International Journal of Science and Healthcare Research
Vol. 8; Issue: 1; Jan.-March 2023
Website: ijshr.com
Case Report *ISSN: 2455-7587*

A Rare Case of Polyostotic Fibrous Dysplasia

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DOI: https://doi.org/10.52403/ijshr.20230113

ABSTRACT

This case repost discusses clinical and biochemical features of polyostotic fibrous dysplasia, a rare entity which leads to significant morbidity and multisystem manifestation in children. The diagnosis ,differentials, treatment and complications are discussed. The role of FGF 23 in the pathogenesis of the disease is briefly discussed. This case description would improve the understanding of the clinical presentation as well as the diagnosis and treatment of fibrous dysplasia.

Keywords: polyostotic fibrous dysplasia, FGF 23, hypophosphatemia

CASE DESCRIPTION

This case report describes the clinical and biochemical features of fibrous dysplasia in a 11 years old male child who came to the OPD with a chief complaint of Recurrent fractures for last 7 years, Bending of lower limbs for 4 years, bending of arms for 2 years. The child was non ambulatory for last four years. There was no history of urinary complaints, visual or hearing disturbance, drug intake, brittle teeth, fever, rash, alopecia, trauma, weight loss/ loss of appetite, blood transfusions .The child had Sustained 3 fractures till date: first at 5 year of age, Second at 7 year of age, and third at 8 year of age.

The child was born out of a non consanguineous marriage and antenatal and birth history was uneventful. The child had pallor and clubbing, rachitic rosary, Harrison sulcus, deformed bilateral humerus (forward bending), bilateral wrist joints twisted, deformed lower limbs , bilateral femur twisting along with kyphosis and scoliosis. The weight of the child was at -4.04Z and height was normal at -2.01Z. Laboratory investigations showed normal complete blood counts and normal urine examination, serum calcium (Ca++) level of 8.7mg/dl (8.8-10.8mg/dl) low serum phosphate of 1.7mg/dl (4-7 mg/dl), while alkaline phosphatase was elevated at 1696 IU/L(44-147IU/L).Serum electrolytes, liver and renal functions were within normal limits.

Patient had normal blood gas analysis. The parathyroid hormone (PTH) level was normal 51.6 pg/ml (7-53 pg/ml) ,25 hydroxy vitamin D3 was normal 40.7ng/l(20-40ng/ml). 24-hour urine examination showed marked phosphaturia, with phosphate of 26.6 mg/dl (4.5-6.5 mg/dl) and Fractional excretion of phosphate 48%.The urine electrolytes, and calcium creatinine ratio was normal. The TMP GFR was normal 1.11(1.15-2.44).

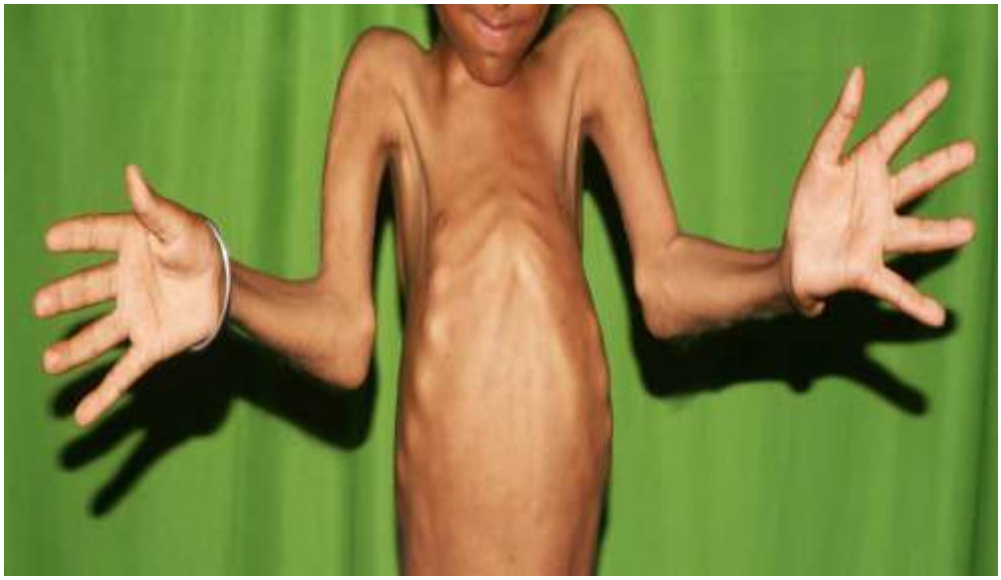

Figure 1 The index case has rachitic rosary and bending of both humeri.

