Parry-Romberg syndrome

Jon Stone

Practical Neurology 2006;6;185-188
doi:10.1136/jnnp.2006.089037

NEUROLOGICAL RARITY

Practical Neurology 2006; 6: 185-188

Parry-Romberg syndrome

Jon Stone

Parry-Romberg syndrome, which is also called progressive facial hemiatrophy, overlaps with a condition known as linear scleroderma "en coup de sabre". It is a rare, acquired, neurocutaneous syndrome of unknown aetiology. The principle features are atrophy of the soft tissues, and sometimes the bone, on one half of the face or forehead without facial weakness. Sometimes the atrophy extends to the limbs, usually on the same side, and there may be various ophthalmological and neurological complications. As a neurologist you should know about this syndrome because you will probably encounter at least one or two cases during your career, perhaps to assess new onset facial asymmetry, hemi-masticatory spasm, or because a patient already with the diagnosis has developed epilepsy or difficult migraine. Patients with Parry-Romberg syndrome present to various other clinics—especially dermatology, plastic surgery, and rheumatology.

TABLE 1 Parry-Romberg syndrome: clinical features (with estimates of their frequency)

- Facial hemiatrophy of fat, skin, connective tissue, muscle, and/or bone (100%)
- Hemiatrophy of contralateral or ipsilateral arm, trunk, or leg (20%)
- Atrophy of tongue (25%)
- Dental abnormalities (50%) and trismus/jaw symptoms (including hemi-masticatory spasm) (35%)
- Migraine/facial pain (45%)
- Ocular abnormalities including globe retraction, uveitis, pupillary abnormalities, restrictive ocular myopathy (mimicking Duane's syndrome), heterochromia
- Epilepsy (10%), sometimes associated with ipsilateral brain changes on MRI (5%)
- Vitiligo, hair depigmentation/hyperpigmentation (20%)
- Brain MRI abnormalities—usually ipsilateral but sometimes contralateral in grey and white matter

Jon Stone
Consultant Neurologist and Honorary Senior Lecturer, Department of Clinical Neurosciences, Western General Hospital, Crewe Road, Edinburgh EH4 2XU, UK; Jon.Stone@ed.ac.uk

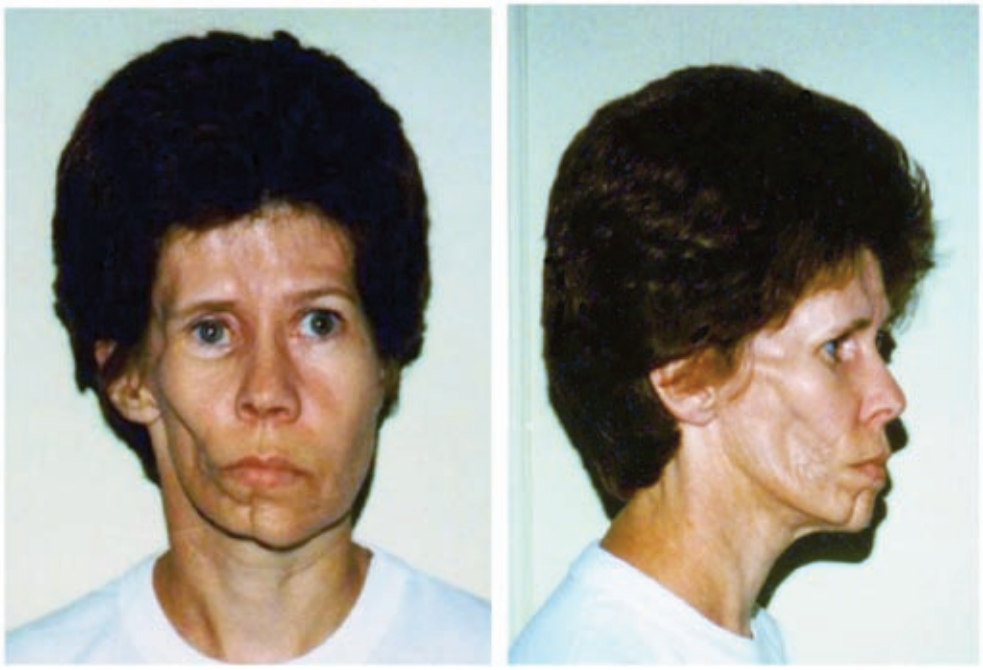
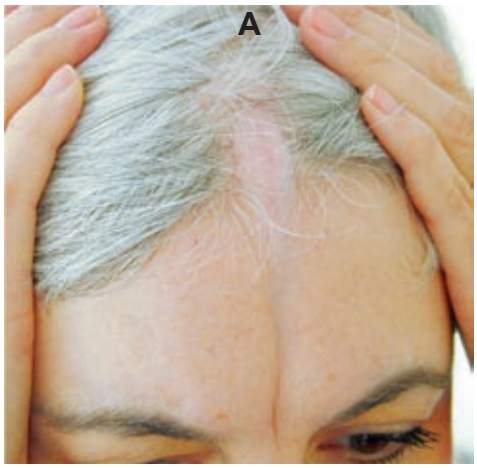


