OMCR

Oxford Medical Case Reports, 2016;8, 166–169

doi: 10.1093/omcr/omw031
Case Report

CASE REPORT

Lower limb onset Parry–Romberg syndrome: an unusual presentation of a rare disease

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Abstract

Parry–Romberg syndrome (PRS) is characterized by progressive degeneration and atrophy of the cutaneous, subcutaneous connective tissues, muscles and bones. Classically, PRS is restricted to unilateral face but in 20% of patients may extend to other parts of the body including ipsilateral or contralateral arms, trunk and legs. We report a case of 24-year-old male who presented with insidious onset, gradually progressive deformity and muscle wasting of right lower limb followed by right side of face and chest for 8 years. The right side of the face showed hemiatrophy, coup de sabre and deviation of nose and chin toward the same side. The magnetic resonance imaging showed atrophy of right lower limb. Computed tomography with 3D facial reconstruction revealed atrophy of facial bones on right side. He was managed with physiotherapy and symptomatic treatment and planned for facial and ankle reconstructive surgery on follow-up.

INTRODUCTION

Parry–Romberg syndrome (PRS), also known as progressive hemifacial atrophy, is a rare disorder of unknown etiopathology [1]. This disorder is characterized by progressive degeneration of skin, subcutaneous connective tissues, muscles and bones. Bone and cartilage tissues are rarely affected, unless the onset occurs before the second decade [2]. Mostly, it is sporadic and occurs predominantly in females with a female-to-male ratio of 3:2. It begins in the first decade of life, although late onset has also been described in the literature.

CASE REPORT

A 24-year-old male presented with insidious onset, gradually progressive thinning and weakness of right lower limb for 8 years. He started walking on toe and had difficulty in squatting position on right side. Later, he developed progressive muscle thinning and deformity of right half of face and chest for 3 years. He had no history of fever, headache, fasciculations, sensory symptoms, vision loss, hearing impairment or seizures. There was no history of trauma, chronic infections, drug abuse, hypertension or diabetes mellitus in the past. The family history was negative. On examination, right lower limb showed muscle atrophy, foot deformity and contracture (Fig. 1). The face was asymmetric due to atrophy of right half of the cheek, chin and lips (Fig. 2). A hyperpigmented area and coup de sabre (scar defect) are seen on the right side of the central frontal and chin (Fig. 2). The right mandible, maxillary, zygomatic and frontal regions were depressed. Left side of the face was normal. Intraoral soft tissue examination showed atrophy and fissuring of tongue on right side (Fig. 2). Right side of chest was depressed due to wasting of subcutaneous tissue and thoracic muscles (Fig. 3). Higher mental functions and fundus examination were normal. Muscle power was medical research council grade 4/5 proximally and distally on right side of lower limb and normal in other

Received: December 30, 2015. **Revised:** March 14, 2016. **Accepted:** March 25, 2016

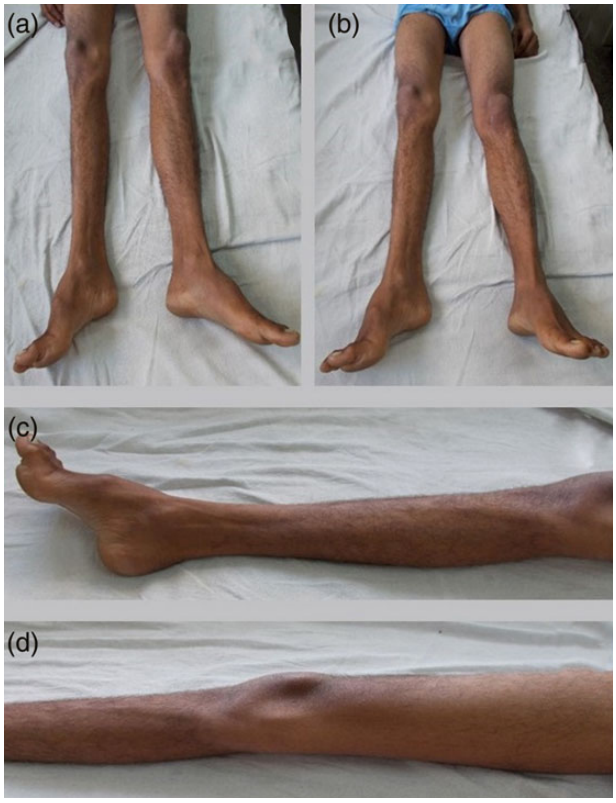

Figure 1: The figure of lower limbs is showing atrophy of subcutaneous tissue and underlying muscles on right side. There was a difference of 2.5 cm on right side of the thigh and leg when compared with left side of the thigh and leg (**a–d**). Right foot deformity and contracture are also present (c).

limbs. Rest of the neurological examinations including deep tendon reflexes, plantar, sensory and cerebellar system were unremarkable.

