Jaw involvement in sclerosteosis: a case report

R E WOOD, G KLEYN, C J NORTJÉ and F GROTEPASS

From the Department of Maxillofacial Radiology, Faculty of Dentistry, University of Stellenbosch, Tygerberg, Republic of South Africa.

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A case is presented in which a patient with the sclerosing bone dysplasia, sclerosteosis, also had a lobulated torus palatinus, torus mandibularis and increased height of the mandible. The patient was initially diagnosed as having benign, dominantly-inherited osteopetrosis. The characteristic oral features of sclerosteosis helped to establish the correct diagnosis.

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Sclerosteosis is one of the group of disorders known as sclerosing bone dysplasias (4). The condition was first described by Truswell (13) but the term sclerosteosis is attributed to Hansen (8). It is most common in the Afrikaner population of South Africa but is otherwise considered rare. It is inherited as an autosomal recessive (2), in contrast to benign osteopetrosis which is dominant (9), and is similar to another sclerosing bone dysplasia, van Buchem's disease, in that both are recessive, exhibit the radiographic features of osteopetrosis and are associated with jaw enlargement. However, the gnathic distortions in sclerosteosis are said to be more marked (4).

The clinical features of sclerosteosis include severely distorted facies, malocclusion, relative midface hypoplasia, tall stature, transient facial nerve palsy, progressive enclosure of skull foramina and progressive diminution of cranial capacity with elevated intracranial pressure (1,3,4). Syndactyly is frequent but not universal. Skull radiographs show cranial thickening, especially of the posterior cranial base, and a massive increase in height of the mandible (4).

It is vital that this condition be differentiated from osteopetrosis since sclerosteosis may be associated with sudden death secondary to impaction of the medulla oblongata in the foramen magnum due to the elevated intracranial pressure (4). In addition, mandibular osteomyelitis, a common feature of osteopetrosis (10), is not associated with sclerosteosis (3). The purpose of this paper is to present a case of sclerosteosis which was originally misdiagnosed as osteopetrosis and review the distinguishing oral features of this disease as compared with the other sclerosing bone dysplasias.

CASE REPORT

M.D., a healthy 24-year-old male, presented to Tygerberg Hospital complaining of painful headaches, back pain and a jaw deformity: his chin and lower jaw had become so enlarged that the chin strap on his motorcycle helmet no longer fitted. He reported he had been well until 7 years previously when he began to experience painful bilateral temporal headaches: five years later he had a transient paralysis of the left facial nerve and six weeks ago he had spontaneous lacrimation of the left eye and dilation of the left pupil.

He denied any fractures despite a history of contact sports and was otherwise healthy. There was no relevant family history.

Physical examination revealed a healthy, tall (194 cm), adult male with large facial features but clinically normal hands: there was marked increase in the height of the mandible (Fig. 1) with relative midface hypoplasia. Intra-orally, there were lobulated tori at the apex of the hard palate and lingual to the right and left mandibular premolars (Fig. 2). The alveolar process of the maxilla was widened bilaterally but the occlusion was normal. The patient had received routine dental care, including extractions, with no untoward effects. Cranial nerve examination revealed a slight paralysis of the left facial nerve which was verified by electromyography. Both eye fields were normal. Serum electrolytes, calcium, phosphate and alkaline phosphatase, and renal and liver function tests, including arginine renal acidity, were all normal. A diagnosis of osteopetrosis complicated with mandibular prognathism and facial nerve paralysis was made.