Figure 1
Marilyn—one of the moderators of the "Romberg Connection", a web organisation for people with Parry-Romberg syndrome (reproduced with permission from Lippincott, Williams and Wilkins).[3]



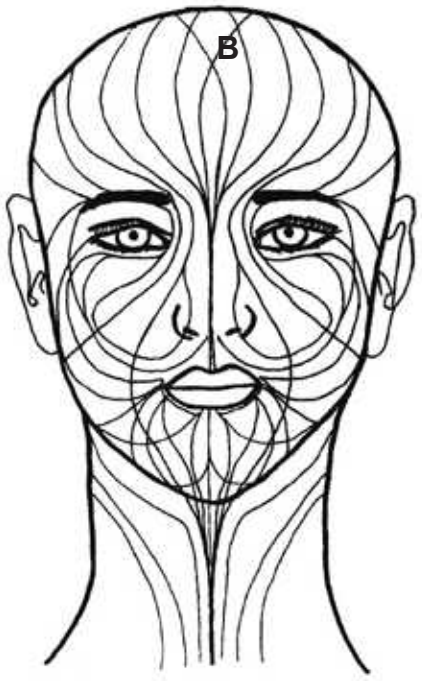


Figure 2
(A) Scleroderma "en coup de sabre" often occurs together with facial hemiatrophy in Parry-Romberg syndrome (reproduced with permission from Dr Ken Katz).[6] It tends to follow Blaschko's lines (B), which are embryological lines that many skin diseases favour (reproduced with permission from Elsevier).[1]

CLINICAL FEATURES

The main feature is hemiatrophy of facial tissues, typically fat, but variably skin, other connective tissue, and sometimes bone (fig 1). The jaw, mouth, cheek, or forehead may all be affected. Patients range in severity from those with barely perceptible asymmetry to severe disfigurement. The range of clinical features is outlined in the table. Where the atrophy meets normal tissue on the other side of the face, it may produce a "line". Around 25% of patients with facial hemiatrophy have a more definite vertical or diagonal "line" on their forehead as a result of cutaneous sclerosis (rather than atrophy of the deeper tissues) (fig 2). These lines tend to follow "Blaschko's lines" which are the diagonal/vertical lines on the forehead, of uncertain origin which some skin diseases tend to follow.[1] When a pathological "line" is present it is called scleroderma "en coup de sabre" ("in a sabre cut") and is classified separately in many textbooks as a form of linear scleroderma affecting the head. Scleroderma "en coup de sabre" does, however, appear to be an overlapping condition with Parry-Romberg syndrome and shares a similar list of associated features.[2] The other terms to be familiar with in this area are "morphoea" (which is an umbrella term for sclerodermatous or fibrotic lesions of the skin) and Gower's panatrophy (which is the name for a "Parry-Romberg like" lipoatrophy just affecting a limb). Systemic sclerosis is an unlikely association of any of these conditions.

