Cinacalcet Treatment in Girls with Hereditary Vitamin D Resistant Rickets

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Objectives: HVDRR is characterised by hypocalcaemia, secondary hyperparathyroidism and severe early-onset rickets in infancy and is diagnosed easily especially associated with alopecia. Objective and hypotheses: Successful treatment requires reversal of hypocalcaemia and secondary hyperparathyroidism and is usually failed by high dose calcitriol but sometimes accomplished by administration of high doses calcium. Some patients need enteral or parenteral continuous calcium replacement that has low compliance or high complication risk.

Methods: Cinacalcet trial for HVDRR in 3-year-old girl who failed with high dose calcium and calcitriol.

Results: The patient has been administered conventional treatment for 2.5 years and did not response to therapy. Metabolic and clinical deterioration progressed. Parathormone and alkaline phosphatase levels could not reach to normal levels during treatment (about 400 pg/ml and above 1 000 U/l). Cinacalcet 16 mg/d was given and parathormone level decreased to normal at 4th day. Calcium and calcitriol doses were reduced and the phosphate replacement was ended. Metaphyseal cupping and fraying were improved, PTH and ALP was decreased to 36 pg/ml, 675 U/l respectively in 5th month of cinacalcet.

Conclusions: Cinacalcet should be considered in HVDRR patients who were not responsive to conventional treatment.

References: