A rare form of Vitamin D Receptors dysfunction (vitamin D-dependent rickets (type II) with alopecia. A case Report.

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Introduction

Vitamin D-dependent rickets type II is an autosomal recessive disorder caused by a defect in the vitamin D receptor gene located on chromosome 12q12–q14. So far, 13 mutations have been identified. It is characterized by hypocalcemia, secondary hyperparathyroidism, and early onset severe rickets. We report a case of severe form of rickets associated with alopecia.

Case Report

This 23-month-old term boy born to a consanguineous parents.
He was referred for evaluation of his short stature, persistent hypocalcemia that not responding to oral calcium, and vitamin D therapy.
He had progressive alopecia capitis, delayed gross motor milestones, bow legs and macrocephaly. He had no family history of vitamin D or calcium related abnormalities.
He had been on vitamin D (1400 units/day) and oral calcium prescribed by a pediatrician.

He did not have any gastrointestinal or urinary symptoms, and did not develop any seizure or impaired consciousness.

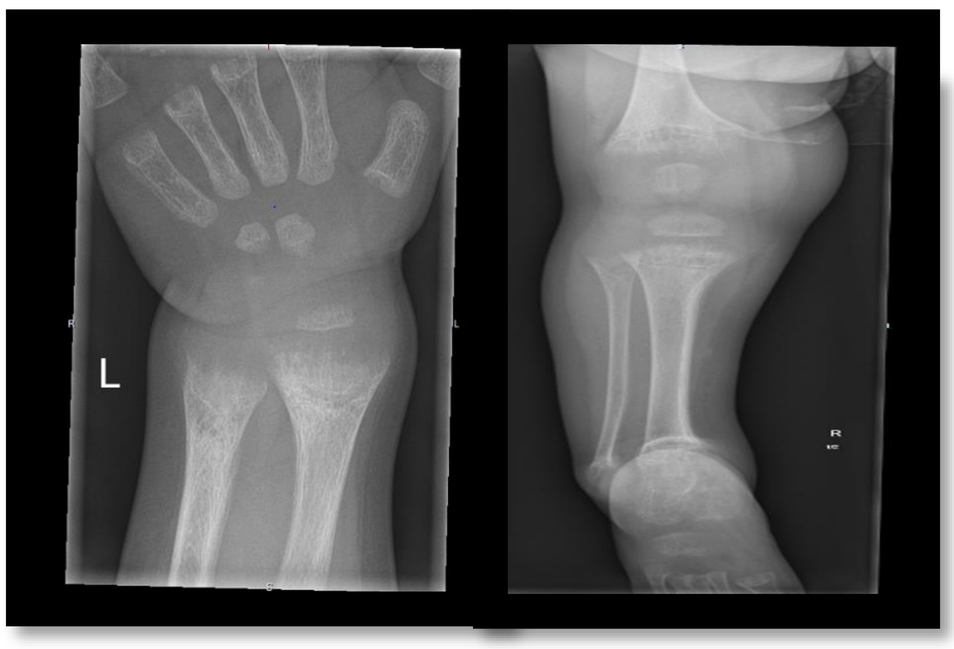
Clinically has alopecia capitis and closed fontanelle. His weight = 12.15kg (0.01SD), length = 79.5cm (-2.7 SD) and head circumference 48.8cm (0.48SD)

He had parietal and frontal bossing, rachitic rosaries, Harrison sulcus, widening of the wrist and bowing of the distal radius and ulna and bowing of the femur and tibia. His systemic examination otherwise was unremarkable

Radiologic investigation revealed extensive cupping, fraying and splaying of the distal metaphysis of the both femora and proximal and distal metaphysis of the tibia and fibula with diffuse osteopenia and cortical thinning of the shafts of the tibia and fibula.

Initial laboratory investigation, as follow.

	Before treatment	4 weeks after treatment	Normal value
Serum total calcium	1.72 mmol/L	1.62 mmol/L	2.32-2.64
Serum phosphate	1.11 mmol/L	1.31 mmol/L	1.45-2.33
Serum creatinine	15 mcmol/L	16 mcmol/L	17-36
Serum Alkaline phosphatase	1531 IU/L	1521 IU/L	134-315
Serum Magnesium	0.81 mmol/L	0.86 mmol/L	0.70-1.00
25 OHD level	69 nmol/L	-	-
PTH – intact molecule	46.3 Pmol/L	52.7 Pmol/L	1.3-5.8
1,25 OH2 vit D	870 Pmol/L	-	58-207



Patient was started on oral elemental calcium (100 mg/kg/day) divided q8 hourly and alfa calcitriol (0.15 mcg/kg) 2 mcg daily. Monitoring (table) showed no improvement of biochemical or radiological parameters after 4 weeks of this treatment.

Conclusions

Therapy with high dose of vitamin D analogues with 100 mg/kg of elemental calcium orally was not effective in our case. Higher doses of oral calcium or in combination with intravenous calcium. therapy has been discussed with the parents as a next possible therapy.

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