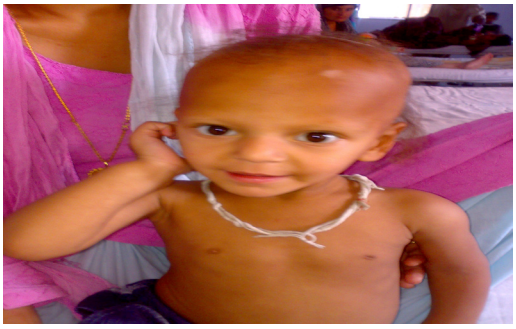
SPOT DIAGNOSIS (IMAGE GALLERY)



RICKETS (VITAMIN D DEPENDENT RICKETS TYPE II)

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A two years and eight months old female child presented to us with complaints of progressive falling of hair after neonatal period and delay and difficulty in walking. She also had history of excessive sweating mostly on forehead, delayed dentition and delayed milestones. She was second in birth order born of non-consanguineous parents. Family history of similar ailments was negative. On examination, she had almost alopecia totalis (Fig.1). She had florid rickets in the form of widened wrists, widened ankles, open anterior fontanelle and mild frontal bossing along with slight bowing of legs and waddling gait. Occipitofrontal circumference (OFC) was 48 cms. She was short statured with a height of 80 cms (Less than 5th centile). Her biochemical parameters were in favour of rickets showing low levels of serum calcium (8.6mg/dl) and moderately low levels of phosphorus (2.03 mg/dl), alkaline phosphatase was markedly raised (1764.8 IU/ l). Serum parathormone (PTH) levels were also raised (354.9 pg/ml) indicating secondary hyperparathyroidism. Levels of 1,25-dihyroxyvitamin D (1,25-D) were also markedly raised [1053.6 pg/ ml (normal 19.6-54.3 pg/ ml)] which strongly points to vitamin D dependent type II rickets. Renal and hepatic function parameters were within normal limits. Her roentgenographic findings revealed metaphyseal flaying, fraying and cupping of proximal and distal tibia, distal femur, distal radius and ulna. Before reporting to us the child had received treatment in the form of injectable vitamin D for few months with no improvement in the condition of child.

What is the diagnosis?

Vitamin D dependent rickets type II also known as Vitamin D resistant rickets Type II is a rare genetic disorder which presents as rickets not responsive to high doses of vitamin D. It is characterized by presence of elevated serum levels of 1,25 dihydroxyvitamin D (1,25-D) and in many cases alopecia. (1) It is a rare autosomal recessive disease resulting from end organ resistance to 1, 25-D caused by defect in the vitamin D receptor (VDR) gene (2). Two siblings with vitamin D dependent type II rickets were reported for the first time from India in 1990 (3). Some patients, especially those without alopecia, respond to extremely high doses of vitamin D2, 25-D, or 1,25-D. This response is due to a partially functional vitamin D receptor. All patients with this disorder should receive a 3 to 6 month trial of high-dose vitamin D and oral calcium. To start with dose of 1,25-D should be 2 µg/ day but some patients require doses as high as 50–60 µg/ day. (4) Calcium doses range from 1,000–3,000 mg/ day. Patients who do not respond to high-dose vitamin D may be treated with long-term intravenous calcium, with possible transition to very high-dose oral calcium supplements. Treatment of patients who do not respond to vitamin D is difficult.

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E-published: April 2011 . **Art**#22