INTRODUCTION

The human calcium-sensing receptor (CASR) plays an essential role in the regulation of extracellular calcium homeostasis. Heterozygous germline inactivating mutations in the CASR gene cause a decrease sensitivity to extracellular calcium, impairing the ability to sense and correct hypercalcemia, and have been reported in cases with hypocalciuric hypercalcemia (FHH). When two inactive gene copies are inherited (or one single mutant allele acts in a dominant negative manner), the patients may manifest neonatal severe hyperparathyroidism (NSHPT), presenting life-threatening hypercalcemia in the neonatal period. Other related clinical manifestations include bone lesions, feeding difficulties, respiratory distress and hypotonia. The clinical management of NSHPT is difficult. Surgical treatment with total parathyroidectomy has been recommended for the most severe cases, but it can lead to iatrogenic hypoparathyroidism.

Some cases benefit from major medical interventions (like bisphosphonates and calcimimetics) before surgery or as an alternative to surgery. To our knowledge, long-term follow-up of these patients is scarce.

First daughter of a healthy, young, unrelated couple. Uneventful pregnancy and birth: Birth weight: 3100 g (10.25th centile)

Severe hypercalcaemia, respiratory distress and hyperventilation. Transfer to Neonatal Intensive Care

After an initial period fraught with difficulties, meticulous metabolic control of calcium and 1,25-dihydroxyvitamin D allowed for normal growth and development.

CLINICAL CHARACTERISTICS AND LONG-TERM FOLLOW-UP OF A PATIENT WITH NEONATAL SEVERE HYPERPARATHYROIDISM

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CONCLUSIONS

In this girl, a mutation in the CASR gene was identified in a family with FHH/NSHPT.

The complexity of this clinical condition requires a multidisciplinary approach to minimize morbidity. In the present case, total parathyroidectomy with reimplantation was needed to control hypercalcaemia. After a challenging admission to control calcium levels, a good clinical evolution was reported at fifteen-year follow-up.

Identification of a CASR mutation allowed family genetic counseling.

REFERENCES


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