AFEBRILE 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.

Intravenous calcium gluconate was titrated up to a maximum of 1.15 mmol/kg/day.

Oral calcium carbonate was started (1g/day).

Oral calcitriol was started (0.25µg/day).

Maximum doses of oral calcium (6g/day) and calcitriol (21µg/day).

Discharge with:
- Oral calcium 6g/day
- Calcitriol (21µg/day)

Intravenous calcium was withdrawn

Plasma calcium stabilization (8.7 – 9 mg/dL)
Reduction of iv calcium gluconate and parallel increase of oral calcium

D1
D4
D12
D15
D43
D59
D61

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Capillary blood gas showed severe hypocalcaemia (ionized calcium 0.76 mmol/L)

Calcium (mg/dL) 7.7 ↓
Parathyroid hormone (PTH) (ng/mL) 375 ↑
Phosphorus (mg/dL) 3.5 ↓
25(OH)D (ng/mL) 11.9
Alkaline phosphatase (ALP) (UI/L) 860 ↑
1,25(OH)2D (ng/mL) > 189.0 ↑

Genetic analysis found a homozygous mutation c.133A>G p.(Lys45Glu) in VDR gene, confirming VDDR2

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.

REFERENCES


CONTACT INFORMATION

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