

OSTEPETROSIS - A CASE REPORT

S.M.S.B. TABIB¹, M.A. HALIM², M.N. ISLAM³

Introduction :

Osteopetrosis is a genetic disorder of bone¹. It is also known as marble bone disease, Osteosclerosis fragilis; Osteitis condensans generalisata and Albergs Schonberg disease. This rare metabolic bone disorder is characterized by dense bone which was first described by Albergs Schonberg in 1904 and Karshner in 1926². Several forms of osteopetrosis have been described with overlapping spectra of clinical and roentgenographic features. Among these only osteopetrosis of infantile variety with precocious manifestations and osteopetrosis with milder benign disease of childhood and adolescence has been clearly defined. The two forms appear differently both clinically and genetically³. Intermediate forms occur and include a type of osteopetrosis with renal tubular acidosis and cerebral calcification⁴.

This case presented to us with gradual increasing mass in the abdomen, low grade fever, respiratory tract infection and enlargement of head. Though osteopetrosis is not unknown in the country, this case is presented here with review of literatures to develop awareness about the presence of this rare disease in children in our country.

Case Report :

A 2 year old male child was admitted to the department of paediatrics, IPGMR with irregular low grade fever, recurrent respiratory tract infection, gradually increasing mass in abdomen and enlargement of head for last six months. He was born normally and antenatal and natal history was normal. There was no history of consanguinity of marriage. Other two sibs were active and alive showing no stigmata of the disease.

On examination he was restless, fretful, moderately pale with poor nutritional status. His weight was 5 Kg, which was 45% of 50th centile for age with a length of 58cm corresponding to 73% of 50th centile and head circumference 47cm.

Fontanelle was widely open and eye balls were slightly protruding with normal fundi. Liver was enlarged 3 cm from right costal margin, smooth, firm nontender. Spleen was enlarged 12 cm, smooth & firm. (Fig 1 & 2).

1. Dr. S.M. Shahnawaz Bin Tabib, FCPS, Asstt. Professor of Paediatrics
2. Dr. M.A. Halim FCPS, Associate Professor of Paediatrics
3. Prof. M.N. Islam FCPS; FRCP, Professor and Head of the Deptt. of Paediatrics
Institute of Post Graduate Medicine and Research, Dhaka.



Fig 1
Two years boy with osteopetrosis



Fig. 2
Patient of Osteopetrosis with marked
hepatosplenomegaly

Auscultation of lung revealed vesicular breath sound with scattered crepitation all over the lung field.

Investigations :

His haemoglobin was 58% Total WBC 16000/cumm with 48% Neutrophil, 46% lymphocyte, 04% Monocyte and 02% Eosinophil. Peripheral blood film was suggestive of leuco erythroblastic picture.

X-ray skull showed sclerosis of cranial bones specially at the bases. There was increased density of long bones with loss of medullary cavity with periosteal reaction. Sclerosis of the vertebral bodies specially end plates were noted 'FIG-3'.

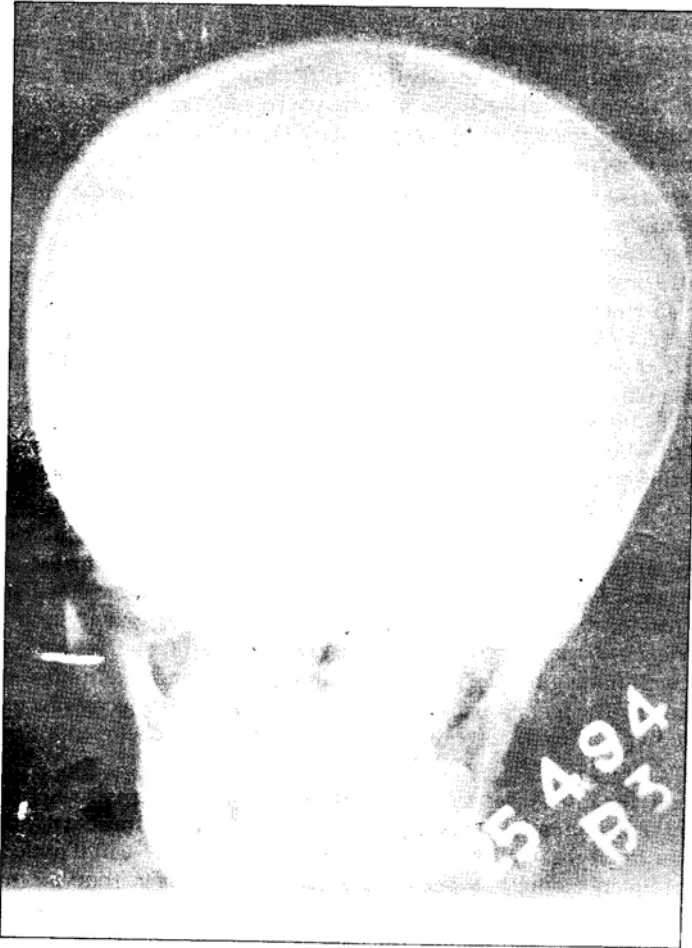


Fig. 3 Skeletal changes of the case (Increased density of bone)

Ultrasonography of head showed dilated ventricles and ultrasonography of abdomen revealed mild hepatomegaly with gross splenomegaly.

