

Idiopathic acquired progressive left facial hemiatrophy (Parry-Romberg syndrome) in a 21-year-old man in semi-urban, south-west Nigeria

Imarhiagbe FA,

Department of Medicine, Federal Medical Centre, Owo, Ondo State, Nigeria

Adeoti AO

Department of Medicine, University of Benin Teaching Hospital, Benin City, Nigeria

Correspondence to: Dr Frank Imarhiagbe, e-mail: drnime@yahoo.com

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Abstract

Idiopathic progressive hemifacial atrophy, or Parry-Romberg syndrome, is a rare entity, seldom described in medical texts. Though first described in 1825, as yet there are no clear-cut diagnostic criteria. It is of interest mainly because of the numerous features with which it may be associated, which are largely in the central nervous system, and, more importantly, because it can easily be confused with localised scleroderma *en coup de sabre*.

Some classify it as one of the trophoneuroses; others believe it is a variant of localised scleroderma. Most of the sufferers present because of the attendant cosmetic loss from the facial disfigurement. Treatment is multifactorial, and includes facial reconstructive surgery and immunosuppressants. Here we present the case of a 21-year-old Nigerian with idiopathic progressive left hemifacial atrophy. To the best of our knowledge, it is probably the first case from Nigeria to be reported in literature.

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Introduction

Idiopathic progressive facial hemiatrophy, or the Parry-Romberg syndrome, is a very rare entity of unknown aetiology. It is seldom described, even in standard medical texts. It was first reported in 1825. In its simplest form, it involves atrophy of the facial subcutaneous fat and muscles. In the more extensive and severe forms, it presents with atrophic changes in the skin, muscles and bone of one side of the face, and it may be associated with epilepsy, encephalitis or trigeminal neuropathy. It is closely related to other causes of acquired localised atrophy of the face, like linear scleroderma *en coup de sabre*.

Though classified by some as one of the trophoneuroses (a group of diseases with localised dysfunction of the autonomic nervous system and trophic changes), others believe that it may be a variant of localised scleroderma or lupus profundus. The exact nature of this condition is currently not fully known. However, it is amenable to reconstructive cosmetic surgery in its simplest form, for the facial disfigurement it causes.

Here a case is reported in a 21-year-old man with progressive wasting of the left facial muscles without skin or bone involvement, and no other associated conditions or family

history. To the best of our knowledge, it is probably the first case presented in Nigeria to be reported in literature.

Case study

The patient was a 21-year-old secondary school leaver, who presented with a progressive history of wasting of the left side of the face over a one-year period prior to presentation at the Consultants' Outpatient Clinic of the Federal Medical Centre, Owo, Nigeria. He claimed to have been in good health, with no associated changes in skin colour or texture of the face, and denied any pain, photosensitivity, or alteration in sweating pattern.

There was no known family history of a similar condition, or any previous medications or epilepsy. No abnormality of mastication was reported. He claimed to have presented in hospital because of the facial disfigurement and the attendant cosmetic effect.

Examination revealed a young man who was not ill-looking, except for the obvious wasting of the left facial muscles (See Figures 1 and 2.) There were no other stigmata of chronic illness, the skin was normal, and the cardiovascular and respiratory systems and abdomen were unremarkable. The nervous system was essentially normal, and remarkably, higher mental functions, cranial nerves and sensation were

