ALBERS-SCHONBERG DISEASE*

Report of Case

L. S. N. PRASAD, K. P. SINHA AND D. K. SEN

Patna

Albers-Schonberg disease is a rare hereditary condition. It has been known by various names since its first clinical description in the medical literature in 1904 by Albers-Schonberg. Later in 1921, Laurell and Wallgren described this condition under a different name of “osteosclerosis iragilis generalisata”, which was subsequently modified to “congenital osteosclerosis”. It was Karshner (1926) who suggested the term “osteopetrosis” which signified that hardness and brittleness of the bones were both present simultaneously.

It is essentially a disease of childhood manifesting itself during the first year of life but in some cases the symptoms may not be apparent until adolescence or adult life. The exact type of inheritance in osteopetrosis is uncertain. Males and females are equally affected. It may be sporadic and in only about half the cases can a familial history be demonstrated. A study of the families suggests the operation of a rare recessive trait, though exceptionally, a family has been reported in which more than one generation has been involved. McPeak (1936) reported 8 cases occurring in three generations in one family. Harnapp (1937) discovered diffuse osteosclerosis in a father and in five of his seven children.

This disease is congenital in origin and starts in utero. The affected children may be born with hydrocephalus and often show symptoms suggestive of compression of the cranial nerves specially of the optic and the acoustic. They are markedly anaemic and seldom survive in spite of repeated transfusions of blood. The etiology remains obscure. There is no evidence that endocrine disturbances, dietary deficiency, toxae mia or faulty calcium and phosphorus metabolism have an etiological role. It is a true developmental disease which is manifested by the persistence of a primitive type of bone and seems to be due to an abnormal property of the parental germplasm.

Biopsy material of accepted instances of osteopetrosis suggests strongly that the disease is characterised by faulty resorption of the cartilagenous ground substances and of the newly-formed bone; faulty osteogenesis with the spongiosa with combinations of decreased, poor or metaphastic bone formation; and delayed or faulty remodelling of the
cortical and membrane bone. The long bones in a well-developed case are clubbed or hourglass-shaped with the most extensive clubbing at the ends, from which maximum growth in length normally occurs. Clubbing is usually symmetrical and equal but according to Pirie (1930), not necessarily so.

There is no specific treatment. All attempts to arrest the progress of the disease or to produce decalcification have failed. Inasmuch as osteopetrosis seems to be due to an abnormality of bone histogenesis, mere decalcification would be of no ultimate advantage. Fractures should be treated as in other cases. As consanguinity seems to be a factor in the occurrence of some of these cases, inter-marriages are inadvisable.

The following is a brief account of a case of osteopetrosis admitted to the Hospital for Children, Patna Medical College, for the treatment of extreme pallor and anaemia. This is the second case reported from this department (Prasad 1957).

**REPORT OF A CASE**

Baby Munna, aged 3 months, a Muslim male child, was admitted on 20.3.66 with the complaints of convulsions since the age of 20 days, frequent loose motions with blood and mucus since the age of one month, marked pallor and weakness, occasional episodes of low grade fever and failure to thrive. This was the first child of the family, born after full-term normal delivery. There was no history of abortion or consanguinity. At age of 3 weeks, the child started getting repeated attacks of convulsions throughout the day and night, 10-15 times in 24 hours. After 5 days, the severity and frequency of attacks diminished, but the convulsions did not stop. The child could never take to the breast even though there was no anatomical abnormality of the nipple nor any cleft lip or cleft palate. Pallor and weakness were of a progressive nature. He was given two whole blood transfusions of 100 cc each.

Examination showed an irritable child, with the limbs hypertonic. He could see and focus vision on the objects around. There was extreme pallor and the gums were markedly hypertrophied with a high arched palate. Patchy pigmentation was seen over the legs and abdomen indicating old hemorrhages under the skin. The upper eyelids appeared boggy and no lymphadenopathy was detected. The important measurements were, length 22", weight 10 lbs, and circumference of head 14.5". The skin was somewhat rough and a soft systolic bruit was heard all over the precordium, probably haemic. The lungs were clinically normal. The spleen was enlarged to 8 cm. below the costal margin, the liver was 4 cm., both being firm.

**Investigation.** The total R.B.C. count was 1,430,000/cmm, total W.B.C. count 5,320/cmm with polymorphs 27%, lymphocytes 35%, eosionphils 3%, monocytes 3% late normoblasts 25%, intermediate normoblasts 3% and myelocytes 4%. The hemoglobin value was 6.75 G.% and the platelet count 92,000/cmm. The fragility test was normal and Coomb's test was negative. The Wasserman reaction of the mother was positive. Tests for abnormal hemoglobins proved negative, serum iron was found to be