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Progressive hemifacial atrophy (Romberg's disease): skeletal involvement and treatment

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SUMMARY. **Progressive hemifacial atrophy (Romberg's disease) manifests variable involvement of the skin, soft tissue and underlying cranio-facial skeleton. Significant bony deformation has been identified in those patients with early onset disease, the result of factors both intrinsic to the disease process and secondary to the abnormal environment in which the skeleton develops (functional matrix). Treatment demands combined osteotomy and augmentation of the skeleton in concert with conventional approaches to soft tissue correction.**

Progressive hemifacial atrophy, eponymously referred to as Romberg's disease, remains an aesthetically, functionally and psychologically challenging disruption of the cranio-facial region on one side. The variable, yet inexorable, progressive atrophy of skin and soft tissues is starkly contrasted by the apparently normal contralateral side.

The approach to management of Romberg's disease demands a detailed three-dimensional documentation of all soft tissue and skeletal disturbances, and how these interplay with the normal temporal sequence of cranio-facial growth.

This paper retrospectively reviews a consecutive series of 11 patients with Romberg's disease, examining their clinical features, with particular reference to the associated underlying skeletal anomalies and their influence on the management approach.

Patients and methods

Between 1978 and 1991, 11 patients with Romberg's syndrome were referred to the Australian Cranio-Facial Unit for assessment. This series included both local patients and those tertiary referrals from interstate and overseas in whom the deformity was manifestly more extreme.

A complete review of their medical records, clinical photographs and radiographic assessments was undertaken, recording details of the disease course, associated features and all soft tissue and skeletal distortions involving the head and neck region. In particular, the skeletal deformity was recorded from axial 2-D CT scans and since 1983 by 3-D CT reconstructions. Several patients, in addition to standard cephalometric radiographs, had sequential CT scan assessment prior to definitive treatment, allowing observation of the change or progression of bony involvement. The approach to treatment and its outcome were studied.

Results

The details of the 11 patients are summarised in Table 1. There were 9 females and 2 males, with involvement of the right and left sides in equal proportion. The age of onset ranged between 9 months and 40 years. Two female patients had distinctly delayed onset of signs after age 35 years.

Only one patient described an episode of local trauma prior to onset, whilst another was noted to have a parent with focal scleroderma not involving the face.

Ipsilateral tongue atrophy was common, in one patient being associated with soft tissue atrophy involving the ipsilateral trunk and upper and lower limbs. Intracerebral calcification was evident in two cases, in one of whom there had been a middle cerebral artery occlusion.

The soft tissue involvement in the head and neck corresponded to the areas of distribution of the trigeminal nerve, without predilection for any one division.

The relationship between the region of skeletal deformity and the age of onset is shown in Table 2. Where onset was as an infant or child less than 5 years of age frontoorbitozygomatic distortion was noted, in addition to midfacial and mandibular asymmetries. Later onset resulted in relative sparing of the orbitozygomatic complex, but consistent findings of progressive changes in the lower face. Figures 1 and 2 graphically record the failure of normal maturation of the facial skeleton on the involved side compared to an apparently normal increase in facial height, width and projection on the contralateral "normal" side.

Table 1 Patient data (n = 11)

Sex		*Age of onset*		*Side involved*	
Female	9	Before 5 years	6	Left	7
Male	2	After 5 years	5	Right	4

Table 2 Facial skeletal involvement according to age of onset

	Frontal	*Orbitozygomatic*	*Maxillary*	*Mandibular*	*Dentoalveolar*
Onset before 5 years (n = 6)	3	5	6	4	4
Onset after 5 years (n = 5)	0	2	3	3	4

