Cinacalcet experience in hypercalcemia due to CaSR mutation

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Introduction:

Heterozygous inactivating mutations of the CaSR gene (CaSR) generally result in mild, asymptomatic hypercalcemia in the familial hypocalcuric hypercalcemia syndrome. Homozygous inactivating CaSR mutations end up with neonatal severe hyperparathyroidism. Calcimimetics are drugs that interact with the transmembrane part of CaSR and make the receptor more sensitive to calcium. Cinacalcet, a type II calcimimetic, suppresses PTH levels and increases renal calcium excretion.

Case:

A 7-years 9 months-old girl presented with hypercalcemia. She was diagnosed with hypercalcemia because of vomiting and abdominal pain when she was 1,5 years old. She had taken hydration, diuretic, steroid and pamidronate treatments. Genetic analysis revealed a p.R185Q (c.554G> A) heterozygote mutation and p.A986S (c.2956G> T) polymorphism in the CaSR gene. 1 mg / kg / day furosemide treatment was learned. On physical examination, weight: 24.4 kg (25-50p), height: 118.7 cm (10p), blood pressure 90/60 mmHg, and other system examinations were normal. When Ca: 14.1 mg / dl was considered, i.v hydration, i.v furosemide and prednisolone treatments were started. Despite these treatments, Ca: 13,2 mg / dl, 1x30 mg / day cinacalcet treatment was started. Ca: 12.2 mg / dl at the 24th hour of treatment and Ca: 11.6 mg / dl at the 96th hour. She was treated with cinacalcet for 3 years and her calcium level was between 11,1-12,3 mg / dl. No side effects were observed. Renal ultrasonography was normal.

Result:

Cinacalcet treatment has been reported to be effective in the treatment of severe hypercalcemia due to CaSR mutations, especially in the neonatal period. Calcium levels of the patient were at the upper limit of normal for 3 years and no side effects were observed. In cases of hypercalcemia resistant to other therapies, treatment with cinacalcet may be given.