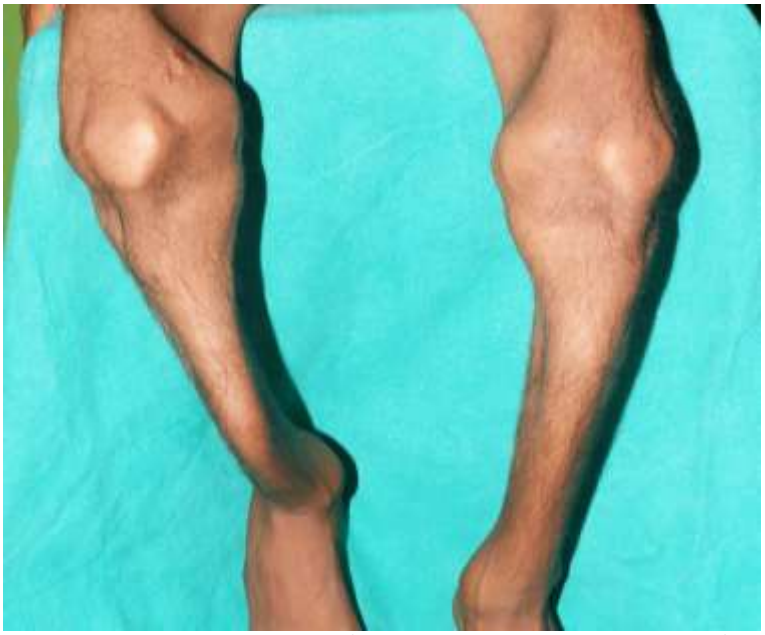

Figure 2 The tibia on both sides are bent forward and the lower extremities are deformed.

Figure 3 Bending and expansion of distal end of arms and wrist joint is seen. Clubbing is seen.

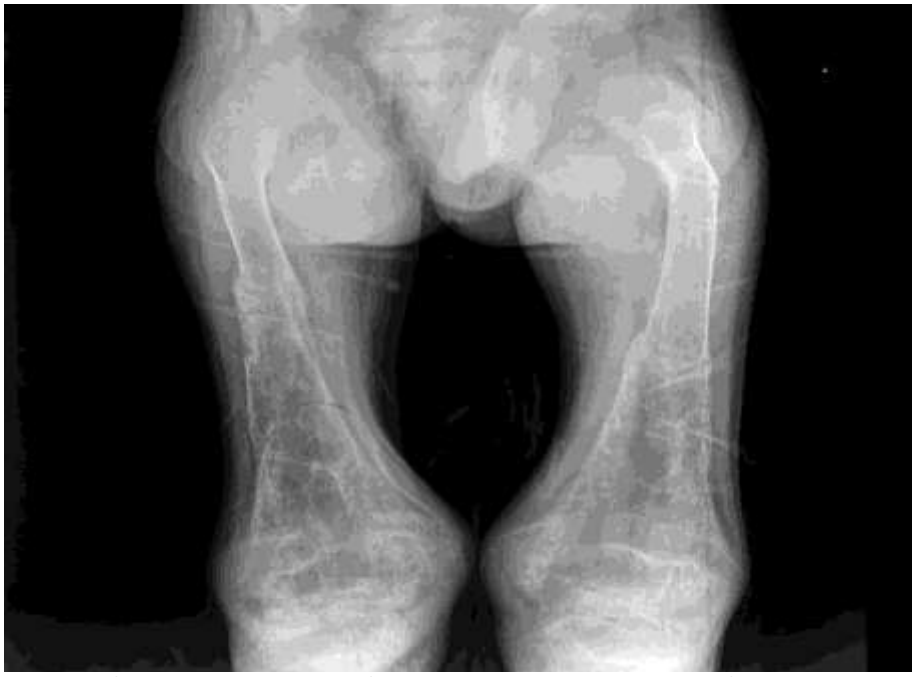

Figure 4: Multiple fractures in various stages of healing are noted, there is expansion of disphysis and metaphyses.