Fig. 1

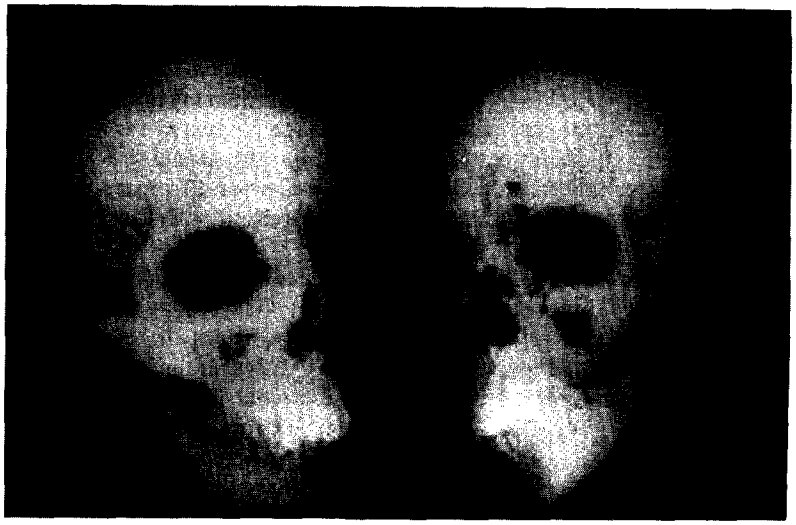

Fig. 2

Figure 1—Severe left hemifacial atrophy with deviation of the facial midline to the affected side. **Figure 2**—3D CT reconstruction of the patient in Figure 1 showing the extent of the underlying bony asymmetry.

Significant bony asymmetry, requiring surgical correction, was present in 6 patients. All had the initial onset of the disease process earlier than 5 years of age (Tables 2 and 4). Surgical correction of orbital dystopia was required in two patients, whilst a further case underwent repeated bone grafting of the orbital walls to correct enophthalmos. Bimaxillary surgery was used to centralise the facial midline and correct the tilted occlusal plane, at the completion of facial growth. In two of these situations, a free vascularised composite tissue transfer was incorporated both to augment soft tissue volume and to bring appropriately contoured vascularised bone to the lower face for mandibular reconstruction.

Free microvascular soft tissue transfer was employed to augment the facial contours in patients with both early and late onset of the disease (Table 3). Groin flaps based on either the deep or superficial circumflex iliac vessels were most commonly employed. All were associated in the long term with a subsequent tendency to sagging due to gravity, requiring revision. Difficulties were also experienced in correcting the paramedian soft tissues in the chin, upper and lower lips and around the alar base. This resulted from failure to achieve interdigitation of the free flap across the midline into the normal opposite side. Where smaller soft tissue volumes were required, lateral arm and forearm flaps were utilised, the latter in

