsy has varied from structural lesions, to chronic encephalitis. We report here a girl with PRS, who developed intractable epilepsy. The management of this girl provides newer insights into the management and pathogenesis of this disorder.

**Case Report**

A 17 year old right handed girl presented with partial onset seizure involving left side since last 1 year. The seizures were mainly tonic with occasional clonic components and involved left face, hand and leg. She had a few episodes of generalized tonic clonic seizures. Born at full term out of a non-consanguineous marriage she had normal developmental milestones. At the age of three years the parents noted a linear discoloration of skin in the right supraorbital region. This was diagnosed as localized scleroderma and the girl was prescribed topical steroid. Soon afterwards, she started developing atrophy of right side of face. This gradually progressed to involve right temple, right cheek, and right jaw. The atrophy in linear scleroderma usually does not spread below the forehead. So at the age of 7 years the diagnosis was changed to PRS. No skin biopsy was done as histology cannot always reliably differentiate between the two conditions.

She had her first episode of generalized tonic-clonic seizure at the age of 16 years. EEG and CT scan of brain were normal. Carbamazepine was started but she continued to have episodes of left focal motor seizures involving face and upper limb. The seizure frequency increased progressively and was uncontrolled in spite of adequate dosage of carbamazepine, clobazam, sodium valproate, phenobarbitone, and lamotrigine. At the time of examination, she was having prolonged episodes of tonic and clonic seizures involving left arm, leg, and face, which recurred 6-7 times daily. Physical examination showed atrophy and hyper pigmentation of right side of face. There was no atrophy of limbs. Systemic examination was normal. Neurological examination showed normal intelligence, cognition and speech. There was no evidence of any developmental regression. Cranial nerves were normal. There was left hemiparesis (power 4/5). Deep tendon reflexes were brisk on left side and plantar response was extensor. She had impaired fine movements in left hand. However, she was able to walk without aid. Routine hemogram and biochemical tests were normal. Cerebrospinal fluid study was within normal limits. There were only 5 lymphocytes. No oligoclonal bands were

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found. EEG, done on two occasions was normal. MRI scan, performed using T1, T2, and FLAIR sequence revealed focal atrophy in right fronto-parietal cortex [Figure 1]. Considering the intractable nature of her focal seizures, progressive neurological deterioration and focal atrophy of right fronto-parietal lobe, the differentials of Parry Romberg’s disease with epilepsy and Rasmussen’s encephalitis were considered. Patient was advised to have evaluation for surgical resection. The parents denied consent. Autoimmune markers were negative.

The girl was started on corticosteroids (oral prednisolone at 1 mg/kg/day) after which her seizures stopped within one month. No other anti-epileptic drugs were modified during this period. After 3 months steroids were gradually tapered. Last 6 months, she has had only one episode of convulsion. That too occurred when we tried to stop the steroids completely. She is now on her eighth month of oral prednisolone. Presently she is on 10 mg/day Her anti-epileptics are also being tapered. We could not repeat MRI imaging as the girl has undergone a plastic surgery with a MRI incompatible facial prosthesis.

**Discussion**

PRS is a rare disorder of unknown etiology. The similarity between PRS and scleroderma has raised the speculation that PRS may be an autoimmune disease. The presence of autoantibody in serum of patients of PRS and occasional co-existence of this condition with other established autoimmune disorders like SLE and rheumatoid arthritis further supports the autoimmune hypothesis.\[8-10\] Resolution or improvement of the lesion after immunosuppressive treatment also supports the autoimmune mechanism. However, despite all these points, there are only a handful of cases of PRS where systemic steroids have been used for neurological management.\[11\] Ours is in fact one of the first reports to document stabilization of epilepsy after use of steroids in such cases. We had considered the option of using intravenous immunoglobulin. But keeping the prohibitive cost in mind, we kept it as a second choice. Moreover, as the patient responded dramatically to steroids we have not used it till date.

The mechanism by which epilepsy develops in PRS is not clear. Traditionally, the epilepsy has been ascribed to cortical dysgenesis and other such structural abnormalities. Two recent reports have highlighted the co-existence of PRS and Rasmussen syndrome.\[12,13\] The clinical description of Rasmussen syndrome consists of intractable seizures, progressive hemiparesis and focal atrophy of brain. Brain biopsy shows feature of chronic encephalitis. Evidence suggests that Rasmussen’s encephalitis (RE) is an autoimmune disorder. Chughani et al. recently reported a case of PRS in a boy of 7 years who developed intractable focal seizures. Based on clinical, radiological, PET scan, and brain biopsy findings they concluded that their patient had RE. Our patient had resistant epilepsy, progressive hemiparesis, neurocognitive decline and MRI evidence of focal brain atrophy. All these features closely resemble the clinical features of RE. But in addition she had facial hemiatrophy. Thus, the condition cannot be explained only by RE.

Despite the limitations, our case could be explained by two hypotheses. Firstly, there may be an association of two rare disorders, namely PRS and RE. At present, neither PRS nor RE has a known underlying cause. The possibility that they share a common autoimmune mechanism should be explored. The second possibility is that this girl had Parry Romberg’s disease with intractable epilepsy. But the condition responded to steroids. Either way, steroid responsiveness may suggest that chronic autoimmune encephalitis is responsible for the epilepsy in some such cases rather than dysgenesis.\[11,13,14\] The possibility of a coincidental remission cannot be ruled out completely. However, this seems unlikely considering the fact that the seizures recurred immediately on stoppage of steroids. West syndrome, Lennaux Gastaut syndrome, Landau Kleffner’s syndrome, and CSWS are also responsive to steroids though the exact basis of action is not known. The epilepsy of PRS may be an addition to this expanding list.

**References**

5. Wartenberg R. Progressive facial hemiatrophy. Arch Neurol Psy-
The year 2008 is the Birt
year of Dr. J. C. Patel. Some of his students/admirers felt that it would be a good idea to celebra
this Centenary Year by organizing CMEs, Orations/Lectures, Conferences, etc during the year. He was associated with many professional bodies, which meet regularly every year; during these annual meetings/conferences, a lecture/symposium, etc can be organized as a part of Centenary celebrations. We would like to form a Dr. J. C. Patel Birth Centenary Celebrations Committee. All his past students/admirers are invited to join the committee (without any financial commitment). Kindly communicate your name, designation, postal address, telephone number and E-mail ID to Dr. B.C. Mehta at Flat 504, Prachi Society, Juhu-Veerva Link Road, Andheri (W), Mumbai - 400053. E-mail: drmehta.bc@gmail.com.

Announcement

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