

A FEBRILE SEIZURE IN A TODDLER GIRL WITH ALOPECIA: A CASE REPORT

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INTRODUCTION

- Calcium homeostasis is primarily regulated by vitamin D. In the absence of the active hormone or a functional receptor, bones are inadequately mineralized, leading to the development of rickets.
- Vitamin D dependent rickets type 2 (VDDR2) is a rare autosomal recessive disorder caused by mutations in vitamin D receptor (VDR) gene.

CASE REPORT

- 12 month-old girl
- Three-minute 1st episode of seizures: unconsciousness, hypotonia, ocular reversion, cyanosis, and sialorrhoea.

- ∅ fever
- ∅ trauma
- ∅ accidental drug ingestion

- Pregnancy and delivery were unremarkable.
- Parents were nonconsanguineous.
- She had alopecia since birth and yet she was not able to walk, even with support.

Examination showed: **total alopecia**, closed anterior fontanelle, dentition according to her age, **enlarged wrists and bowed legs**.



Photos were taken after parental consent



Capillary blood gas showed **severe hypocalcaemia** (ionized calcium **0,76 mmol/L**)

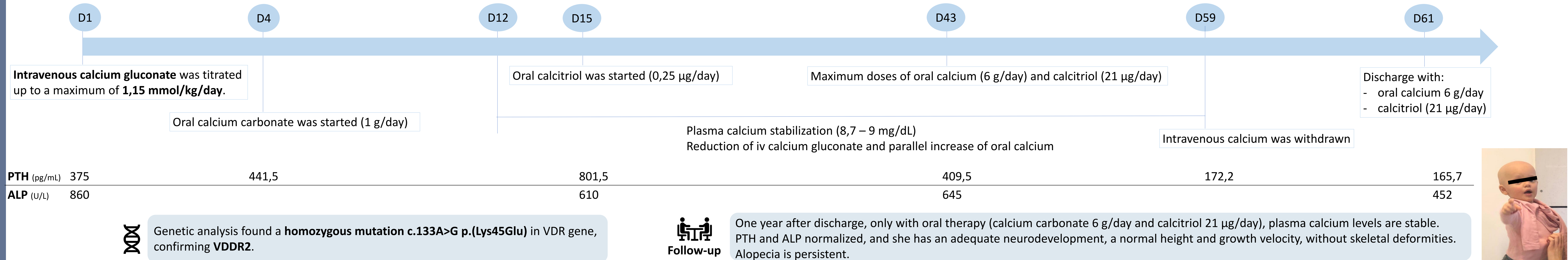
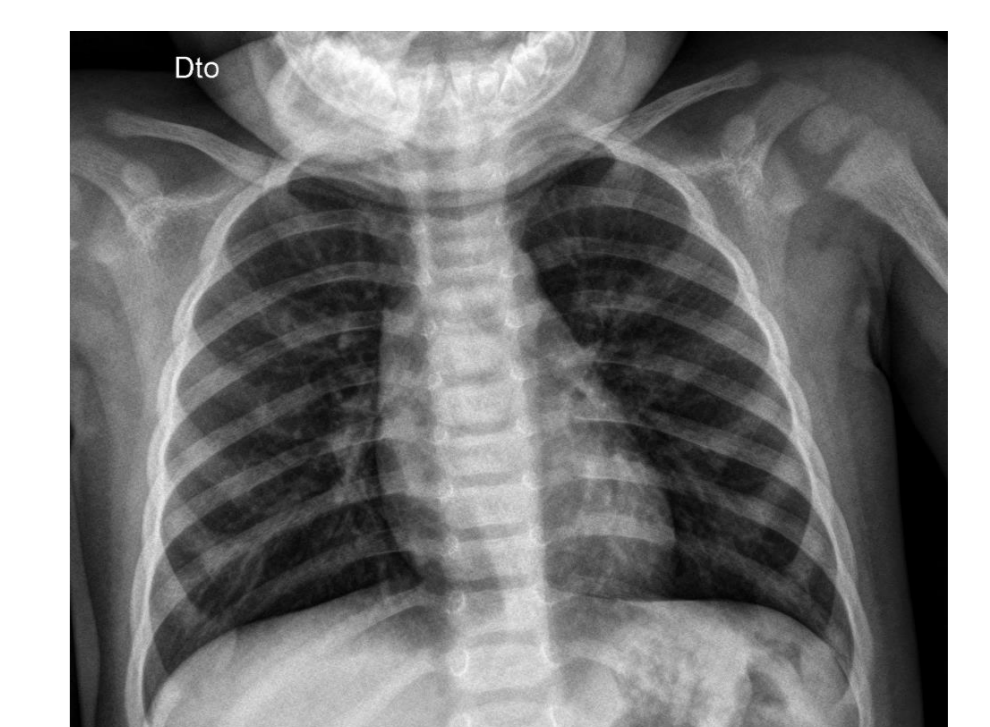
Calcium (mg/dL)	7,7 ↓	Parathyroid hormone (PTH) (pg/mL)	375 ↑
Phosphorus (mg/mL)	3,5 ↓	25(OH)D (ng/mL)	13,9
Alkaline phosphatase (ALP) (U/L)	860 ↑	1,25(OH) ₂ D (pg/mL)	> 189,0 ↑



- Full body skeleton X-ray:
 - Diffuse bone demineralization
 - "Rosary ribs"
 - Metaphysis flaring, fraying, and cupping

- Renal ultrasound was normal.

Vitamin D dependent rickets type 2 ?



CONCLUSIONS

- Genetic mutations are the cause about 13% of rickets.
- VDDR2 secondary to Lys45Glu mutation prevents VDR from activating gene transcription.
- **High levels of 1,25(OH)₂D and alopecia** are the distinct points of this disease, and alopecia is thought to be a sign of disease severity.
- Control of secondary hyperparathyroidism is the therapeutic goal, decreasing bone demineralization.
- Intravenous calcium treatment for several months, followed by high doses of oral calcium and calcitriol, seems to be an effective approach.
- Unfortunately, alopecia is persistent and a heavy feminine psychosocial burden.

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