HAIR DISORDERS

HEREDITARY VITAMIN D RESISTANT RICKETS: A RARE CAUSE OF ALOPECIA: REPORT OF 3 TUNISIAN CASES

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Background: Hereditary vitamin D resistant rickets (HVDRR) is an autosomal recessive disorder resulting from end-organ resistance to 1, 25(OH) 2D3. Alopecia may be the initial clinical finding in affected neonates or occurs during the first few months of life.

Observation: Three patients, including two siblings aged 9 and 18 years old and a 4-year-old girl, born to a consanguineous marriage presented to our clinic with alopecia since few months after birth. Clinical examination revealed growth retardation, severe deformation of the lower limb, total scalp and body alopecia, reduced eyebrow and eyelash hair and conic teeth with severe dental caries. Biological studies revealed reduced calcium levels, hypophosphatemia, higher levels of alkaline phosphatase and elevated serum 1, 25 (OH) 2 D levels. Radiographs of the legs and wrists showed advanced signs of rickets.Thus the diagnosis of hereditary vitamin D resistant rickets associated with alopecia was assessed. Treatment included high doses of the active form of vitamin D3 with calcium supplement with dramatic improvement of their rickets but alopecia didn't improve. A genetic investigation was performed for the two siblings showing a mutation located in the exon 2 of the vitamin D receptor (VDR) gene.

Key messages: In front of a congenital total alopecia associated with clinical and biological signs of rickets, vitamin D resistant rickets must be suspected. The mechanism causing alopecia is unknown but VDRs are present in the hair follicle which explains the maintenance of alopecia despite high doses of the active form of vitamin D3 with calcium. Therefore, there is probably a role for the vitamin D pathway in regulating hair follicle biology in humans.