Proceeding S.Z.P.G.M.I vol: 10(3-4) 1996, pp. 88-89.

Sclerosteosis: A Case Report

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SUMMARY

Hansen in 1967 first proposed the term sclerosteosis to describe a disorder characterized by familial generalized osteosclerosis associated with hyperostosis of the calvarium, mandible and clavicle, syndactly and other abnormalities of the digits, thus differentiating it from osteopetrosis and others sclerosing disorders.

CASE REPORT

This patient was a 26 years old Pakistani woman. She was normally delivered, the mother and father were in good health. It was noted at the time of birth that the baby had dysplasia of nails and syndactly of left index finger and thumb. At the age of 5 years, it was first noted that the facial movements were slightly limited on the right side and right eye was somewhat smaller than left. At 23 years of age, she suffered from mild intermittent headache which gradually increased. When she was 25 years old she presented to us with low backache and pain in the lower limbs.

Physical examination showed a relatively well nourished tall woman. She had exophthalmos and broad flat nose. Mandible was slightly broadened. She had slight trouble in hearing and high pitched voice. Right facial palsy was also noted. Visual acuity was normal. Terminal phalanx of right index finger deviated radially. On the left side there was syndactly between thumb and index finger and terminal phalanx of left index was hypoplastic. There was syndactly of second and third toes of left side and second, third toes of right side. The range of motion was normal. Sensations were diminished on right side and right lower limb was weak.

Roentgenographically the cranial vault, base of the skull and mandible were markedly sclerotic and hyperostotic. Frontal sinuses were remarkably enlarged. The clavicle and ribs were widened and slightly sclerotic.

The vertebral bodies and pelvis were normal in shape but showed marked diffuse condensation of bone. The long tubular bones showed reduced tabulation with increased bone density. The metaphyseal flare was not present in the hand, bones were cigar or rod shaped and marked diaphyseal thickening was noted in all metacarpals and phalanges. The metatarsals were markedly thickened in the diaphyseal portion. Her brother had similar problems physically & roentgenographically. No such case was known to the family in the past.

DISCUSSION

This disorder was considered to be a type of osteopetrosis before Hansen's work in 1967. Truswell 1958 recognized the existence of this disorder when he described six patients in an article Osteopetrosis with syndactly, a morphological variant of Albers - Schonberg disease[2]. Sclerosteosis is inherited as an autosomal recessive. About 60 cases have been reported, majority among the African community of South Africa, a woman from Switzerland a girl in Japan and a consanguineous family in Brazil [3-9].

Parental consanguinity was also observed[4,6,8-10]. All of the characteristic clinical features can be observed in the early years of life. There is progressive overgrowth and sclerosis of the skeleton particularly the skull. Typical facies in infancy is characterized by high forehead, ocular hypertlorism, broad flat root of the nose and broadened squared mandible. Height and weight are often excessive. The abnormalities in the fingers and toes are variable. Commonly there is bilateral, often asymmetric, cutaneous syndactyly. Th nails of the involved fingers and toes are often dysplastic or absent. Another important clinical feature is the

involvement of cranial nerves. Facial palsy may be a presenting feature. Characteristically it is unilateral for many years. In some cases optic atrophy, reduction in visual field and nystagmus or exophtholmos are present. In adulthood, raised intracranial pressure may cause headache. Several patients have died suddenly from impaction of brainstem in the foramen magnum.

Cutaneous or bony syndactyly of 2nd and 3rd fingers serves to distinguish sclerosteosis from the other disorders.

Roentgenographically, gross widening and sclerosis of the skull is the predominant feature. The base of skull is especially thickened, the body of mandible is greatly thickened and the angle is wider than normal. The clavicles and ribs are broadend and scapulae, pelvis, vertebral bodies and long bones are uniformly sclerotic but their contours are normal. The long tubular bones show decreased diaphyseal modelling resulting in a straight, rodlike shape and similar changes are observed in short tubular bones of hands and feet. Deviation of one or several fingers are also noted.

Nature of the basic defect is unknown, but following comprehensive histological studies Stein et al. (1983) suggested that the condition might be a disorder of osteoblast hyperactivity.

Differential diagnosis

The Roentgenograms of the hands reveal cardinal findings for the differentiation of each of the hereditary sclerotic disorders that cause symmetrical widening and hyperostosis or osteosclerosis of the tubular bones or both.

In osteopetrosis there is club shaped metaphyseal flare with marked amorphous condensation of bone shadow. Longitudinal striations of lesser degrees are observed.

In pyknodysostosis, all the tubular bones show markedly diffuse condensation of bones. Metaphyseal flare is not observed, acro-osteolysis of terminal phalanges is one of the most characteristic feature of this disease.

In familial metaphyseal dysplasia (Pyle's disease), marked metaphyseal splaying with decreased bone density in the metaphysis is noted. Thickening and condensation of bone shadow are also noted at the distal 2/3rd of the diaphysis of the each phalynx and of the first metacarpal and at the proximal 2/3rd of each diaphysis of the 2nd to 5th metacarpals.

In pachydermoperiostosis, remarkable symmetrical external hyperostosis is noted at diaphysis of each tubular bone. The shape of the metaphysis and epiphysis remain quite normal.

In sclerosteosis, each of the tubular bones show rod-shaped or cylindrical deformity with diaphyseal thickening and diffuse condensation of bone shadow.

In Camurati Engelmann's disease or progressive diaphyseal dysplasia. The diaphysis of each long tubular bone shows hyperostosis with marked condensation of bone shadow and irregular contours, while the short tubular bones are rarely affected.

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