Hemogram, biochemistry including thyroid function tests and serum vitamin B12 level were normal. Abdominal ultrasonography showed no evidence of organomegaly or free fluid. Serology for human immunodeficiency virus (HIV) and viral hepatitis (HBsAg, HCV) was negative. Serum antinuclear antibodies, anti-dsDNA, anti-histone antibodies, rheumatoid factor and anti-centromere antibodies were negative. Two-dimensional echocardiography and electrocardiography were normal. Electrophysiology tests (nerve conduction studies and electromyography) of face and limbs were normal. Radiographically, there was a clear discordance in the right and left thighs and tibial bone. Magnetic resonance imaging (MRI) of lower limbs showed thinning of right leg and thigh. Computed tomography (CT) with 3D reconstruction of face revealed atrophy of the mandibular, maxillary, zygomatic and frontal bones on the right side. MRI of the brain and spine was normal. Multimodal therapies including physiotherapy and supportive treatment were given to the patient, and facial and ankle reconstructive surgery were planned in follow-up.

DISCUSSION

PRS is a rare acquired disorder, characterized by progressive degeneration and atrophy of the skin, subcutaneous connective tissues, muscles and bones. Classically, PRS is restricted to unilateral face but in 20% of patients may extend to other body parts including ipsilateral or contralateral arms, trunk and legs [3]. Our patient presented with progressive right lower limb atrophy, which has not been described in the literature thus far.

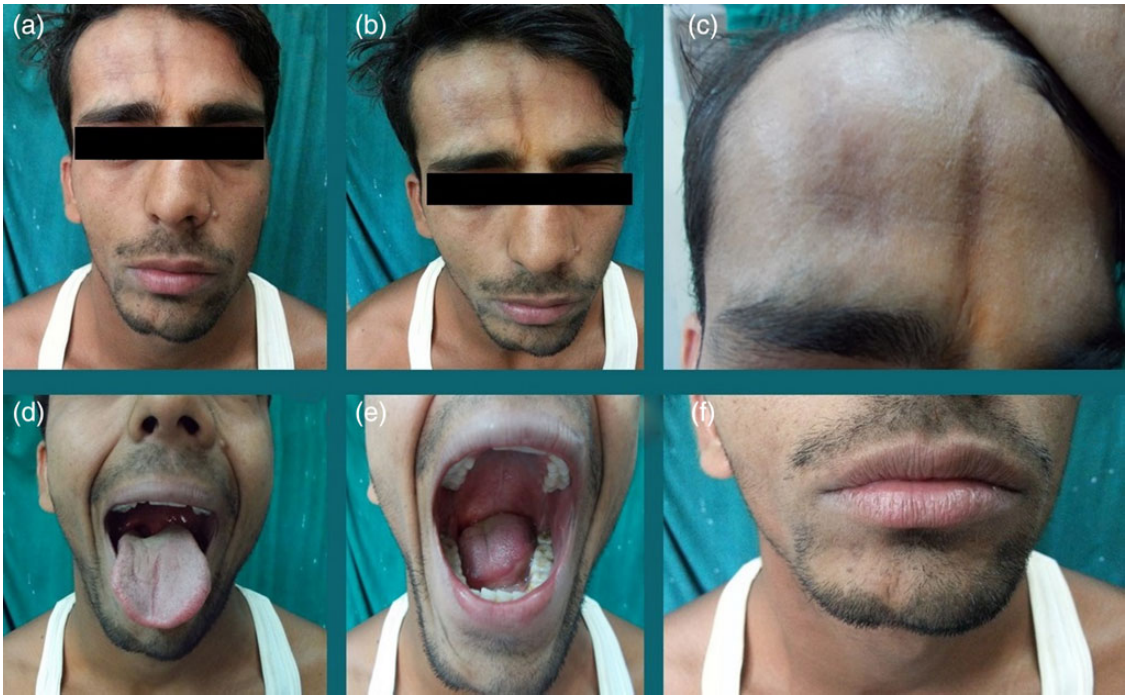

Figure 2: The facial features of the patient showing hemiatrophy of the skin, subcutaneous tissues and muscles on the right side (**a–f**). There is hyperpigmented patch and en coup de sabre (scar defect) appearance on right side of frontal (c) and mandibular (f) regions. There is atrophy of right half of the cheek, chin and lips (a and b). The right mandible, maxillary, zygomatic and frontal regions appear depressed when compared with left side. The nose and chin are deviated to the right side. There is atrophy and fissuring of the right side of the tongue (d and e). Left side of the face is normal.

