

# VETERINARY TECHNICAL DATASHEET

Hereditary Vitamin D-Resistant Rickets Type II, (HVDRR)



Mutation Found In :Pomeranian

## Disorder Type

- Skeletal

## Disease Severity

- Severe

## Background

Hereditary vitamin D-resistant rickets (HVDRR) is characterized by defects in the vitamin D receptors of the end-organs, preventing vitamin D from reaching its target tissues. Bone mineralization is inhibited in HVDRR causing softening and bending of the bones and skeletal malformations. The causative mutation for a form of HVDRR has been identified in Pomeranians.

## Key Signs

- Hypocalcemia
- Secondary hyperparathyroidism
- Hypomineralization of bones
- Bone malformations
- Rickets
- Joint pain
- Lameness
- Limb deformities
- Spontaneous fractures
- Alopecia

## Clinical Description

HVDRR is characterized by low calcium levels leading to secondary hyperparathyroidism. Vitamin D enhances bone mineralization and calcium is essential for hardness and strength of the bones. Vitamin D-resistance causes dysfunctional bone mineralization leading to the softening of bones and skeletal deformities. The clinical signs of HVDRR include joint pain, lameness, limb deformities, spontaneous fractures, jaw deformities, and neurological signs. HVDRR can also cause alopecia (hairlessness).

## Mode of Inheritance

- autosomal recessive

## Gene Name

- VDR

## Next Steps

As soon as a diagnosis of HVDRR has been confirmed affected dogs should be considered for treatment. Affected dogs can be treated by oral administration of active vitamin D and calcium supplements. The bone deformities are, however, irreversible.

## References

LeVine DN, Zhou Y, Ghiloni RJ, Fields EL, Birkenheuer AJ, Gookin JL, Roberston ID, Malloy PJ, Feldman D. Hereditary 1,25-dihydroxyvitamin D-resistant rickets in a Pomeranian dog caused by a novel mutation in the vitamin D receptor gene. *J Vet Intern Med* 23:1278-83, 2009.

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