中文題目：腎上腺腫瘤表現似非典型先天性腎上腺增生症

英文題目：Adrenal tumor mimicking non-classic congenital adrenal hyperplasia

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Background: Elevated 17-hydroxyprogesterone may be caused by congenital adrenal hyperplasia, ovarian or adrenal tumors. A positive cosyntropin stimulation test result for 17-hydroxyprogesterone may be found in functional or non-functional tumors and be related to tumour size.

Patients and Methods: We present a case of a 36-year-old woman with a 4-year history of infertility. Laboratory test results revealed elevated progesterone and 17-hydroxyprogesterone, with normal luteinizing hormone, follicle-stimulating hormone, oestrogen, testosterone, dehydroepiandrosterone sulphate, and anti-Mullerian hormone levels. The 250-μg cosyntropin stimulation test revealed a 17-hydroxyprogesterone level of 11.3 ng/mL and 31.8 ng/mL at 0 min and 60 min, respectively. Non-classic congenital adrenal hyperplasia was diagnosed initially; however, genetic testing revealed no 21-hydroxylase deficiency. She received dexamethasone but progesterone and 17-hydroxyprogesterone levels remained high. Abdominal computed tomography found a 4.5 × 4.8-cm left adrenal tumor.

Results: Subsequent pathological report was compatible with an adrenal cortical adenoma. Progesterone and 17-hydroxyprogesterone levels returned to the normal range postoperatively and the 250-μg cosyntropin stimulation test of 17-hydroxyprogesterone showed a normal response.

Conclusion: When biochemically diagnosed NCCAH is diagnosed, gene mutation analysis should be performed if available. If the patients suffer from poor response to steroid supplementation, the physician should consider adrenal and ovarian imaging. Adrenalectomy may demonstrate promising improvement in suppression of 17-OHP and progesterone.