He was diagnosed as a case of Osteopetrosis (dominant type) with 3rd degree malnutrition. He was treated with antibiotic cotrimoxazole and full energy feeding. Though his nutritional status was improved but other condition remained static and he was discharged after 18 days with advice to attend follow up clinic in paediatric outpatient department.

Discussion :

Osteopetrosis is a rare metabolic bone disorder manifested clinically by tendency to fracture, optic atrophy, cranial nerve palsies, anaemia and thrombocytopenia with hepatosplenomegaly, pathologically by brittle bones. There is no age and sex difference⁵. There is a persistence of primary calcified cartilaginous matrix with apparent failure of resorption by osteoclasts. Remodelling of bone is defective and cortical compact bone is poorly differentiated as the marrow space fails to form.⁶

The mode of inheritance of osteopetrosis has appeared confused many years. The complex nature of inheritance is seen in the classification of Schinz in 1944 and Hanhart in 1948. The genetic problems were well reviewed by Johnston et al in 1968 and they also described two mode of inheritance - a autosomal dominant form with a rather benign course and a rare autosomal recessive form which runs a malignant course with marked anaemia, myeloid metaplasia, infection and early death³. There is no history of consanguinity of our patient and other sibs are normal. So this case may be dominant form due to spontaneous mutation. Anaemia, failure to thrive, recurrent infections, protruding eye ball and progressive hepatosplenomegaly were marked in this case which is consistent with osteopetrosis. Anaemia in osteopetrosis is leucoerythroblastic in type. This patient also had leucoerythroblastic anaemia. Radiological findings are quite consistent with the findings of Silence and Robert Berkow^{2,4}.

Radiological findings showed generalised increased in bone density, endobones form in vertebra, Pelvis and tubular bones; the skull became thickened specially at the base, loss of demarkation of cortex and the medullary cavity and the spine showing "rigger Jersey" appearance. This patient has dilated ventricles which is consistent with finding by Bard P.A. et al that Osteopetrosis is an unusual cause of hydrocephalus⁷. The management of osteopetrosis is aimed at decreasing or arresting progressive hyperostosis, correcting anaemia and thrombocytopenia and treating infection promptly and vigorously⁴.

Treatment for the severe malignant form of osteopetrosis includes blood transfusion for anaemia, antibiotics for infection and standard orthopaedic treatment for fractures. Optic nerve decompression may save sight if optic atrophy is developing. Splenectomy has been performed on the basis that the haemolytic process is due to hypersplenism. Corticosteroids have controlled the anaemia and thrombocytopenia in some patients but have been of no obvious benefit in other⁶. However high dose IV. methyl prednisolon in the treatment of recessive osteopetrosis

is highly effective⁸. A low calcium diet if necessary supplemented with cellulose phosphate to inhibit calcium absorption has led to demineralization of bone and prevented progression of the disease. High dose calcitriol promotes bone resorption leading to clinical improvement⁹. Bone marrow transplantation with appropriately HLA matched donor marrow may be dramatically beneficial in some patients^{10,11}. Until the basic pathogenesis of the disorder is more fully understood, some of all these various forms of treatment may reasonably be adopted¹². Usually prognosis is guarded and death in the first few months or years is not uncommon.

References :

1. Cockburn Forrester, Hutchison James H. Albers-Schonberg disease (Osteopetrosis) Practical paediatric problems, 6th ed. 1986.
2. Robert Berkow. The Marck Manual of Diagnosis and therapy. Osteopetrosis 14th ed. MSD, 1982 S 1234-38.
3. YU IS et al. Osteopetrosis. Arch Dis Child 1971; 46:257.
4. Sillence David, O. Genetic Skeletal dysplasias Nelson - Text Book of Paediatrics 14th ed. W. B. Saunders Company, 1992 P-1743.
5. Marshall A Schorin. Bleeding and Purpura Bed side pediatrics. Diagnostic evaluation of the child. Ea Mohsen Ziai 1983, Little Brown and Company, Boston P. 542.
6. Campbell A.G.M, Mcintosh Neil, Albers. Schonberg Disease (Osteopetrosis, Marble Bone disease), Forfar and Arneils Text Book of Paediatrics 4th ed. ELBS, 1993, p-1656-57.
7. Baird PA, Robinson GC, Hardwich DF et al. Congenital osteopetrosis : unusual cause of hydrocephalus. Can Med. Assoc. Journal 1986 : 98:382-385.
8. Ozsoylu S, High dose I.V. Methyl Prednisolon in the treatment of osteopetrosis (A.R.) Arch Dis Child 1987; 62 (2); 214-5.
9. Key L et. al Treatment of congenital osteopetrosis with high dose calcitriol. New Eng. Journal of Medicine 1984. 310:409-415.
10. Coccia PF, Krivit W, Cervenka J et al, Successful bone marrow transplantation for infantile malignant osteopetrosis. N Engl J Med. 302:701. 1980.
11. Sieff C.A. et al. Bone marrow transplantation in osteopetrosis, Lancet, 1983, 1:437.
12. Moe PJ, Skjaeveland A. Therapeutic studies in osteopetrosis. Acta Paediatrica Scandinavia, 1969 58:593-600.

Electricity cannot flow through rubber, mica, procelain, oil, and dry wood, which are called non-conductors of electricity.

Turtles and tortoises have no teeth but horny sheaths to the edges of the jaws.