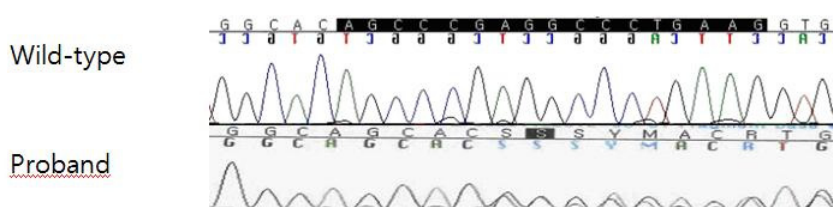


Concurrent type 2 diabetes and multiple endocrine neoplasia type 1 with a novel ME N1 gene mutation

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Multiple endocrine neoplasia type 1 (MEN 1) is an autosomal dominant inherited disorder that is characterized by the combined occurrence of primary hyperparathyroidism, enteropancreatic neuroendocrine tumors and anterior pituitary adenomas. Increased insulin resistance and a higher prevalence of glucose intolerance have been noted in MEN 1 families and are associated with increased cardiovascular mortality. Mutations in the MEN1 tumor suppressor gene cause multiple tumors in MEN1 but can also be associated with glycemic dysregulation. A 52-year-old man was admitted for blood glucose control. He had been diagnosed with diabetes three years previous. Hypercalcemia was incidentally detected during a blood test. After examination, he was diagnosed with primary hyperparathyroidism caused by parathyroid hyperplasia. On genetic analysis, he was found to have a novel mutation in the MEN1 gene (exon 10, c.1590_1606_del; p.Gly532AlafsX20). We report a case of MEN type 1 with a novel MEN1 gene mutation associated with type 2 diabetes in a patient with no family history of diabetes. This is the first case of MEN 1 associated with type 2 diabetes before pancreatic surgery in Korea.



A Case of Camurati-Engelmann Disease with clinical features of Van Buchem Disease

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Camurati-Engelmann disease is a rare autosomal dominant type of bone dysplasia. The mutation of the gene encoding transforming growth factor(TGF)- β 1, results bilateral and symmetric cortical thickening of the periosteal and endosteal surfaces of long bones. Although half of the patients have sclerosis of skull, a few patients have mandible sclerosis and enlargement. Van buchem disease is a rare autosomal recessive bone dysplasia, which is characterized by progressive cortical thickening of long bones and mandible enlargement. Here we report a 30-years-old woman who diagnosed as Camurati-Engelmann disease by detect TGF- β 1 gene mutation but had typical clinical features of Van Buchem disease.

