

Open Peer Review on Qeios

Vitamin D Dependent Rickets 2b

National Cancer Institute

Source

National Cancer Institute. <u>Vitamin D Dependent Rickets 2b</u>. NCI Thesaurus. Code C131076.

Rickets caused by a post-receptor defect in the vitamin D signaling pathway producing vitamin D resistance due to constitutive overexpression of a nuclear ribonucleoprotein that competes with the vitamin D receptor-retinoid X receptor dimer binding with DNA vitamin D response elements. This condition has a similar phenotype to vitamin D receptor deficiency rickets including elevated 1,25-dihydroxyvitamin D (calcitriol) concentrations.

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