ORIGINAL RESEARCH PAPER

Paediatrics

A RARE CASE OF VITAMIN D DEPENDENT RICKETS TYPE-2A PRESENTED AS ALOPECIA TOTALIS

KEY WORDS: alopecia,end organ resistance,vitamin D, 1,25[OH]2 vitamin D3

M.V.Sreevani yadav Post graduate,Department of paediatrics,Kurnool medical college,kurnool,Andhrapradesh,India

B.Himabindu* Post graduate,Department of paediatrics,Kurnool medical college,kurnool,Andhrapradesh,India*Corresponding Author

DR.B.Vijay anand babu professor and HOD,Department of paediatrics,Kurnool medical college,kurnool,Andhrapradesh,India

DR.S.Srilatha Associate professor,Department of paediatrics,Kurnool medical college,kurnool,Andhrapradesh,India

ABSTRACT

- Rickets is a disease of growing bone caused by unmineralized matrix at the growth plates in children only before fusion of the epiphyses.
- There are many causes of rickets, including vitamin D disorders, calcium deficiency, phosphorus deficiency, and distal renal tubular acidosis
- VDDR-2A presents as refractory rickets and growth retardation presenting within first year of life and is frequently associated with alopecia totalis

INTRODUCTION

- Vitamin D dependent Rickets Type 2A is an Autosomal Recessive disorder due to mutations in the gene encoding the vitamin D receptor, preventing a normal physiological response to 1,25-Dihydroxy cholecalciferol resulting in extremely elevated levels of 1,25-Dihydroxy cholecalciferol.
- Most patients present during infancy, although rickets in less severely affected patients might not be diagnosed until adulthood. Less severe disease is associated with a partially functional vitamin D receptor.
- Approximately 50 to 70% of children have alopecia, which tend to be associated with a more severe form of disease and can range from Alopecia areata to alopecia totalis.

CASE REPORT

- A 22 months old female child presented with complaints of progressive Alopecia since 3 months of age which started as loss of scalp hair followed by loss of eyebrows followed by loss of hair in the entire body.
- Associated with inability to walk .
- Associated with recurrent respiratory infections in the past
- History of Sibling death with similar complaints present.

ON EXAMINATION

- Vitals were stable
- Short stature present
- Delayed dentition present
- Mild frontal bossing present
- Rickety rosary present
- Pot belly abdomen present
- Scoliosis present
- Bow legs present
- Widening of wrist present
- Alopecia totalis present
- No gross abnormalities found in CVS,RS,ABDOMEN, GENITOURINARY and CNS.

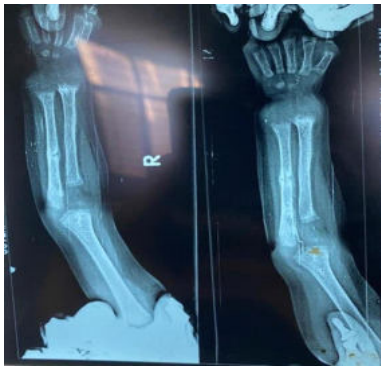
BIOCHEMICAL INVESTIGATIONS

- Serum calcium, total-normal(9.2mg/dl)
- Serum calcium, ionised-normal(1.20mmol/l)
- Serum Phosphate –decreased(2.7mg/dl)
- Serum PTH-grossly Elevated(695pg/ml)
- Serum 25 OH Vitamin D-Normal(67nmol/l)
- Serum 1,25 OH Vitamin D-Grossly elevated(170pg/ml)
- Serum Alkaline phosphatase-Grossly elevated(2200 U/l)
- Urinary calcium decreased(4mg/dl)
- Urinary phosphate levels Increased(10mg/dl)

RADIOLOGICAL INVESTIGATIONS

- Plain AP and lateral radiographs of right upper limb and B/L lower limbs showing:

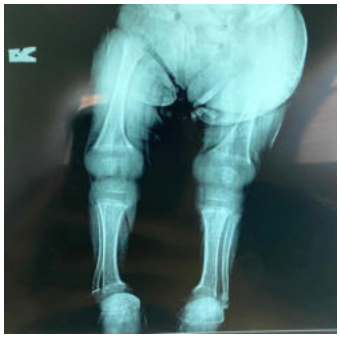
diffuse osteopenia with widened epiphysis of distal radius and ulna

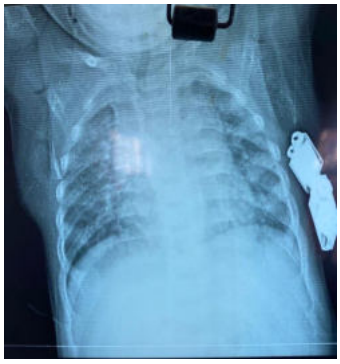


- B/L distal femur with :

splaying and fraying ,insufficiency fracture with adjacent callus formation noted in mid shaft of right ulna.

- Plain X-ray AP view of chest showing beaded appearance of left 2nd to 5th costochondral junctions.





TREATMENT

- High doses of vitamin D3(1,25 dihydroxy cholecalciferol) started as 2 mcg per day
- High dose of oral calcium 2000 mg per day

FOLLOW UP

- Child completed one month of high dose of VitaminD3 and Oral Calcium and is on follow up once in every 2 weeks without any significant improvement till now.
- Advised to continue therapy up to 3 to 6 months to expect clinical,biochemical and radiological improvement.

DISCUSSION

- Child with partially functioning vitamin D receptor will have less severe disease and have some degree of response to high doses of vitamin D3 and calcium therapy.
- Child with totally nonfunctioning vitamin D receptor will have severe form of disease and treatment of such cases will be difficult and have grave prognosis

CONCLUSION

- VDDR is a autosomal recessive disorder with history,physical examination findings and investigations support VDDR TYPE-2A caused by mutations in gene encoding the vitamin D receptor,hence preventing the normal physiological response to 1,25-D hence needs extremely high doses of vitaminD2[25D or 1,25-D]

CONFLICTS OF INTEREST: NO

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