Progressive facial hemiatrophy (Parry Romberg syndrome) presenting with partial seizures and migraine headache a rare case report

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Abstract:
Abstract Progressive facial hemiatrophy is also known as Parry-Romberg syndrome. It is a rare, slowly progressive, neurocutaneous syndrome of unknown aetiology. It is characterised by unilateral facial atrophy of subcutaneous tissue, skin, muscles and bone. The common neurological complications associated with this disease are migraine and epilepsy. Method Case report A 45 years old female with history of thinning of right side of her face for the past 16 years, migraine type of head ache for the past 8 years and left focal seizure for the past 6 years duration. On examination she had atrophy of right side of her face, involving cheek, buccal and zygomatic areas with facial asymmetry. The skin over this area was not indurated and not adherent to underlying tissues. Her neurological, ophthalmological and dental evaluations were normal. Biochemical parameters were within normal limits. Anti nuclear antibody, Rheumatoid factor and Anti-double stranded DNA were negative. Imaging showed atrophy of soft tissues on right side of her face and bilateral globus pallidus calcification. Discussion and conclusion In our patient the facial hemiatrophy started at about 29 years of age, on the right side of the face, which progressed slowly over a period of 10 years and became static for the past 5-6 years. Most common type of seizure associated with Parry-Romberg syndrome is partial seizure (11 percent) as seen in our patient. Migraine may occur in up to 45 percent of these patients. Reported imaging findings were meningeal enhancement, intra cranial calcification and cerebral atrophy. There is no definitive treatment. Symptomatic treatment for symptoms like seizures and reconstructive surgery for facial disfigurement can be considered. While dealing with common neurological symptoms like head ache and seizures, we have to consider such uncommon disorder in appropriate settings. 
Keyword: Progressive facial hemiatrophy, Parry-Romberg syndrome, seizures, epilepsy, migraine, head ache
Introduction:
Progressive facial hemiatrophy is otherwise known as Parry-Romberg syndrome. It is a rare neurocutaneous syndrome of unknown aetiology, characterised by progressive, unilateral facial atrophy of subcutaneous tissue, skin, muscle and bone (1, 2).

First description of the progressive facial hemiatrophy was by Caleb Hillier Parry (1755-1822), a British physician (3). In 1846 Moritz Heinrich Romberg (1795-1873) described it as a syndrome (4). The word ‘Progressive facial hemiatrophy’ was coined by German neurologist Albert Eulenburg (1840-1917) in 1871 (5).

It is a unilateral disease, but 5-10 % cases may have bilateral involvement (6, 7). It is more common in females with a prevalence of 1/700,000 (2, 7, 8). It usually manifests within first two decades of life (9). But it can occur at any age and sex (8, 10, 11). If the disease onset is earlier, its intensity will be severe (12). It usually occurs sporadically, but there are few familial presentations as well (2, 13). The disease progression is rapid during the first 2-10 years following the onset after which it stabilises (2, 13, 14). Progressive facial hemiatrophy is associated with various neurological problems like epilepsy, migraine, trigeminal neuropathic pain, hemimasticatory spasm, mild hemiparesis, cognitive impairment, cranial neuropathy and brainstem signs (15, 16). It is also associated with various ophthalmological complications (17, 18, 19). Cerebral calcification, white matter lesions, meningeal enhancement, cerebral atrophy, infarctions in corpus callosum and cortical thickening were the reported neuroimaging findings (15, 20, 21).

Though this is a rare disease, it can present with common neurological symptoms and signs of right side of her face since the age of 29 years. She attributed repeated trauma as the cause. The wasting started over the right cheek and it slowly progressed over a period of 10 years. It remained static for the past 5 to 6 years. She developed headache over the past 8 years, which has features suggestive of migraine without aura (unilateral, lasted for more than 12 hours, 2 to 3 attacks in a week with photophobia and phonophobia). She had left focal seizure of 6 years duration which is hypermotor, complex partial seizure of extra temporal origin. There was no history of difficulty in chewing or swallowing. No history of eye lid drooping, weakness in opening the mouth, drooling of saliva or limb weakness. She was on oral sodium valproate, amitriptyline and propranolol. She is not a diabetic or hypertensive. There was no family history of seizures, migraine or facial hemiatrophy. She is the first born of non consanguineous parents with normal birth and development.