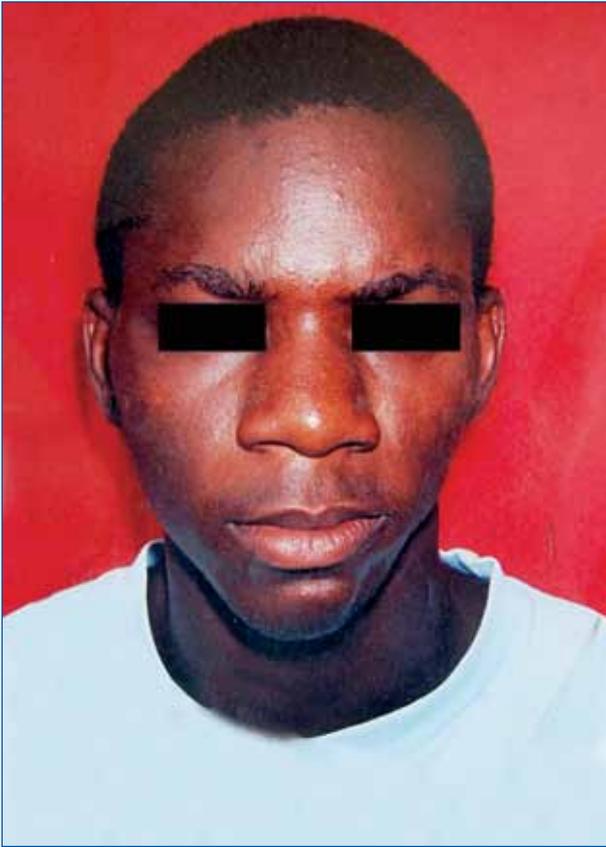


Figure 1. Front view of the face



Figure 2. Left side view of the face

all intact. Obvious wasting of the left masseter, temporalis and buccinator muscles was evident. There was no sign of fasciculations, or clinical evidence of denervation or tenderness. Other muscle groups and joints in the body were essentially normal. The results of a complete blood count and the erythrocyte sedimentation rate were normal. Human immunodeficiency virus serology was negative. Results of available autoimmune tests for antinuclear factor, anti-double-stranded DNA and creatine kinase levels were within normal limits. Lipid panel was also normal. An impression of idiopathic acquired left facial hemiatrophy was made. The patient was referred for a consultation in the plastic surgical unit.

Discussion

Idiopathic acquired progressive hemifacial atrophy, Parry-Romberg syndrome, is a rare entity. Although it was first described in 1825, it remains incompletely understood.¹ This is evident in the nosology and possible aetiology of the condition. While some classify it as one of the trophoneuroses (a group of diseases with autonomic dysfunction and trophic changes), others have classified it as a variant of localised scleroderma, like scleroderma *en coup de sabre* ("like the stroke of a sword") on the upper face and lateral scalp, and others still see it as a variant

of lupus profundus with panniculitis involving the face – all underscoring the poor understanding of the entity.^{2,3} However, it is hypothesised that a disturbance of cerebral fat metabolism may be the primary cause. This can be a result of trophic malformation of the cervical sympathetic nervous system, possibly triggered by pathogenetic mechanisms (such as trauma), viral infection, autoimmunity, heredity or endocrine disturbance.³

In its simplest form, it involves progressive unilateral wasting of the muscles of the face, as in the case described above, without any changes in the skin or bone or manifestations of central nervous system abnormality. However, in the more severe forms, it might be associated with unilateral structural changes in the brain, with atrophy and epilepsy. Localised encephalitis of the Rasmussen's type has been described.⁴⁻⁶ It also has been associated with status migrainosus, an alien hand syndrome from contralateral thalamic degeneration, as well as an infection with *Borrelia burgdorferi* (the aetiological agent of Lyme disease) – all underscoring the complex nature of this clinical entity.⁷⁻¹⁰ The clinical depression from the attendant cosmetic loss as a result of the facial disfigurement is one of the major reasons the patient presents in hospital.¹¹ It may start in childhood, or first present in adulthood.

It is important to differentiate this condition from others that may present with facial disfigurement, so that treatment is not misapplied. Such other conditions include linear scleroderma *en coup de sabre*, with which it is easily confused. Several sources suggest that they are one and the same entity and that they may be different presentations of the same illness.¹² The characteristic skin changes, a tendency of linear scleroderma to involve the upper face and scalp more often than Parry-Romberg syndrome, which has a predilection for the lower face and cheeks, and the autoimmune test results seen in scleroderma may be important differentiating factors.^{4,12,13}

The next condition is acquired localised lipodystrophy, in which there is atrophy with or without hypertrophy of subcutaneous fat. This tends to be more widely distributed beyond the face, even in the idiopathic form (Barraquer-Simmons syndrome), where lipoatrophy may start from the face before involving other parts of the body. Its association with metabolic disorders like hyperglycaemia, dyslipidaemia, positive antinuclear factor and anti-double-stranded DNA in the majority of the patients may be helpful in distinguishing this condition.¹⁴ Localised lipoatrophy may also occur following prolonged, highly active antiretroviral therapy, particularly with the protease inhibitors. But this lipoatrophy usually tends to be more widely distributed.¹⁴ There are currently no clear-cut criteria for the diagnosis of Parry-Romberg syndrome, and hence it remains a diagnosis of exclusion.

Treatment options for Parry-Romberg syndrome include cosmetic surgical reconstruction and possibly the use of immunosuppressants. The latter have been used in some situations with good results. An interdisciplinary approach is usually required.¹⁵⁻¹⁷

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