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A rare case of familial hypocalcemia

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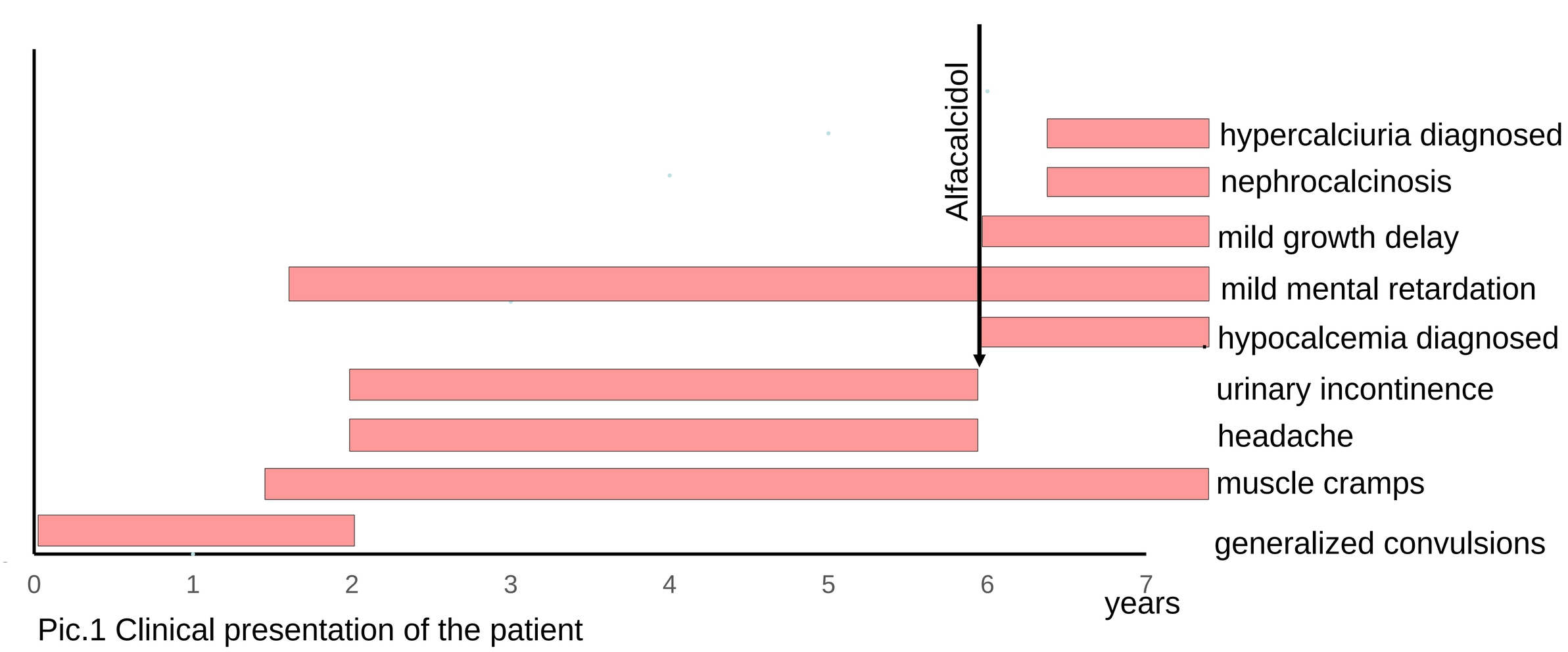
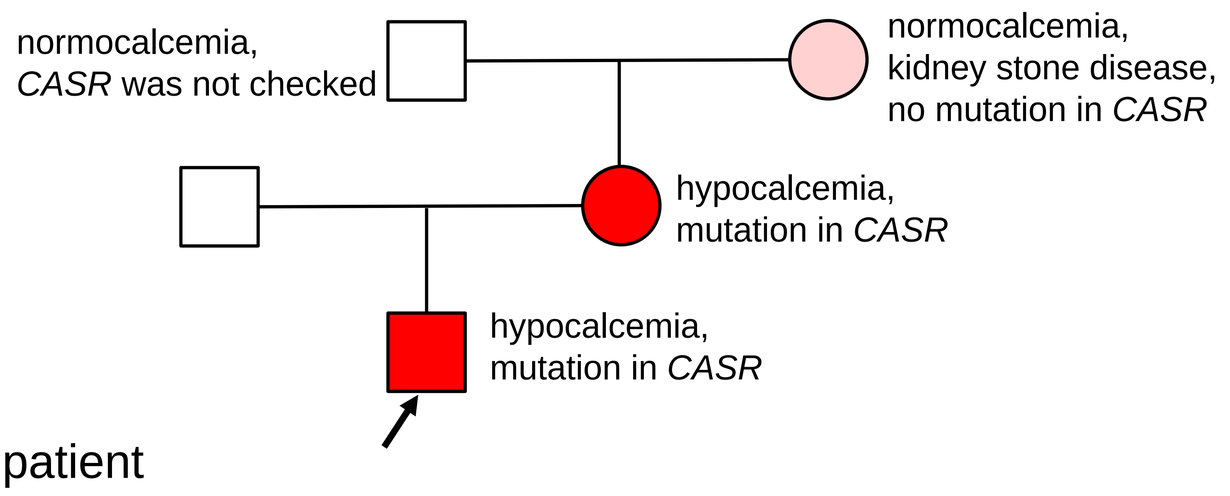
Nothing to disclose

Background: Familial hypocalcemia is a rare autosomal dominant disease characterized by hypercalciuric hypocalcemia. The disorder is caused by heterozygous mutation in the *CASR* gene that encode a calcium-sensing receptor in parathyroid glands and kidney tubules.

Clinical case: The boy was born at term from non-consanguineous parents with normal length and weight. On the second day of life he was admitted to an intensive care department with convulsions. Calcium level was not checked, anticonvulsant drugs were prescribed. Treatment was not effective and generalized convulsions repeated every month. After he turned 2 years old generalized convulsions stopped, but muscle cramps, headache and urinary incontinence persisted. Hypocalcemia (1.62 mmol/l), hyperphosphatemia (3.41 mmol/l) and low PTH (0.57 pm/l) were detected at the age of 6 for the first time. Hypoparathyroidism was diagnosed and alfacalcidol was initiated. At the age of 6.5 he was admitted to our hospital. Chvostek's sign, mild mental retardation, mild growth delay (SDS – 1.69) were observed at the examination. Signs of nephrocalcinosis were found by ultrasound. He had mild hypocalcemia (total calcium 1.96 mmol/l, ionized calcium 0.96 mmol/l), and hypercalciuria (calcium/creatinine ratio was 1.05). Autosomal dominant hypocalcaemia was suspected.
The patient's mother didn't have a history of convulsions, but she had mild muscle cramps of arms and legs and episodes of lockjaw. She was found to have hypocalcemia, low PTH and nephrocalcinosis.
Previously undescribed heterozygous deletion c.344-358del in *CASR* was detected in both, the boy and his mother. Grandparents of the boy had normal levels of calcium and phosphate. Maternal grandmother had a history of kidney stone disease, but the mutation in *CASR* was not detected.



Pic.1 Clinical presentation of the patient



Pic.2 Pedigree of the patient

Conclusion

Familial autosomal dominant hypocalcemia due to mutations in CASR gene is a rare cause of hypocalcemia. Clinical presentation of the disease could be variable within one family.