Normal serum calcium levels and vitamin-D dependent rickets type 2 (VDDR-II): A novel vitamin D receptor mutation

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The authors declare there is no conflict of interest

Background: VDDR-II, is an autosomal recessive disorder characterized by the early onset of rickets with hypocalcemia, secondary hyperparathyroidism and hypophosphatemia and is caused by mutations in the vitamin D receptor (VDR) gene.

Case:

- 2 years old girl
- admitted to: total alopecia and bilaterally genu varum deformity (Fig 1 and 2).

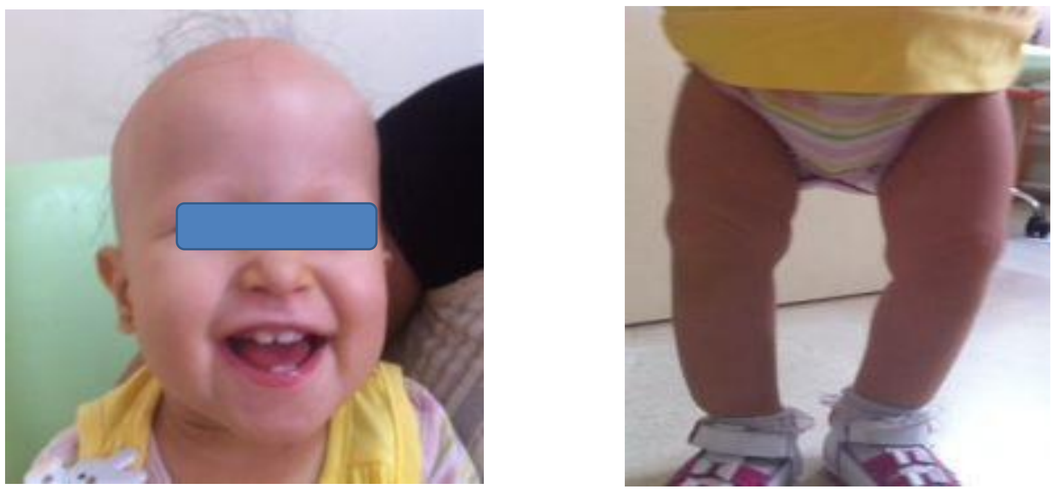


Figure 1-2: The patient have total alopecia and genu varum

History:

- Hair loss was observed at 3 months
- Motor and mental development was normal
- Used daily oral 400 IU vitamin D regularly
- First offspring of consanguineous parents

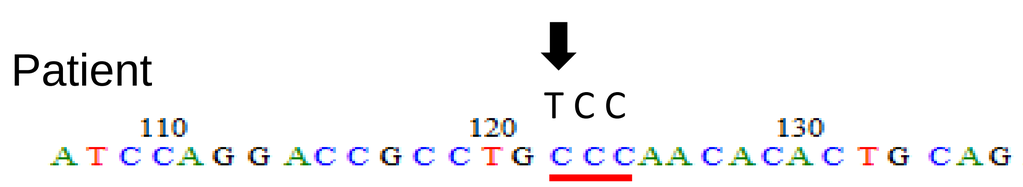
Physical exam:

- Weight: 9.2 kg (SDS: - 2.1)
- Height: 78.5 cm (SDS: -1.31)
- Total alopecia, frontal bossing, prominence of costochondral junctions, widening of wrists, and bilaterally genu varum deformity.

Laboratory

- Serum calcium:8.5- 9.4 mg/dl (8.5-10)
- Serum phosphorus: 2.2-3.1 mg/dl (3-5.5)
- ALP:988-1132 U/L (100-350)
- PTH: 450- 778 pg/ml (12-60)
- 25(OH) D3: 32 ng/dl (20-100)
- Blood gas analyses, renal and tubular functions tests were normal.
- X-ray of the left hand wrist and lower extremities were compatible with severe rickets.

- Molecular analysis of VDR gene : a novel homozygous missense mutation (S360P)
- Her parents were heterozygote (Figure 3)



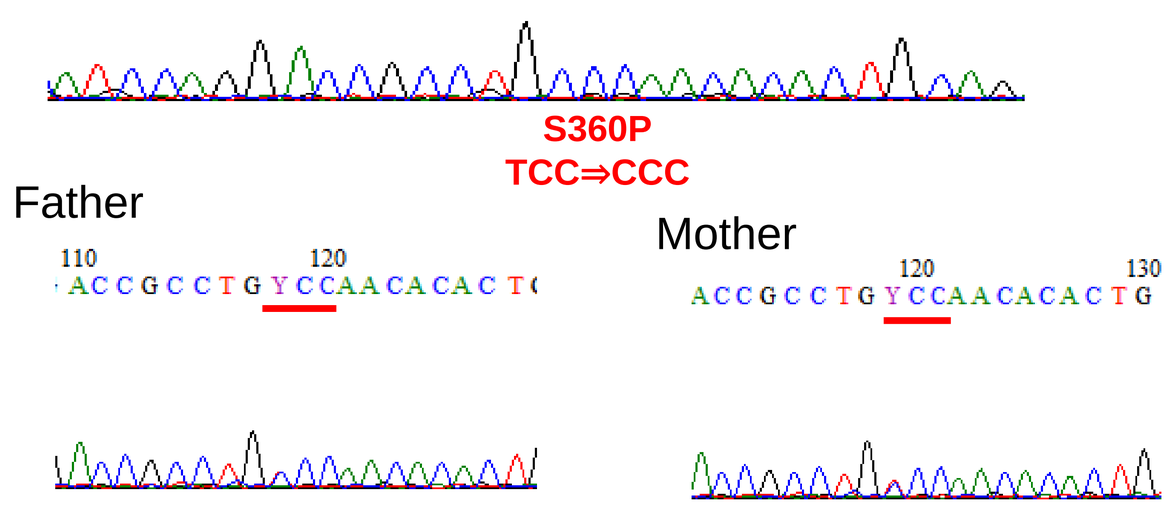


Figure 3: VDR gene molecular analysis from patient and her parents

Conclusion: The clinical spectrum of VDRR-II varies widely, probably reflecting the type of mutation within the vitamin D receptor and the amount of residual vitamin D receptor activity. S360P mutation in the vitamin D receptor may be associated with normocalcemic VDRR-II.