On examination, the patient had atrophy of skin and subcutaneous tissue predominantly over the buccal, masseter and zygomatic regions of lower half of right side face with facial asymmetry (Figures 1 and 2). Skin over this area was thin and pinchable. There were no induration or adherence of skin to underlying tissues. Prominence of right zygomatic arch was present. Patient also had melasma (hyper pigmented patch over bilateral cheek). She had no pallor or thyroid swelling. Her pulse rate was 78/minute, blood pressure was 110/70 mm Hg, respiratory rate was 15/minute. Her neurological examination including lobar functions, cranial nerves, spino motor system, sensory, cerebellar, gait, spine and cranium were normal.

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There was no atrophy or fasciculation of tongue. The limbs, neck and body were symmetrical. There was no autonomic disturbance on the involved side. There was no vitiligo. Ophthalmological, Dental and Cardiac evaluations were normal. Dermatological opinion was Parry – Romberg syndrome with melasma.

**Figure-3: CT Brain axial view showing bilateral globus pallidus calcification**

**Figure-4: MRI Brain axial gradient sequence showing bilateral globus pallidus calcification**

**Figure-5: CT paranasal sinuses coronal view showing right side soft tissue atrophy( white arrow)**

**Figure-6 CT paranasal sinuses axial view showing right side soft tissue atrophy( white arrow)**
Discussion:
Progressive facial hemiatrophy (Parry-Romberg syndrome) was described as a slowly progressive atrophy of soft tissues including skin, subcutaneous tissue and occasionally muscle and bony structures. It usually affects one side of the face but bilateral involvement has also been reported. Rarely, the atrophy extends to unilateral or contralateral upper limb. Hyper pigmentation of skin can occur (22). Even though the typical age of onset is first or second decade, it can occur at any age (9, 10, 11). Our patient had unilateral atrophy of face with melasma and the age of onset was 29 years (Fig 1 and 2). The lesion sometimes begins after trauma or surgery to the face, which was also noted by our patient (2, 15). Parry-Romberg syndrome usually progresses for 2-10 years following the onset and then it stabilises or goes into remission as in our case (2, 13, 14). It usually occurs sporadically, some familial cases have also been reported (2, 13). There was no family history of similar illness in our case. The disease is associated with various neurological problems like epilepsy, migraine, trigeminal neuropathic pain, hemi masticatory spasm, mild hemiparesis, cognitive impairment, cranial neuropathy and brainstem signs in various combinations (2, 15, 16). The common neurological complications are Migraine (45%) and partial seizures (11%) (2, 23). Seizures may even precede the onset of atrophy. The mechanism by which seizure develops in Progressive facial hemiatrophy is not clear but traditionally, it has been ascribed to cortical dysgenesis and other such structural abnormalities (1). Our patient had partial seizures and migraine. Progressive facial hemiatrophy can have ophthalmological and dental abnormalities. Ophthalmic complications associated with this syndrome are enophthalmos, decreased corneal sensitivity, ocular myopathy, episcleritis, ipsilateral Horner syndrome, blepharophimosis, cataract and secondary glaucoma. Enophthalmos is the most commonly noted association (2, 14, 17, 18, 19). Dental and ophthalmological evaluations were normal in our case. Cerebral calcification, white matter lesions, meningeal enhancement, cerebral atrophy, infarctions in corpus callosum, cortical thickening and intra cranial calcification are the reported neuroimaging findings (15, 20, 21). In our case, CT brain and paranasal sinuses showed bilateral globus pallidus calcification and atrophy of soft tissues on the right side of face. MRI brain also confirmed the same (Fig 3 to 7). Ultrasonography of abdomen may reveal the presence of atrophy of ipsilateral internal organs (14). However it was normal in our patient.
The exact aetiopathogenesis of Parry-Romberg syndrome is poorly understood. Various theories have been proposed. As per the trigeminal theory, vascular insult to the trigeminal ganglia and trauma to the superior cervical ganglia were the triggering factors of the wasting process (14). Some consider it as an autoimmune disorder, because of high frequency of auto antibodies than in general population, the overlap with linear scleroderma, vitiligo and the presence of transient high signal lesions on brain MRI. But these findings are not consistently present in all cases (2, 14). The other proposed mechanisms are cortical dysgenesis, dysfunction of the sympathetic nervous system, chronic localised meningoencephalitis, slow viral infections and disturbances of angiogenesis during growth and development (14, 15). There is case report of Progressive facial hemiatrophy and Rasmussen’s encephalitis (1). Shain et al, reported a case of Parry-Romberg syndrome, in whom the aetiology was thought to be Lyme disease, but the aetiological association was not established (22). Even though various mechanisms are proposed, they are inadequate to explain all the clinical features of the disease (15). Anti nuclear antibody, Rheumatoid factor and Anti-double stranded DNA were negative in our patient. The disease was often confused with en coup de sabre (French term which means ‘cut of the sword/like the stroke of a sword)(2, 13, 24). Localised scleroderma of the en coup de sabre may be associated with some degree of facial hemiatrophy, particularly if it begins early in life. But, en coup de sabre is a more superficial process than Progressive facial hemiatrophy. The skin will be adherent to underlying tissues. Hair loss and pigmentary changes are conspicuous. The skin is mobile and grossly normal in Progressive facial hemiatrophyBoth the disorders may coexist (9). Localised scleroderma of the en coup de sabre has a tendency to involve the upper face and scalp more often than Progressive facial hemiatrophy, which has a predilection for the lower face and cheeks (24, 25, 26). The diagnosis is based on history and clinical signs, since the skin biopsy cannot always reliably differentiate between these two conditions (1). In our case lower face and cheek on right side was affected, skin was mobile, non adherent and there was no induration of the skin. The clinical diagnosis of Progressive facial hemiatrophy was made. There is no definitive treatment for this disease. However symptomatic treatment for complications like seizures and migraine has to be given. Plastic surgery for cosmetic disfigurement can be considered, which includes autogenous fat grafts/lipofilling or silicone injections, flap/pedicle grafts, bone implants or cartilage grafts (2, 6). Most patients do not have severe disease to warrant immunosuppressive therapy. In case of severe and progressive disease methotrexate, corticosteroids, azathioprine and cyclophosphamide were used but there are no randomized controlled trials to evaluate the benefits of these drugs (2, 6, 8, 13). In our patient, the disease is inactive at present and her seizure and migraine are under control with sodium valproate. Since our patient was already on sodium valproate, the same drug was titrated. Other drug like amitriptyline and propranolol were tapered and stopped. She declined surgical options for facial disfigurement.

Progressive facial hemiatrophy is a rare neurocutaneous syndrome of unknown aetiology. It is characterised by progressive, unilateral facial atrophy of subcutaneous tissue, skin, muscle and bone. It may present with various neurological complications like seizures and migraine. We must be aware...
of this rare disorder and one must suspect this disorder in appropriate settings. We are presenting this disorder for its rarity.

**Note:** Informed consent was obtained from our patient for publication of photos and information, which is available with the author.


9) Facial hemiatrophy, Rook’s Textbook of dermatology, eight ed. Vol.3; 45.11-45.12.


11) The Romberg’s connection- 2009 survey results-an international support group made up of over550 individuals and families whose lives are affected by Parry-Romberg syndrome.


