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Stunted growth and alopecia totalis : A case report

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Introduction

Hereditary resistance to vitamin D (HRVD), formerly known as Vitamin D receptor resistance type II (VDRR II), is an autosomal recessive disease caused by mutations in the vitamin D receptor (VDR). It is a very rare form of rickets, with only about 100 cases reported. The typical clinical and biological signs are severe rickets, hypocalcemia, hypophosphatemia, secondary parathyroidism and markedly increased serum levels of calcitriol [1,25(OH)2D]. Children often exhibit growth impairment, poor bone structure (bowed legs, widening of the wrists, craniotabes...) and alopecia totalis (complete lack of body hair) in 2/3 of cases. The disease presents a broad clinical picture that largely depends on the genotype.

Aim

Case Report

Methods

The patient is a female infant whose parents continually sought medical advice regarding a marked alopecia. It was only when she reached the age of two, owing to relative psychomotor retardation and poor overall body growth, that the suspicion of rickets was raised. The clinical examination showed the typical signs of rickets: bowed legs, widening of the wrists, rachitic rosary and alopecia. X-ray examination confirmed a generalised lack of bone mineralisation and uncovered fractures of the distal forearms bilaterally. A blood test showing hypocalcemia, hypophosphatemia, excessive 1,25(OH)D, increased PTH levels and markedly high alkaline phosphatase strengthened the suspicion of HRVD. Genetic analysis uncovered a mutation in the coding region of the VDR gene, confirming the diagnosis. The treatment initiated was oral administration of calcium carbonate (1g three times a day) and vitamin D (25000 U/week). However, owing to poor response, it was switched to a daily intravenous administration of high doses of calcitriol and calcium. The patient was initially hospitalised for her treatment and later discharged with arrangements made for her intravenous infusions to be undertaken at home.

Results

The human VDR results from a single gene located on the chromosome 12(12q13-14), and mutations in the coding region of this gene lead to severe functional disorders. Among the diverse functions of vitamin D, calcium homeostasis is the most apparent one. Thus, malfunctioning VDR leads to poor calcium uptake by the digestive system. With a decrease in calcemia, the secondary parathyroidism that follows in turn depletes the calcium stock in the bones and favours phosphate excretion in the urine. While the physiopathology for the alopecia is still unclear, its presence indicates a more acute presentation of the disease. Treatment aims at enhancing growth, restoring normocalcemia and normal PTH levels, and improving bone mineralisation. Response however varies greatly and depends on the severity of the affection. HRVD patients presenting alopecia generally do not respond to oral treatment and intravenous infusions are therefore required.

Conclusions

This case illustrates a very rare and probably known-to-few affection of end-organ resistance to vitamin D, and depicts the cruciality of the latter for proper mental and