The main feature is hemiatrophy of facial tissues, typically fat, but variably skin, other connective tissue, and sometimes bone

10.1136/jnnp.2006.089037

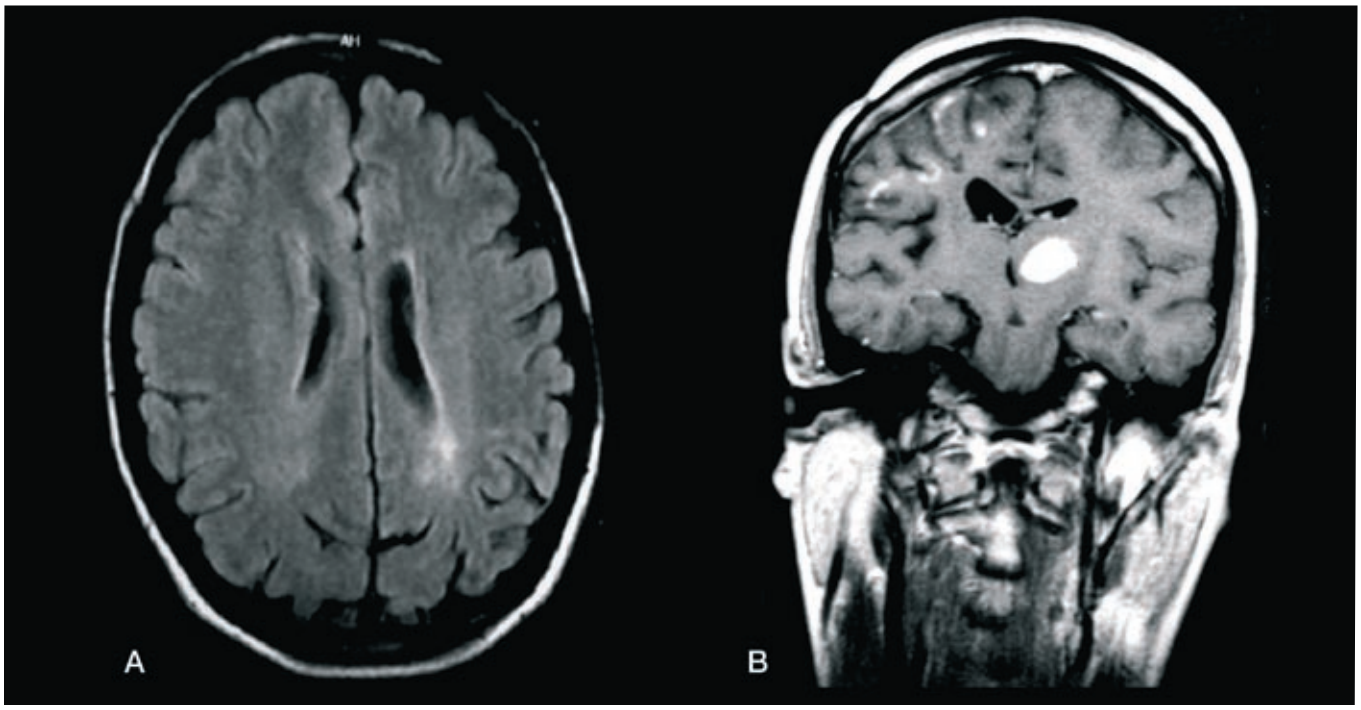


Figure 3
(A) Left sided Parry-Romberg syndrome with ipsilateral cerebral hemiatrophy and white matter hyperintensity (reproduced with permission from Elsevier).[7] (B) Right sided scleroderma "en coup de sabre" with ipsilateral cortical abnormality and a contralateral lesion in the thalamus (reproduced with permission from the BMJ Publishing Group).[8]