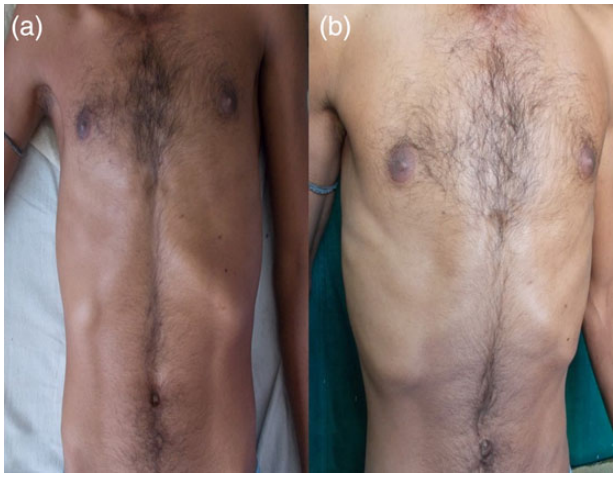

Figure 3: Right side of the chest is depressed due to wasting of subcutaneous tissue and thoracic muscles (**a** and **b**).

Skin and subcutaneous connective tissue changes may range from focal skin discoloration to severe muscle atrophy and functional disability. There are progressive atrophy and deformity of the face, nose, chin, ear and orbit, resulting in deviation of face toward the same side. Typically, disease starts with atrophy of buccinator, masseter and temporalis muscles. Involvement of frontal area of the scalp is usually associated with hair loss and depressed linear scar ('coup de sabre') [4]. In 5–10% of the PRS patients, disease may extend to bilateral face and intraoral structures like lips, gingiva, tooth and hemiatrophy of the tongue. Commonly associated neurological manifestations are migraine, trigeminal neuralgia, focal seizures, mental retardation, cerebral atrophy, intracranial vascular malformations and brain tumors. Enophthalmos is the most common ophthalmological abnormality due to loss of subcutaneous tissues and muscles around the orbit. Other common findings are extraocular muscle thinning, eyelid atrophy, ptosis, third nerve paresis, uveitis, glaucoma and band keratopathy on the affected side of the face.

The etiopathogenesis of this acquired disorder is unknown; however, various hypotheses have been proposed including autoimmune, trauma, sympathetic dysfunction and chronic infections [5]. There is strong evidence of autoimmune disorder due to high prevalence of disease in females, very close relationship with linear scleroderma en coup de sabre, high prevalence of autoantibodies in serum and frequent involvement of multiple systems. Some authors relate this disease to alterations in the sympathetic nervous system due to trauma (accidental, postoperative sympathectomy, dental avulsion) of cervical plexus or sympathetic trunk. Slow viruses (herpes virus, rubella) or chronic bacterial (*Borrelia burgdorferi*, diphtheria, syphilis, tuberculosis) infections have also been hypothesized as a possible causative factor in PRS, although no organism had been identified [6].

The closest differential diagnosis such as wasted leg syndrome, muscular dystrophies, spinal muscular atrophy, linear scleroderma and Rasmussen's syndrome were ruled out on detailed clinical history, examination and laboratory investigations [7]. Differentiating PRS from linear scleroderma en coup de sabre (LSCS) is very challenging. The most differentiating features of LSCS are site, severe atrophy and the presence of inflammation or induration. Rasmussen's encephalitis is a chronic progressive inflammatory disorder, which usually affect one side of cerebral hemisphere [8]. The neurologic symptoms include focal seizures, progressive hemiparesis and cognitive decline. The MRI of the brain shows ipsilateral brain atrophy, patchy gyriform cortical enhancement and areas of unilateral hyperintensity corresponding to cytotoxic edema.

Corticosteroids and immunosuppressive drugs including azathioprine, hydroxychloroquine, methotrexate, cyclosporine and cyclophosphamide may be considered in active phase of disease, neurologic manifestations or associated comorbid autoimmune disorders. Usually, spontaneous stabilization of disease occurs after 8–20 years. Once the disease stabilizes, esthetic therapy consisting of augmentation of the atrophic region and restoration of the symmetry of affected body part can be offered [9].

CONFLICTS OF INTEREST STATEMENT

None declared.

FUNDING

None.

AUTHORS' CONTRIBUTIONS

All the authors contributed to prepare this manuscript.

ETHICAL APPROVAL

We have followed the ethical norms and have taken proper informed consent from the patient and relatives. Our patient participated voluntarily and did not suffer any harm. We confirm that all the research meets the ethical guidelines, including adherence to the legal requirements of the study country.

CONSENT

We have obtained written informed patient consent for publication of the report and any accompanying images.

GUARANTOR

S.K. is the guarantor of this article.

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