Treatment Dilemma in Osteopetrorickets

A 1½-year-old boy who is followed up in our Nutrition and Growth Clinic was diagnosed to have co-existent osteopetrosis and rickets. At the time of diagnosis, he was seven months old and had anthropometric parameters below the 3rd centile of the NCHS reference standards except head circumference, which was between the 25th and the 50th centile. Clinical examination revealed pallor, hepatosplenomegaly, early optic atrophy and features of rickets like caput quadratum, wide open anterior and posterior, fontanels, Harrison’s sulcus, rachitic rosaries and widening of wrists and ankles. Laboratory data showed hemoglobin 7.5 g/dL, total leukocyte count of 11,200/cumm, polymorphs 52%, lymphocytes 30% and eosinophils 18%, platelet count 1 lakh/cumm and leuko-erythroblastic blood picture with mild thrombocytopenia. He had normal thyroid function and normal karyotyping. The serum calcium (7.2 mg/dL) and phosphorus (1.4 mg/dL) levels were low and alkaline phosphatase (2005 IU/L) was markedly raised. Serum sodium, potassium, bicarbonate and urine pH were normal and associated renal tubular acidosis was excluded. Urine was negative for mucopolysaccharides and aminoaciduria. The liver and renal function tests were within the normal range. Skeletal survey revealed features of osteopetrosis, rickets and pathological fractures with ‘bone in bone’ appearance, increase in medullary density, defective metaphyseal remodeling with fraying and early cupping and widening of ‘physis’, the growth plate (Fig.1). USS showed hepatosplenomegaly and CT scan head showed osteosclerosis and diffuse brain atrophy.

Osteopetrorickets is a rare association of osteopetrosis with rickets(1,2). In osteopetrosis, there is excess calcium reserve and hypophosphatemia(3), but when it is complicated by rickets there is an insufficient Ca:P product to mineralize the growing bone. In coexistent osteopetrosis and rickets, 99% of the calcium is shut off in the bone and is not

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Fig. 1. X-ray of left forearm and hand showing osteosclerosis, ‘bone in bone’ appearance of shaft, fracture both bones, fraying and early cupping of metaphysis and widening of ‘physis’, the growth plate and absence of carpal bones.
available as a source of these minerals and the calcium phosphorus product will become <40, thus insufficient for mineralisation. In such cases, there is chance for hypocalcemic tetany(3). This association is considered a paradox in the midst of plenty(4). Low calcium and high phosphate diet prescribed in osteopetrosis may further decrease the availability of calcium and in rickets liberal calcium is required. Low dose steroid therapy, which is found effective in treating and preventing the hematological problems in osteopetrosis may blunt the intestinal response to maximal vitamin D stimulation as well(4). The general view is to treat rickets in this co-morbid situation as it will improve well being, decrease irritability and decrease infections. This may also help in preparing the patient for future bone marrow transplantation which is the ultimate therapy in osteopetrosis.

Initially, before complete work up, the child was on low calcium, high phosphorus diet and phosphate supplement in the form of Joulie solution. After the confirmation of associated rickets, he was given 2 loading doses of vitamin D3, 6 lakhs units at 3 weeks interval, but there was no line of calcification. Later, he was put on liberal calcium and phosphorus diet with extra calcium carbonate supplements up to 200 mg/kg per day, Joulie solution 4 g/day in 5 doses and 2 mcg/day of 1,25 dihydroxy cholecalciferol. He was also on 1 mg/kg oral prednisolone on an alternate day basis. After three months, serum calcium became 9.4 mg/dL and phosphorus 3.3 mg/dL and serum alkaline phosphatase 677 IU/L and X-ray showed features of healing. The general well being of the child has improved. Growth is continuing in the 3rd centile and he is able to stand with support at 18 months of age. Tooth eruption was delayed up to 18 months.

K. E. Elizabeth,
Gibby Koshy,
Department of Pediatrics,
SAT Hospital,
Government Medical College,
Trivandrum 695 011, Kerala,
India.
E-mail: elizake@hotmail.com

REFERENCES