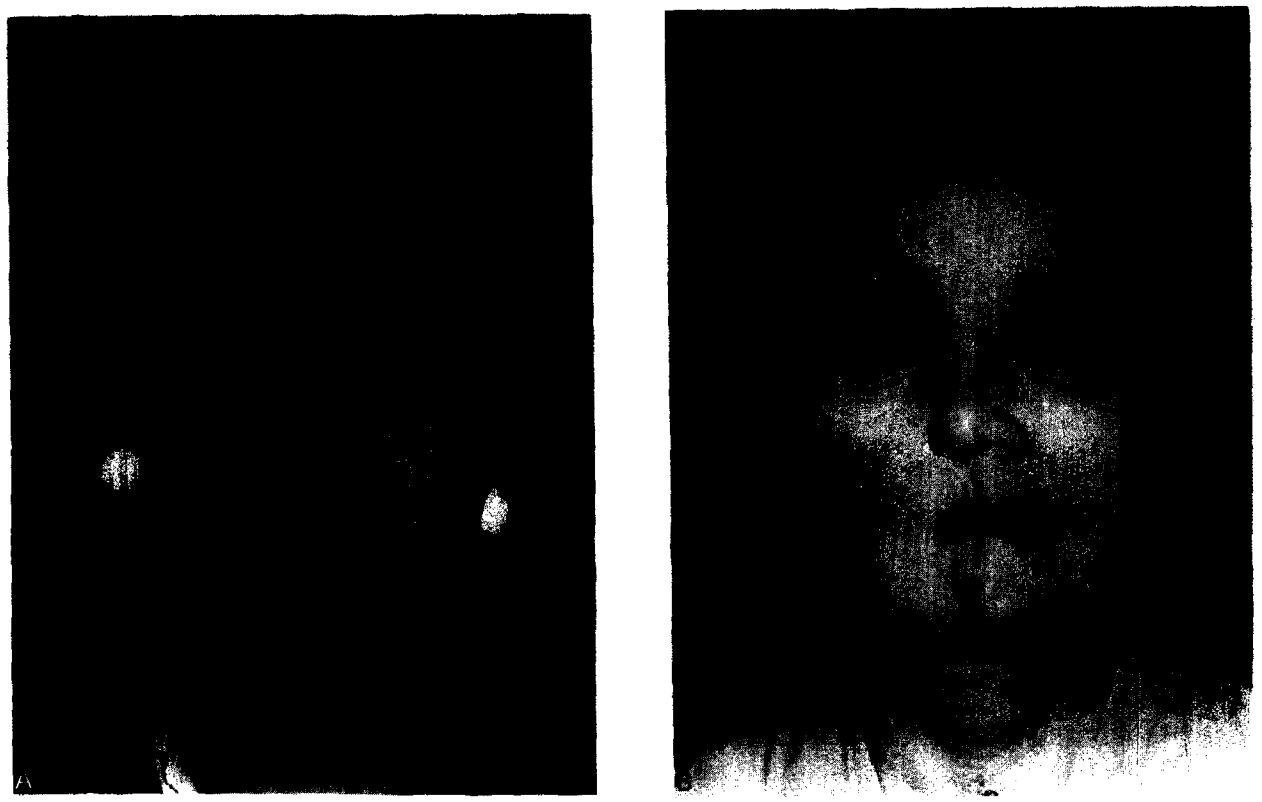

Fig. 3

Figure 3—(A) Early onset (before 5 years of age) right hemifacial atrophy with marked soft tissue and bony deformity. (B) Postoperative reconstruction of the patient in Figure 3 following Le Fort I and bilateral subsigmoid osteotomies, radial forearm and later a lateral arm flap for cheek resurfacing and augmentation.

Table 3 Techniques of soft tissue augmentation according to age of onset

	Non-vascularised augmentation	*Vascularised free flap augmentation*
Onset before 5 years	0	6
Onset after 5 years	1	3

resurfacing the cheek in a case treated elsewhere with multiple injections of liquid paraffin (Fig. 3A). This same patient later required a second soft tissue free flap (de-epithelialised lateral arm flap) to correct the persistently deficient and asymmetric soft tissues (Fig. 3B).

Discussion

Described first by Parry in 1825, but conventionally bearing the name of Romberg (1846), who detailed the clinical findings, Romberg's disease is more precisely referred to as progressive facial hemiatrophy or hemifacial atrophy. The underlying cause remains elusive and largely speculative. Indeed, Rees (1976) suggested that Romberg's disease probably represents a syndrome rather than a specific disease entity.

However, the clinical course and pattern of facial soft tissue involvement is more consistently recorded. Characteristically, beginning during the first or second decade of life as localised and progressive atrophy of the skin and subcutaneous tissues within the dermatome of one or more branches of the trigeminal nerve of one side of the face, it stabilises over 2–10 years. Occurring slightly more frequently in females, both sides of the face are affected with equal frequency. Bilateral involvement has been reported (Rogers, 1963), although no cases were seen in this series.

The classical earliest sign, the "coup de sabre", was recorded in half of our series and reflects the more common early soft tissue involvement in the upper face (frontal and maxillary dermatomes). In the presence of prolonged active disease, there is com-

Table 4 Techniques of skeletal reconstruction according to age of onset

	Onlay bone graft	*Osteotomy*			*Osteocutaneous free flap*
		Orbital	*Maxillary*	*Mandibular*	
Onset before 5 years	6	2	4	4	2
Onset after 5 years	1	0	0	1	1

