International Clinical and Medical Case Reports Journal
Case Report (ISSN: 2832-5788)

A Case Report of Vitamin D Dependent Rickets Type1

Rupal Kashiyani*, Mehul M. Jadav

Second Year Resident, Department of Paediatrics, B.J. Medical College, Ahmedabad

Citation: *Rupal Kashiyani*, Mehul M. Jadav. A Case Report of Vitamin D Dependent Rickets Type1. Int Clinc Med Case Rep Jour. 2025;4(8):1-3.*

Received Date: 01 August 2025; **Accepted Date:** 03 August 2025; **Published Date:** 04 August 2025

***Corresponding author:** Rupal Kashiyani, Second Year Resident, Department of Paediatrics, B.J. Medical College, Ahmedabad

Copyright: © Rupal Kashiyani, Open Access 2025. This article, published in Int Clinc Med Case Rep Jour (ICMCRJ) (Attribution 4.0 International), as described by http://creativecommons.org/licenses/by/4.0/

ABSTRACT

Vitamin D dependent rickets type 1 is rare condition, incidence being 1 in 2,50,000 worldwide. VDDR has 3 subtypes amongst which VDDR type 1 is most common. It is an autosomal recessive condition results from mutation in cyp27B1 gene that codes for 1 α hydroxylase, rate limiting enzyme that converts vitamin D into its active form, calcitriol in kidney.

25(OH) vit D (inactive) ----------> 1,25(OH)2vit D (active)

1 α hydroxylase (rate limiting step)

In this case, 3 years old male child presented with c/o breathing difficulty for 1 month. Patient has consulted multiple doctors in the past for failure to thrive, delayed milestones and multiple deformities, for which he was prescribed vitamin D sachets, but there was no improvement in patient's condition.

On general examination, patient had marked subcostal indrawing, nasal flaring and classical features of rickets such as widening of wrists and ankles, anterior bowing of tibia and femur, genu varus, rachitic rosary, Harrison's groove, frontal bossing, dental carries.

In view of history, examination and X-ray finding, Patient was investigated for rickets. Based on laboratory parameters such as active and inactive form of vitamin D, PTH, Calcium and phosphate level, patient was diagnosed as vitamin D dependent rickets type 1.

Patient initially was given high flow nasal O2 support and gradually weaned off in 3-4 days. Patient was given active vitamin D (CALCITRIOL) in high doses, oral calcium, phosphate sachets, zinc, vitamin c supplements on which patient's condition improved. Proper physiotherapy, dietary advice, course and prognosis of the disease explained to parents and discharged the patient on same oral supplements.

INTRODUCTION

Rickets is a disease of growing bone, characterized by unmineralised bone matrix at growth plates before fusion of epiphyses leading to softening and weakening of the bone leading to various bone deformities.

The disease usually manifest during 1st 2 years of life when child begun to crawl/stand/walk and clinical features similar to vitamin D deficiency rickets, but patient not responding to physiological dose of vitamin D, so called pseudo vitamin D deficiency rickets. Larger doses of vitamin D ranging between 10000-40000 IU daily is necessary as treatment, which needs to be continued lifelong.

CASE REPORT

A 3-year-old male child, referred to pediatrics department, civil hospital Ahmedabad with c/o breathing difficulty for 1 month. On examination, RR=50/min, HR=120/min, marked subcostal recession and nasal flaring was present. On general examination, patient has classical features of rickets such as widening of wrists and ankles, deformities of forearm, anterior bowing of tibia and femur, genu varus, rachitic rosary, Harrison's groove, frontal bossing, and dental carries.

At the age of 1 year, parents noticed swelling at wrist, they consulted in private clinic where patient was stamped having rickets based on X-ray changes and oral supplements were given for 6-7 months. As there was no improvement seen and swelling was increasing in size with bending of legs, they reffered the child to us and on presentation, child was in respiratory distress, and classical features of rickets were noticed. In view of history, examination and X-ray findings, Patient was investigated for rickets in detail. Based on laboratory parameters, patient was diagnosed as vitamin D dependent rickets type 1.

INVESTIGATION

All the routine investigations- hemogram, renal function test, S. electrolytes, liver function test, urine routine-micro were within normal limits. USG abdomen+KUB, renal Doppler and 2D echo all came out to be normal.

S. calcium	7.88 mg/dl (low normal)
Inorganic phosphorus	2.36 mg/dl (normal)
Vitamin D (25-OH)	59.4 ng/ml (high)
Vitamin D (1,25-OH) (active form)	6.83ng/ml (very low)
S. Parathyroid hormone	1396 pg/ml (very high)
Alkaline phosphatase	2667 U/L (very high)

X-ray chest, wrist, joints, bones results:

Severe generalized osteoporosis with thinning of cortex and radiolucent epiphysgen were noticed in visualised bones. Widening, cupping and fraying of metaphysis observed in bilateral wrist joints. Bowing was present, involving bilateral radius and ulna. Pathological fractures seen through mid-shaft of either ulna. Zone of calcification noted in distal end of radius possibly due to healing and widening of anterior ends of ribs.

- Findings suggest changes of rickets.

Genetic workup for the condition has been advised to parents.

TREATMENT

Patient initially was given high flow nasal O2 support and gradually weaned off in 3-4 days. IV antibiotics given for underlying respiratory infection. Patient was given active vitamin D in high doses, oral calcium, phosphate sachets, zinc, vitamin c supplements after consultation with endocrinologist. Proper physiotherapy, dietary advice, course and prognosis of the disease explained to parents and discharged the patient on same oral supplements.

DISCUSSION

Vitamin D dependent rickets has 3 subtypes-VDDR 1A, VDDR 1B, VDDR 2; VDDR 1A being most common. Vitamin D dependent rickets type 1 is an autosomal recessive condition results from mutation in cyp27B1 gene

that codes for 1 α hydroxylase, rate limiting enzyme that converts vitamin D into its active form calcitriol in kidney.

25(OH) vit D (inactive) ----------> 1,25(OH)2vit D (active)

1 α hydroxylase (rate limiting step)

The active form of vitamin D, 1,25(OH)D3 plays an essential role in calcium and phosphate metabolism, bone growth and cellular differentiation. Deficiency of which leads to defective mineralization of bones, which leads to softening and weakening of bones and features of rickets like bowing of bones, widening, cupping and fraying of metaphysis, pathological fractures.

CONCLUSION

The main hormonal regulator of bone mineralization is vitamin D, deficiency of which leads to defective mineralization of growing bone and rickets. VDDR type 1 doesn't respond to normal doses of vitamin D supplements, so higher doses are required to maintain S. calcium level low normal and treatment needs to be continued lifelong. Patients present with recurrent respiratory infections due to muscle weakness and hypotonia.

REFERENCES

1. Nelson textbook of paediatrics 21th edition page no. 442-448
2. Feldman D, J Malloy P. Mutation in the vitamin D receptor and hereditary vitamin dependent rickets. 2014;3:510.