Sclerosteosis in Children

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Abstract. Craniotubular dysplasias and craniotubular hyperostoses are a group of confusing disorders in which the cranial involvement may cause compression of facial and auditory nerves. Two of the hyperostotic conditions that regularly produce cranial nerve compression are sclerosteosis and the recessive form of endosteal hyperostosis (van Buchem's disease). Both have identical radiological features with cranial involvement presenting in childhood. The Dutch and South African cases have come from relatively close-knit communities, those in the latter country being Afrikaners who had their origins in Holland. The radiological features of three such cases of sclerosteosis are briefly presented.

Key words: Sclerosteosis - Craniotubular dysplasias - Bone dysplasias - Bone sclerosis

There are a variety of conditions that produce change in skull and tubular bones and three of them sclerosteosis, endosteal hyperostosis and craniodiaphyseal dysplasia produce symptoms of cranial nerve compression. These craniotubular disorders cause skull sclerosis, hypertrophy of the mandible and cortical hyperostosis of the tubular bones. Craniodiaphyseal dysplasia [10, 13, 20] may represent a group of conditions [13] and will not be further considered as although it causes severe facial and long bone changes, the skull involvement does not inevitably produce nerve compression. Sclerosteosis and endosteal hyperostosis are considered as two separate entities in the International Nomenclature for bone dysplasias [12]. The main features that distinguish sclerosteosis from endosteal hyperostosis (van Buchem type) are syndactyly with minor nail changes and radial deviation and to a lesser degree the relative gigantism and more pronounced cranioptathy that may occur in the former [2]. In sclerosteosis overgrowth of bone in the skull is a major feature and it also occurs in the ribs, clavicles, long bones and hands. The spine and pelvis is usually relatively spared but may be involved in aged patients. In children the skull involvement may be the only notable feature.

The majority of cases of sclerosteosis have been reported from South Africa [3, 4, 5] but reports have also emanated from widely separated areas such as New York [11], Switzerland [16] and Japan [17].

This report concerns the radiological appearances of three cases of sclerosteosis in children which had referrals to the Groote Schuur Hospital in 1978.

Material

Case 1: G. G., female, aged 16 years. This child was born with syndactyly of the thumb and adjacent fingers on the left hand which required surgical operation at the age of five years. Her radiographs were first seen at this hospital when she was aged ten years and a provisional diagnosis of osteopetrosis was entertained.

On the current admission to hospital she was noted to have an enlarged jaw and prominent zygomatic bones. The fingers were deformed and there was an absence of nails. She was deaf and had to wear a hearing aid and on examination was found to have bilateral papilloedema and a conductive nerve deafness. A definitive diagnosis of sclerosteosis was made and a posterior fossa decompression through dense occipital bone was performed by Professor J. C. de Villiers in February 1978.

The radiographs of skull and hand are shown in Figures 1, 2, 3, 4, 5 and 6. Sclerosis was present in the bones of the thorax (Fig. 7), and pelvis but was not marked in the rest of the skeleton.

Case 2: N. H., female, aged 41/2 years. This child had bilateral webbed index and middle fingers at birth. There was no medical history of note till the age of eleven months when she developed a transient left facial palsy. Three months later a right facial palsy developed and further intermittent facial palsies necessitated a surgical decompression this year. The radiographs of her skull, knees and hands are shown in Figures 8, 9 and 10.
Fig. 1. Lateral skull aged 10 years showing thickening of the bone of the calvarium and base

Fig. 2. Lateral skull aged 16 years showing progression of sclerosis at calvarium and base. Arrows indicate a recent craniostomy site

Fig. 3. Oblique of mandible showing the sclerosis and hypertrophy

Fig. 4. Polyaxial tomography showing optic foramina with slight narrowing of the left optic foramen (arrow)

Fig. 5. C. T. scan taken immediately prior to surgery. Calvarial thickening is gross but the ventricles are of normal size

Fig. 6. Right hand. Cortical thickening and faulty modelling of metacarpals and phalanges. Although ungual and digital hypoplasia may occur, deformity of the terminal phalanges of the index and middle fingers was a result of syndactyly correction