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Genetic and Functional Characterization of Vitamin D-Resistant Rickets

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GENETIC AND FUNCTIONAL CHARACTERIZATION OF VITAMIN D-RESISTANT RICKETS

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In this study the 1,25-dihydroxyvitamin D₃ receptor (VDR) in cells of patients with vitamin D-resistant rickets (VDRR) were analyzed for genetic and functional characteristics. The human VDR gene has been cloned and sequenced so the normal gene can be compared with the defective disease gene. The VDR of two patients with VDRR each showed a homozygous point mutation within the third exon, a mutation which substituted a glutamine for an arginine residue in a conservative area on the receptor in the steroid receptor superfamily. The mutant receptor (obtained via site-directed mutagenesis) bound vitamin D hormone with normal affinity, but displayed weak affinity for nuclei and DNA. The receptor was completely inactive in promoting transcription in a cotransfection assay employing a construction containing the CAT gene reporter fused downstream of the VDR-dependent osteocalcin gene promoter-enhancer. These results provide the genetic and functional basis for the disease phenotype of rickets in this inherited human disease.