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NEUROLOGICAL PICTURE

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Intracranial calcifications and activating mutation of the calcium-sensing receptor

A 58-year-old man was referred to us because of asymptomatic hypocalcaemia (calcium 6.5 mg/dl, normal range 8.5-10.4). The patient had one healthy son and two daughters with hypocalcaemia. Analytical study confirmed hypocalcaemia (ionised calcium 0.86 nmol/l, normal range 1.13-1.32), which was accompanied by low levels of intact parathyroid hormone (19 pg/ml, normal <65). Serum phosphorus, 25-hydroxyvitamin D and 1,25-dihydroxyvitamin D were within normal range. Hypocalcaemia was associated with hypercalciuria when oral calcium and vitamin D were administered. Gene study showed a novel activating mutation (Glu(604)Lys) affecting the calcium-sensing receptor in the patient and his two daughters. Morphological studies of the patient showed multiple intracranial calcifications affecting the basal ganglia, cerebellum and brain parenchyma (figs 1, 2). We think that activating mutations of CaSR should be considered as a new cause of hypocalcaemia-induced asymptomatic intracranial calcifications.

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Figure 1 Lateral radiography of the cranium showing basal ganglia and cerebellar calcifications.

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Figure 2 Computed tomography images of the brain showing multiple calcifications of the cerebellum (left), basal ganglia and brain parenchyma (right).