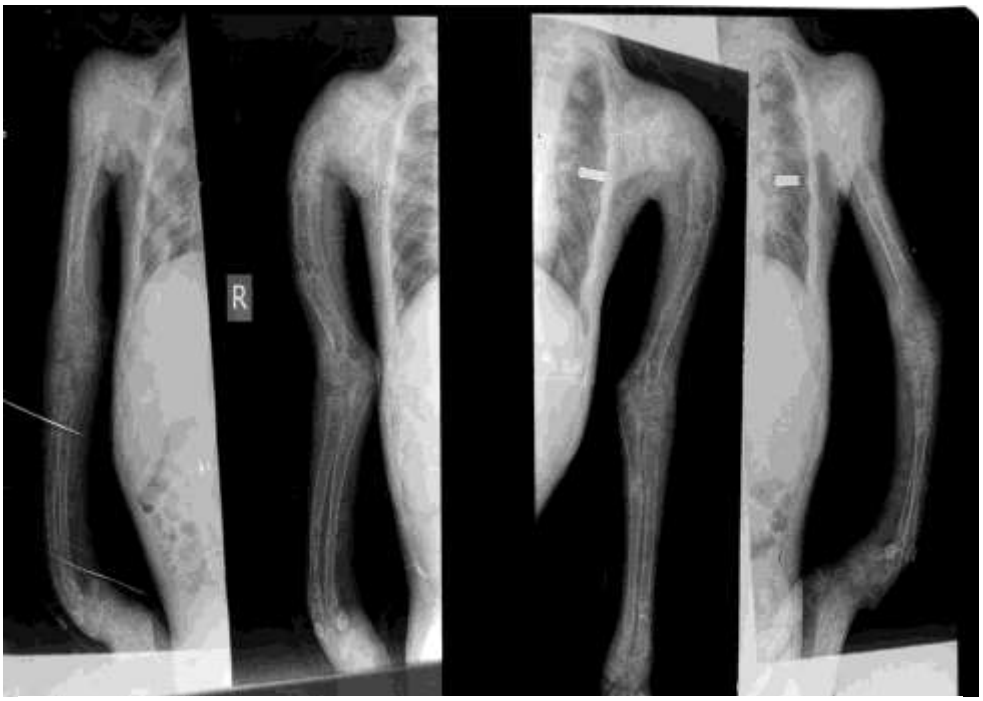

Figure 5: Multiple fractures on both arms and distal twisting of forearm is seen.The bone density is low.

DISCUSSION

In the index case differential diagnosis of polyostotic fibrous dysplasia, hypophosphatemic rickets and osteogenesis imperfecta were considered.

However in view of disphyseal involvement in all bones on X ray, no family history, normal dental examination and very severe deformity of limbs, hypophosphatemic rickets was less likely. There were no hyperlaxity of joints, no history of bruising and normal hearing and eye examination, so osteogenesis imperfecta was considered less likely.

Polyostotic fibrous dysplasia is defined as the presence of fibrous dysplasia in more than one skeletal site without extra-skeletal manifestations(1). Fibrous dysplasia is postulated to occur as a result of a developmental failure in the remodeling of

primitive bone to mature lamellar bone and a failure of the bone to realign in response to mechanical stress. In Fibrous dysplasia Total FGF23(fibroblast growth factor23) levels (a combination of intact FGF23{ifgf23} and circulating FGF23{cfgf23}) are elevated, there was a proportionally greater elevation in cFGF23 levels relative to iFGF23(2). FGF23 reduces serum phosphate levels by three mechanisms:(a) Directly by down-regulation of the NaPi 2a and 2c transporters in the proximal tubule, leading to reduced renal phosphate reabsorption at the level of the kidney,(b) Directly by down-regulation of renal 1α-hydroxylase, which decreases production of active 1,25-dihydroxyvitamin D and as a result decreases intestinal phosphate absorption, and (c) Indirectly by the compensatory increase in PTH(2).However due to cost constraints this test could not be conducted in index patient.

The index case had hypophosphatemia, fractures and significant motor disability.

Phosphate supplementation was started @40 mg/kg/day in 3 divided doses. The dose is to be titrated to maintain serum phosphate at the lower end or just below the normal laboratory reference range for phosphate. In case the vitamin D is low (normal in this case), it is treated with an active metabolite or analogue of vitamin D calcidiol or alphacalcidiol 15-60 ng/kg/day in two divided doses. Calcium supplementation was added @100 mg/kg/day. (3)

Bisphosphonates can be started in severe bone pain defined as VAS score of >3/10, after ensuring that the patient is normo calcemic, hypophosphatemia is corrected and has adequate levels of 25(OH)Vitamin D(4). After excluding endocrine abnormalities, the child was referred to orthopedic surgeon for the treatment of fractures, limb length deformity requires assessment for need of orthotics and prophylactic surgery(5). Long term risk to the child involves monitoring for Fractures, GI manifestations such as polyps and hepatobiliary neoplasms, increased risk of malignancies in mutation bearing and high turnover tissues, facial deformity- hearing and vision disturbances.(3)

Declaration by Authors

Acknowledgement: None

Source of Funding: None

Conflict of Interest: The authors declare no conflict of interest.

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How to cite this article: Sugha Varuna, Bharti Sapna. A rare case of Polyostotic Fibrous Dysplasia. *International Journal of Science & Healthcare Research.* 2023; 8(1): 109-112. DOI: *https://doi.org/10.52403/ijshr.20230113*

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