

Vitamin D Dependent Rickets Type II with Alopecia

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ABSTRACT Vitamin D dependent rickets Type II (VDDR II) is a rare form of autosomal recessive disorder, which is inherited and often caused by mutation of a gene on vitamin D receptor which in turn prevents normal physiological response to 1, 25(OH)₂ vitamin D. The elevated circulating levels of 1,25-dihydroxyvitamin D₃ is a diagnostic feature of VDDR II, while the presence of alopecia differentiates it from vitamin D-dependent rickets Type I. VDDR II is considered to be the best example of aberrant target tissue response. The researchers thereby report a case of a two- year old female child with short stature, typical features of rickets and lack of hair growth. Upon investigation the level of 1,25(OH)₂D was found to be high which is diagnostic of VDDR type II. The child was started on high dose of calcitriol and oral calcium but on follow- up did not show much improvement probably due to poor compliance.