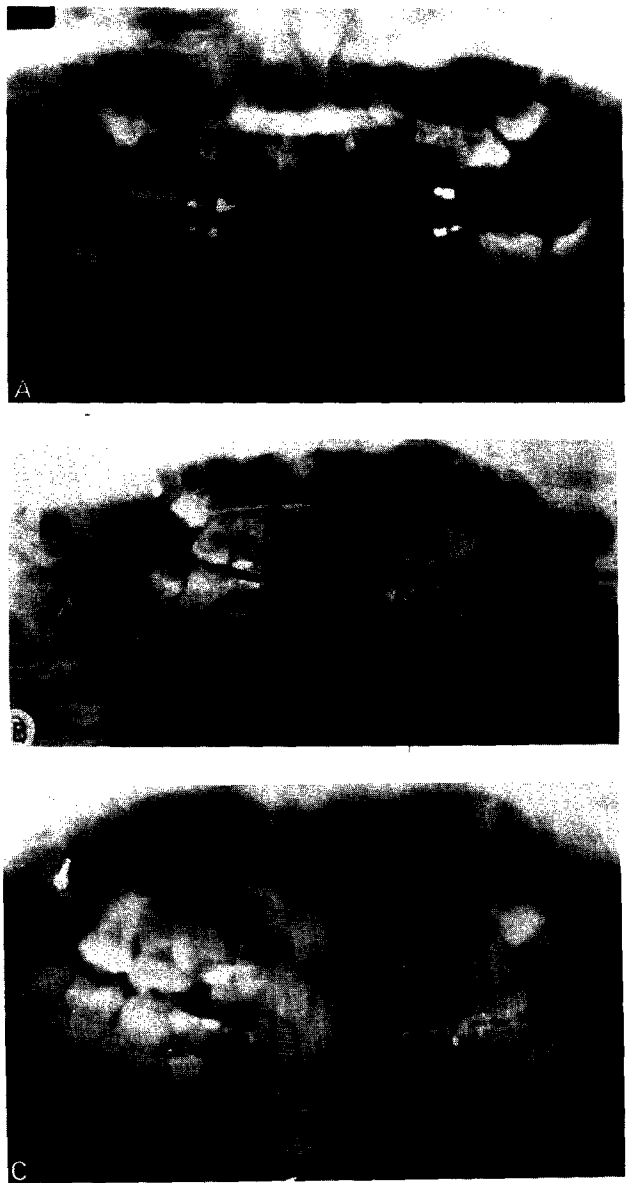

Fig. 4

Figure 4—Delay in right dental maturation associated with decreased bi-maxillary bony development in a patient with right hemifacial atrophy at (A) 9 years of age, (B) 11 years of age, (C) 14 years of age.

monly extension to involve the whole hemiface, *i.e.* soft tissue manifestations are temporally dependent. Late onset of the disease process appears to be characterised by soft tissue atrophy in the lower face.

In contrast, the details of skeletal involvement, whether primary or secondary to the disease process, have been infrequently recorded (Pensler *et al.*, 1990). The advent of cephalometric radiographs and direct and reformatted 2-D CT scans together with techniques of 3-D reconstruction provides the facility to document both qualitatively and later quantitatively the pattern and progression of cranio-facial skeletal disturbance. Most commonly, bony hypoplasia was evident in the mid and lower face. There was relatively infrequent involvement of the frontal region, despite this being the commonest area of early soft tissue disturbance. The orbitozygomatic complex showed marked involvement in three cases, usually with the combination of soft tissue and skeletal changes of enophthalmos and vertical orbital dystopia.

The derangement of cranio-facial skeletal growth in Romberg's disease is unlikely to be solely due to an isolated intrinsic process. Recent reports identify a chronic cell-mediated vascular injury and incomplete endothelial regeneration along branches of the trigeminal nerve (lymphocytic neurovasculitis) as the pathogenetic event in the soft tissues of Romberg's disease (Pensler *et al.*, 1990). To date there has been no confirmation of extension of these electron microscopically demonstrated events into the underlying bone, although this could be expected.

Disturbance to normal dental growth and development, with foreshortening of dental roots, has been previously reported in the mandibular dentition (Foster, 1979). In this series several cases with early onset disease demonstrate this dental hypoplasia in both the maxillary and mandibular dental arches (Fig. 4). Such alterations, as seen in paediatric patients undergoing local head and neck radiotherapy for malignancy, are attributable to the intrinsic pathological process occurring during the development and maturation of the secondary dentition, not the result of an abnormal soft tissue "functional matrix" (Moss, 1972).

