A novel mutation causing an unusual presentation of MEN1 Syndrome

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We report a 16 years old female (IP) of Pakistani origin who presented initially with obvious Cushingoid features noted by attending physician who was treating the patient's older sister for hyperprolactinemia. The diagnosis of Cushing syndrome was confirmed after further hormonal assessment. Magnetic resonance imaging of the abdomen showed a large left adrenal mass (9.7 x 11.8 cm); small pulmonary and hepatic lesions were also noted. She had a left open adrenalectomy; histology was reported as adrenocortical carcinoma. With a presumptive diagnosis of multiple endocrine neoplasia, screening of her two sisters and father ensued and confirmed high serum calcium (2.9-3.1 mmol/l) and parathyroid lesions by ultrasonography. Genetic testing revealed a heterozygous partial gene deletion in Exons 3-10 (c.446-1833) of the MEN 1 gene.

Medical management was compromised by financial and insurance issues; no further specific treatment for the adrenal carcinoma or hyperparathyroidism was possible. In spite of this, and slow progression of metastatic disease she is alive some 15 years after the initial diagnosis. However, over the last year she has received chemotherapy for metastatic tumour- reportedly Ewing's sarcoma.

The case described here is unusual. The mutation found in this family is novel. To our knowledge, the association between Ewing's sarcoma and MEN1 has not been previously reported. Moreover, the slow progression of adrenocortical carcinoma is this case is unusual. The other known affected family members have shown no evidence of malignant disease so far, although all have osteoporosis and hyperparathyroidism.