NEUROLOGICAL AND PSYCHIATRIC COMPLICATIONS

Migraine and facial pain are the commonest neurological symptoms in this patient group. Occasionally, epilepsy may occur and can sometimes be related directly to a brain abnormality ipsilateral to the skin lesion. This may be visible only on MRI as a lesion in the underlying grey or sometimes white matter. Rarely, the brain imaging abnormalities are contralateral or bilateral (fig 3). Very few of these cases have come to biopsy but those that have mostly suggest an inflammatory problem. There are a range of other cerebral abnormalities which have been associated with Parry-Romberg syndrome and scleroderma "en coup de sabre" on imaging and neuropathological grounds. These include cerebral hemiatrophy, meningeal thickening, cortical dysgenesis, calcified lesions, aneurysms, and intracranial vascular malformations. There is one reported fatal case in a child with associated "Rasmussen-type" encephalitis. From a psychological point of view, the disfigurement is often the worst symptom, particularly since this is an acquired condition.

EPIDEMIOLOGY

This is not well described. A few years ago I carried out a study of the condition, recruiting 205 patients from a US patient run website called the "Romberg's Connection"(fig 4).[3, 4] This website brought together many people with Parry-Romberg syndrome who previously had no opportunity to contact anyone else with the same condition. The interpretation of the findings was limited by the methodology but the average age of onset was around 10 years old with about one third starting after the age of 15 and some as late as 40. The disease typically progresses over a few years (but sometimes much longer) and then arrests. It does not spontaneously improve once it is established. There are anecdotal reports of worsening during pregnancy or after childbirth. The prevalence is at least 1/700,000 and it may be more common in females.

HISTORY

The condition was described by an English physician, Caleb Parry, as early as 1815 and subsequently elaborated in 1846 by Moritz Romberg (he of "Romberg's sign"). In the subsequent 190 years there hasn't been a great deal of progress in understanding or treating the condition—most of the literature consists of just case reports.

AETIOLOGY

There are no systematic studies to guide us but the "best guess" is that it is an autoimmune mediated condition. The skin pathology if caught at the onset shows inflammatory changes. Supportive evidence for an inflammatory hypothesis includes: a higher frequency of

PRACTICE POINTS

- Parry-Romberg syndrome is an acquired disorder principally causing progressive facial hemiatrophy, variably of fat, skin, muscle, and bone. The tongue and limbs may also be involved.
- It overlaps clinically with scleroderma "en coup de sabre" which is linear scleroderma affecting the forehead, usually in a diagonal or vertical line.
- Associated neurological problems include epilepsy (which is sometimes associated with brain abnormalities on MRI), hemi-masticatory spasm, and migraine.
- The pathophysiology is uncertain but the disease is likely to have an acquired autoimmune mechanism.
- Plastic surgery can be helpful, and in severe cases immunosuppression should be considered.

Migraine and facial pain are the commonest neurological symptoms in this patient group

autoantibodies than the general population; the overlap with linear scleroderma; vitiligo; the presence of transient high signal lesions on brain MRI; a couple of neuropathological reports of intracerebral inflammation; and oligoclonal bands in the CSF. Cats and rabbits with experimental lesions of the superior cervical sympathetic ganglion share some of the clinical features of Parry-Romberg syndrome. There are very rare cases (<2%) with a possible hereditary aetiology although it is important not to confuse Parry-Romberg with hemifacial microsomia which is a different congenital disorder. There are anecdotal reports of Parry-Romberg coming on after head trauma and surgery to the face.

