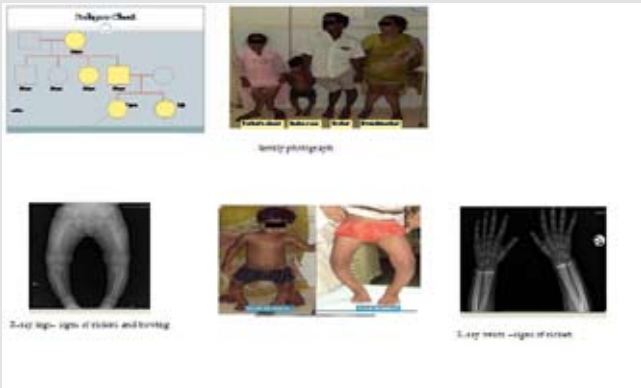
SPOT DIAGNOSIS (IMAGE GALLERY)



FAMILIAL RICKETS

P V Nigwekar, D P Mohapatra.
Department of Pediatrics, Rural Medical College, Loni, Ahmednagar, India

Address for Correspondence: Dr D P Mohapatra,204 Valmik, Nyati Garden, Mahammadwadi, Pune-411060, India. Email: mohapatradp@hotmail.com

Three years old girl, first issue of non-consanguineous marriage presented with progressive bowing of legs since one year of age when she started walking. There was poor gain of height and delayed dentition. Family history revealed that her younger sister of 10 months age had mild bowing of legs. Patient's father, father`s sister and grandmother also had similar complaints since early childhood. She was earlier given two doses of Inj vit D (6 lakh units) each at 4 weeks interval without any response. On examination, weight was 10.8 kg, (Less than 3rd percentile), height was 83 cms, (Less than 3rd percentile), upper segment: lower segment ratio was 1.4:1(increased), intercondylar distance was 12.5 cms (normal Less than 7 cms). Bowing of legs and widening of both wrists and rickety rosary was present. Laboratory investigations revealed normal hemogram, serum phosphorus of 2 mg/ dl (Normal = 4- 6.5mg/ dl), serum calcium of 9.0 mg, dl (Normal=8.4-10.4mg/ dl), serum alkaline phosphatase of 132 IU/ l (Normal =15-110 IU/ l). Renal function tests and blood gases were within normal limits. Serum vitamin D level and urinary excretion of phosphorus could not be estimated due to lack of facilities in the local laboratory. Ultrasound abdomen was normal. X-ray of wrist joints showed retarded bone age with signs of rickets. X-ray legs showed bowing of legs with signs of rickets. Child was put on oral Joulie solution 5 ml five times a day to provide 760 mgs of phosphate. Calcitriol 0.25 mcg daily was also added. Child showed remarkable improvement over period of time and bowing of legs decreased and child started to gain height.

What is the diagnosis?

Hypophosphatemic rickets. Familial hypophosphatemic rickets is a rare vitamin D resistant rickets. It is commonly inherited as an X-web addressed dominant genetic disorder due to defect in a gene called PHEX gene with a prevalence of 1:20,000. (1) The basic defect is increased urinary excretion of phosphorus leading to hypophosphatemia. Signs of rickets and deformities are more marked in lower limbs than chest and upper limbs. Girls generally have less severe disease than boys, probably due to the X-web addressed inheritance. It is characterized by very low serum phosphate levels, high alkaline phosphatase level and resistance to treatment with vitamin D. (2) An outstanding feature of familial hypophosphatemic rickets is short stature. (3) The prognosis for a normal lifespan and normal health is good. Patients respond well to a combination of oral phosphorus and 1,25-D (calcitriol). The daily need for phosphorus supplementation is 1–3 g of elemental phosphorus divided into 4–5 doses. (4) The therapeutic response can be monitored by serum alkaline phosphatase level which is a better indicator than serum phosphorus level. Some children may develop hypertension due to persistent hyperparathyroidism. (5)

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