Diagnostic challenges of Vitamin D-Dependent Rickets Type 1a (VDDR1A) caused by CYP27B1 mutation in resource limited countries: a case series from three families

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INTRODUCTION

Vitamin D dependent rickets type 1A (VDDR1A) is an autosomal recessive condition due to inactivating mutation in CYP27B1 which inhibits 1-alpha-hydroxylase enzyme leading to defective conversion of 25-OH vitamin D to 1,25-(OH)2 vitamin D. (Figure-1)

Clinical and bone profile of VDDR1A mimics vitamin D deficient and others vitamin D dependent rickets. However, normal or raised 25-OH vitamin D in presence of low 1,25-(OH)2 vitamin D is diagnostic of VDDR1A.

In developing countries like Pakistan diagnosing VDDR1A is a challenge due to lack of free availability of 1,25-(OH)2D level and genetic testing.

AIM

To determine the clinical profile and challenges in diagnosis and management of vitamin D dependent rickets type 1A in developing countries.

METHODS

Retrospective review of all children with vitamin D dependent rickets type 1A due to CYP27B1 mutation over last one year in a tertiary care hospital.

CONCLUSION

We should have a high index of suspicion of VDDR1A in children with rickets not responding to cholecalciferol in resource limited countries.

Calcitriol seems to be more efficient than alfalcacidol in term of healing of VDDR1A rickets.

REFERENCES


Figure-1 Pathophysiology Of Vitamin D dependent rickets type 1A