He was therefore referred to the Oral Surgery Department where radiographs of the skull showed

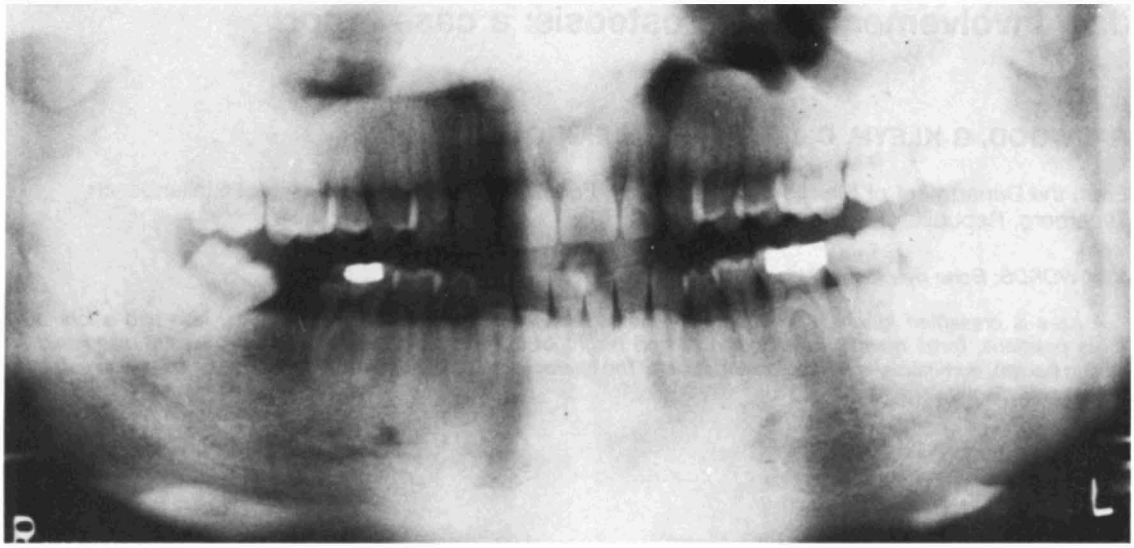

Fig. 1. Panoramic radiograph showing generalized increased radiopacity of bone and gross increased height of the mandible.