The restrictive influence of the abnormal soft tissue envelope (early onset disease) at the time of accelerated growth of the mid and lower face undoubtedly compounds any primary skeletal growth disturbance, manifesting as maxillary and mandibular hypoplasia in all dimensions, with shift of the lower facial midline to the involved side.

Late onset soft tissue changes, as in a well developed frontal "coup de sabre", may be associated with minor notching or ridging of the underlying frontal bone, but no major changes in facial skeletal base bones are recorded in these late cases.

Thus, irrespective of aetiology, if the disease process involves bone, it would seem to exert an effect on skeletal morphology only during periods of facial growth acceleration. Alternatively, even if there is no primary, intrinsic involvement of bone, the presence of an abnormal soft tissue matrix during the period of rapid bone growth produces marked skeletal dysmorphology (Moss, 1972).

All patients in this series with early onset disease required complex skeletal surgery as part of their reconstruction. Where vertical orbital dystopia existed, orbital translocation in concert with extensive inlay bone grafting produced satisfactory repositioning of the globe in both vertical and anteroposterior dimensions. This has been undertaken after the apparent cessation of active disease, and ideally just prior to the midfacial growth spurt when the unerupted secondary dentition is sufficiently inferiorly located to permit safe orbital osteotomy.

Correction of the midfacial and occlusal distortion frequently demands a combined orthodontic-surgical management, the latter involving maxillary osteotomy to rotate the maxilla three dimensionally in space, levelling the tilted occlusal plane and centralising the face, together with mandibular osteotomies (sagittal split or subsigmoid) to re-establish the predetermined

