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Cinacalcet sustainedly prevents pancreatitis in a child with a compound heterozygous SPINK1/AP2S1 mutation

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Abstract

Familial hypocalciuric hypercalcemia is an autosomal dominant genetic disorder characterized by hypercalcemia associated with inappropriate hypocalciuria and normal parathyroid hormone levels. Acute recurrent pancreatitis (ARP) is rare in children. Predisposing factors include hypercalcemia and mutations in the serine protease inhibitor Kazal-type 1 (SPINK1) gene. The disease carries a heavy morbidity and preventive treatment options are scant. Here, we report a child with a novel genetic/metabolic form of ARP associated with compound heterozygous SPINK1/AP2S1 (adaptor protein-2 σ1-subunit) mutations, recurrence of which was completely abrogated for 6 years by cinacalcet treatment.

Keywords: Children; Cinacalcet; Genetic; Hypocalciuric hypercalcemia; Pancreatitis.