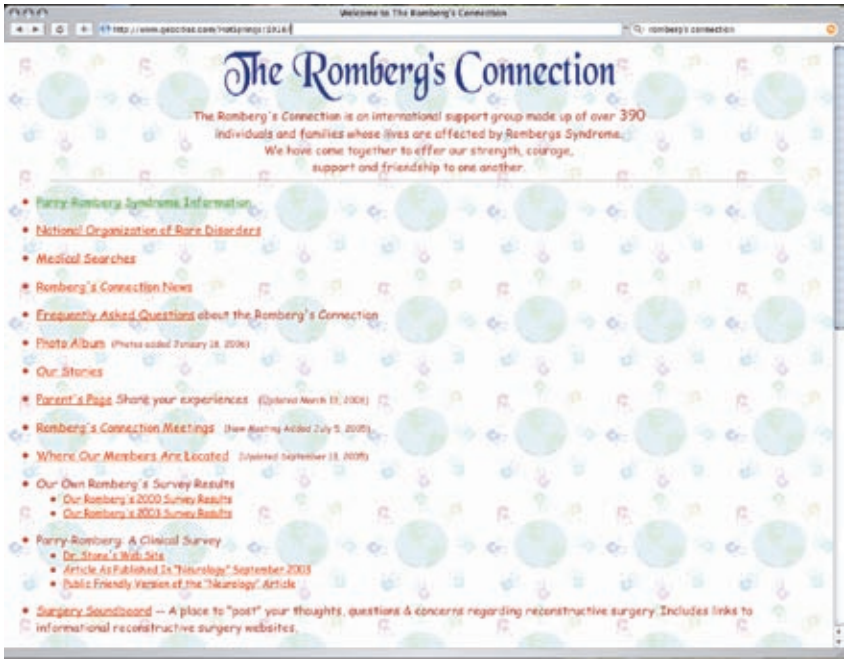


Figure 4
Home page from the Romberg's Connection—an excellent, patient run website.

INVESTIGATIONS

For a patient who only has facial asymmetry, a clinical diagnosis can be made without investigations. MRI is the brain imaging of choice for patients with neurological symptoms. A lumbar puncture and autoantibodies would be reasonable investigations in a patient who is presenting because of epilepsy.

TREATMENT

There are no published trials of treatment. Most patients do not have severe enough disease to warrant immunosuppression but they may be interested in restorative plastic surgery which includes fat or silicon implants, flap/pedicle grafts, or bone implants.[5] Patients tend to be moderately satisfied with the outcome of these procedures. However, they should be made aware that fat injections may simply be resorbed if the disease is still active. For those with more severe and progressive disease, treatments used include methotrexate (for which there is limited evidence in linear scleroderma), corticosteroids, cyclophosphamide, and azathioprine but it is unclear how beneficial they are.

REFERENCES

1. Happle R, Assim A. The lines of Blaschko on the head and neck. *J Am Acad Dermatol* 2001;**44**:612–15.
2. Orozco-Covarrubias L, Guzman-Meza A, Ridaura-Sanz C, *et al*. Scleroderma 'en coup de sabre' and progressive facial hemiatrophy. Is it possible to differentiate them? *J Eur Acad Dermatol Venereol* 2002;**16**:361–6.
3. Stone J. Parry-Romberg syndrome: a global survey of 205 patients using the Internet. *Neurology* 2003;**61**:674–6.
4. Hildebrand T, Neal M. The Romberg's Connection. Available at http://www.geocities.com/rombergs/ (accessed February 2006).
5. Inigo F, Jimenez-Murat Y, Arroyo O, *et al*. Restoration of facial contour in Romberg's disease and hemifacial microsomia: experience with 118 cases. *Microsurgery* 2000;**20**:167–72.
6. Katz KA. Frontal linear scleroderma (en coup de sabre). *Dermatol Online* J 2003;**9**:10.
7. Moko SB, Mistry Y, Blandin de Chalain TM. Parry-Romberg syndrome: intracranial MRI appearances. *J Craniomaxillofac Surg* 2003;**31**:321–4.
8. Stone J, Franks AJ, Guthrie JA, *et al*. Scleroderma 'en coup de sabre': pathological evidence of intracerebral inflammation. *J Neurol Neurosurg Psychiatry* 2001;**70**:382–5.

10.1136/jnnp.2006.089037