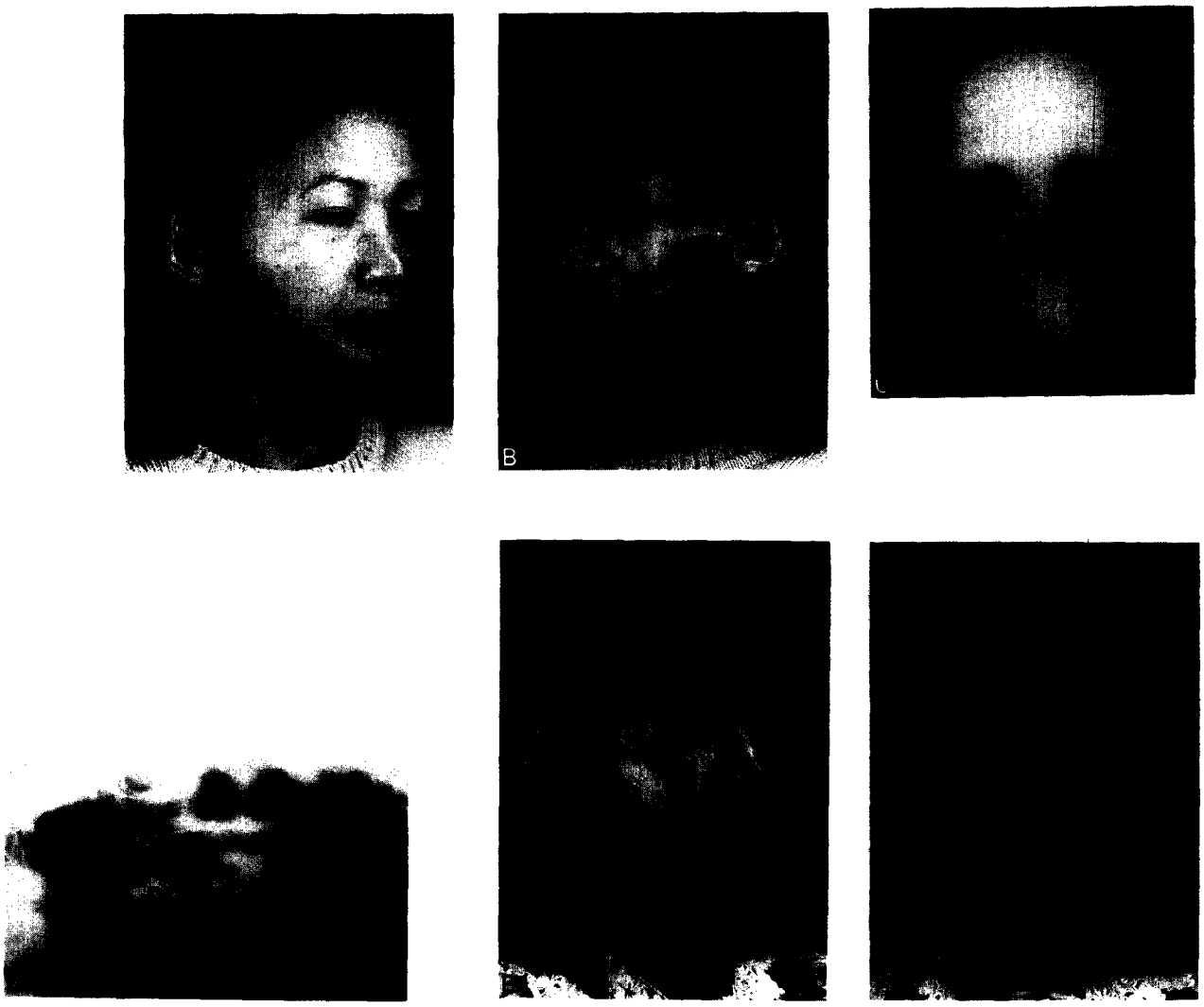

Fig. 5

Figure 5—(A, B) Oblique views of early onset case confirm the extreme facial asymmetry. (C) 3D CT reconstruction details the midfacial and mandibular asymmetry. (D) Orthopantomogram similarly records the left maxillary and mandibular distortion. (E, F) Clinical outcome following bimaxillary surgery in concert with composite osteocutaneous free flap transfer.

occlusion. The surgical elements of this approach have conventionally been implemented following the cessation of active disease and at the completion of facial growth (Fig. 5).

Invariably in grossly distorted cases, the addition of composite microvascular free tissue transfer is useful to augment both the soft tissue and skeletal reconstruction (Fujino *et al.*, 1985; Jones, 1989). The osseous component of the osteocutaneous flap, such as the deep circumflex iliac artery flap, is rigidly fixed as an onlay and inlay vascularised bony augmentation of the mandibular osteotomy site on the affected side (David *et al.*, 1988). It then potentially contributes both to the stability of the orthognathic osteotomies and to the production of a predictable correction of facial skeletal asymmetry.

Soft tissue augmentation for symmetry in the severe cases requires volume replacement beyond that which is initially appreciated, and can be produced predictably only by a de-epithelialised free flap transfer (Shintomi *et al.*, 1981; O'Brien *et al.*, 1981; Jurkiewicz and Nahai, 1985; Dunkley and Stevenson, 1990). On two occasions in this series there was a need for a second free flap, having underestimated the initial soft tissue volume deficit. Conventional placement of the soft tissue elements of the free flap in the subcutaneous plane, with percutaneous bolster sutures to locate the flaps has been unsatisfactory, with uniform sagging of the soft tissues under the influence of gravity. Instead, perhaps, subperiosteal positioning of the deepithelialised soft tissue flap, with suture attachment at multiple points to the underlying skeleton, would prevent this. In the severe cases, this distinction between subcutaneous and subperiosteal is both arbitrary and somewhat artificial, but deep fixation may aid stability of positioning. Inserting the flap with the dermis on the deep aspect may provide a safe and strong layer adjacent to periosteum or bone with which to secure the flap.

By employing multidisciplinary assessment and utilising both cranio-maxillo-facial and microsurgical techniques of reconstructing the underlying skeleton and surrounding soft tissue envelope in this very deforming condition of Romberg's disease, it is now

possible to approach that very challenging functional and aesthetic ideal of the contralateral normal side of the face, seldom in a single surgical procedure, but usually as a planned, staged composite soft tissue and skeletal reconstruction.

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