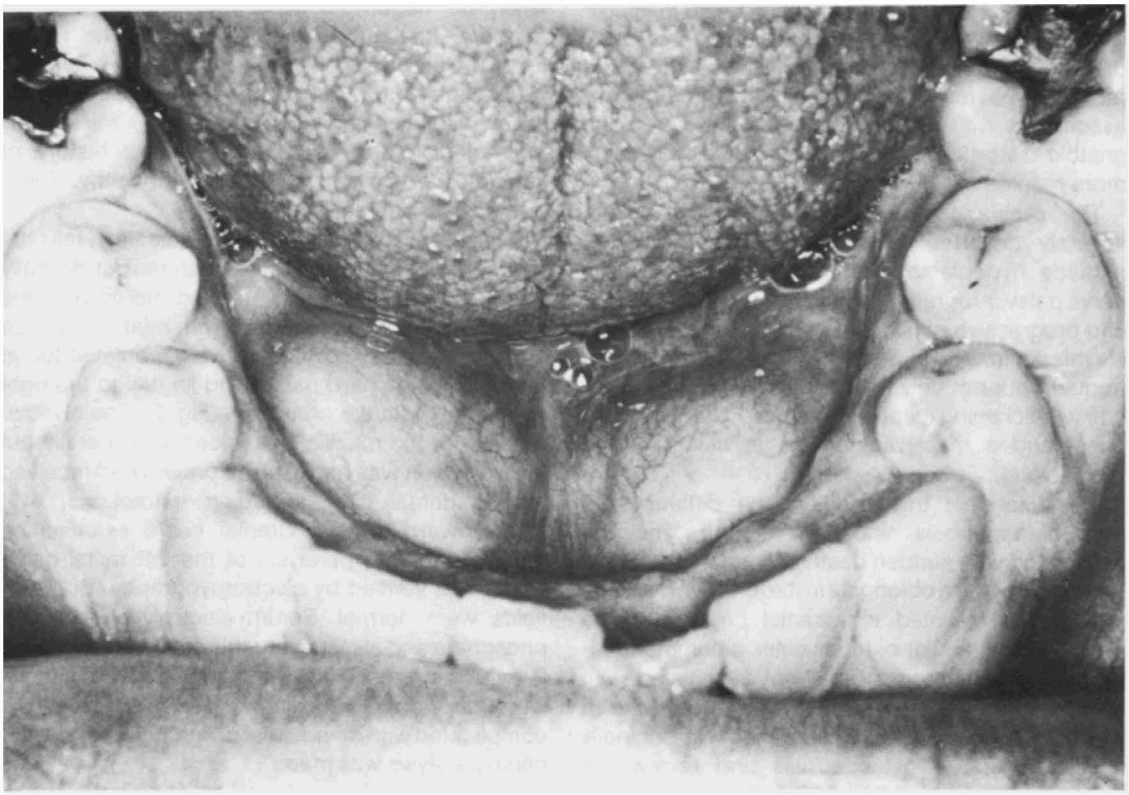

Fig. 2. Intra-oral photograph showing tori mandibularis lingual to the premolar teeth.

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an increased thickness of the vault, especially in the occipital bone and base (Fig. 3). Carpal radiographs revealed involvement of the bones of the hand: there was increased girth of the diaphyses and relative thickening of the phalangeal cortices (Fig. 4), while the metacarpals and phalanges were cigar-shaped. The remainder of the skeleton exhibited a generalized increased density. These new findings were more consistent with a diagnosis of sclerosteosis rather than osteopetrosis and the patient was therefore referred to the neurosurgical service for their advice. Since this condition is progressive and because the bone is excessively hard (5), no cosmetic surgical procedures have been planned.

DISCUSSION

Although the original diagnosis was osteopetrosis, subsequent re-evaluation in light of the clinical and oral radiographic findings led to the correct diagnosis of sclerosteosis. The separation of osteopetrosis and sclerosteosis is vital. Sclerosteosis, unlike benign osteopetrosis, is a potentially lethal condition (4). From an oral surgery standpoint, patients with sclerosteosis do not suffer from dyshaemopoiesis or osteomyelitis (3) but dental extractions are very difficult due to the unyielding nature of the alveolar bone (5).

The presence of mandibular and maxillary tori may be related to sclerosteosis (5). Suzuki and Sakai (11) postulated that these structures are inherited as an autosomal dominant, similar to osteopetrosis (9) but not sclerosteosis (13). It is not known whether this patient's parents had tori.

It is important to realize that radiographic evidence of periapical disease will be obscured in patients with abnormally dense bone (14). Should this be suspected, then the reliability of radiographic detection will be reduced, and other methods of diagnosis will have to be utilized.

Differential diagnosis may prove difficult since the sclerosing bone dysplasias share a number of common clinical features. Malignant, *recessively inherited osteopetrosis* is characterized by early onset, few gross structural jaw changes, dyshaemopoiesis, high incidence of osteomyelitis, and frequent fractures (6). Benign, *dominantly inherited osteopetrosis* exhibits normal mandibular height, a

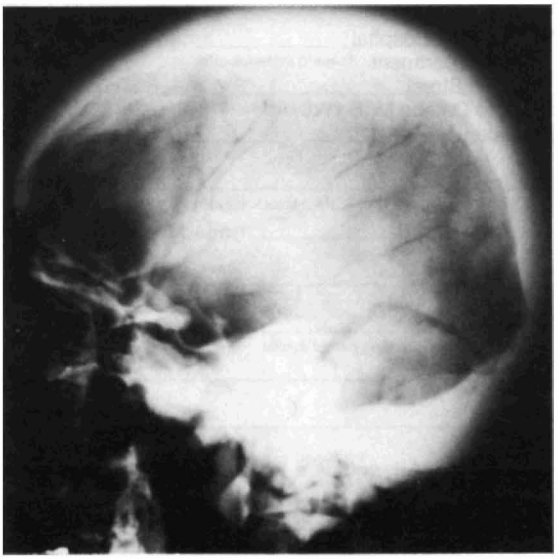

Fig. 3. Lateral skull radiograph showing increased width and density in the calvarium, most conspicuous in the occipital region.

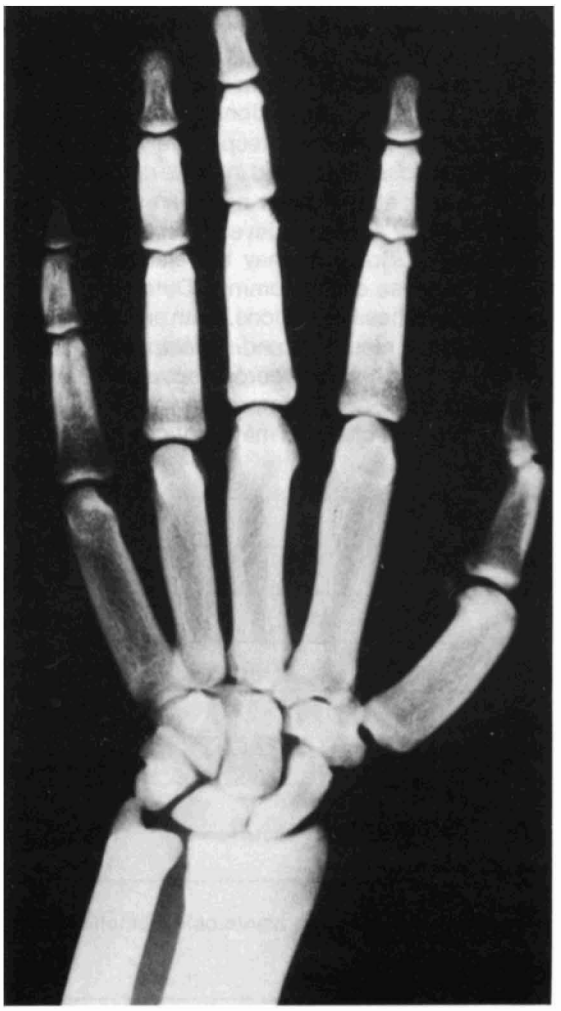

Fig. 4. Hand radiograph showing cigar-shaped metacarpals, opacification of the radius and ulna and moderate metacarpophalangeal cortical thickening.

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high incidence of osteomyelitis and fractures, and an autosomal dominant pattern of inheritance (7,9,12). Both sub-types of osteopetrosis have uniform sclerosis of the skull, whereas sclerosteosis has greater occipital involvement (4).

Patients with *pycnodysostosis* are of significantly shorter stature, prone to mandibular infections (6), and have micrognathia with obtuse gonial angles. Cranial sutures may be widened and fontanelles may persist until adulthood (4).

Jaw involvement is not as marked in *metaphyseal dysplasia* although mandibular prognathism is present. *Craniometaphyseal dysplasia* and *frontometaphyseal dysplasia* are also distinguished on the basis of conspicuous metaphyseal changes (4). *Craniodiaphyseal dysplasia* and *diaphyseal dysplasia*, unlike sclerosteosis, manifest gross diaphyseal changes characterized by thickened and fusiform diaphyses. *Osteopathia striata, osteopoikilosis*, and *melorheostosis* differ from sclerosteosis with respect to the pattern of bone changes (striated, punctate and molten wax respectively). *Ostectasia with hyperphosphatasia* and *infantile cortical hyperostosis* occur at a younger age than sclerosteosis whereas *Paget's disease* develops in older patients. *Van Buchem's disease* may be related to sclerosteosis because of the common Dutch heritage of patients with these conditions. Both are inherited as an autosomal recessive and develop jaw enlargement. However, cranial hyperostoses and alterations in the tubular bones are more marked in the latter and the skeletal changes may continue throughout life (4,5).

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Address:
Dr R. E. Wood
st Michael's Hospital
Dental Department
30 Bond Street
Toronto Ontario M5B 1W8
Canada

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