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A novel pathogenic variant of ARMC5 in a patient with primary bilateral macronodular adrenal hyperplasia: A case report

Wei Wang and **Feng Wei**
First Afliated Hospital of Baotou Medical Collage, China

Background: Primary bilateral macronodular adrenal hyperplasia (PBMAH), also known as adrenocorticotropic hormone (ACTH)-independent macronodular adrenal hyperplasia, is a rare cause of endogenous Cushing's syndrome. In many familial cases of PBMAH, the variants in armadillo repeat containing 5 (ARMC5) gene are found to be associated with the disease. Here, we report a case of PBMAH harboring a novel frameshift variant in ARMC5 gene, which has not been previously reported in the literature.

Case presentation: A 67-year-old woman was referred due to the clinical features of Cushing's syndrome. Radiological imaging and hormonal testing were carried out. The serum levels of cortisol were remarkably increased at late night and did not suppress even after 1 mg of dexamethasone administration, while the plasma levels of ACTH hormone were decreased signifcantly. The patient underwent unilateral left-sided laparoscopic adrenalectomy, and the diagnosis of PBMAH was substantiated by histopathological analysis. Moreover, the partial envelope was incomplete and the cell proliferation index was low. Specifcally, inhibin α-subunit (+), syn focal (+), Ki-67~3% (+), CgA (-) and CEA (-) were observed. DNA sequencing data revealed that a novel frameshift variant (c.363_373delGCCAGTGCGCC, p.Pro122Alafs*61) was identifed in ARMC5 gene. However, this variant was not detected in the daughter of the patient. The rest of the family members, including her sister, son and two brothers, were not consented for genetic testing.

Conclusions: Early detection of ARMC5 variant status and familial screening might have important clinical implications for the diagnosis and prognosis of PBMAH patients. A novel ARMC5 frameshift variant (c.363_373delGCCAGTGCGCC, p.Pro122Alafs*61) was identifed to be associated with the pathogenesis of PBMAH. ARMC5 sequencing may improve the identifcation of a causative gene variant for PBMAH and allow earlier diagnosis of this disease.

Keywords: Cushing's syndrome, ACTH-independent macronodular adrenal hyperplasia, Primary bilateral macronodular adrenal hyperplasia, ARMC5, Variant, Case report.

Recent Publications:

1.Wang W, Wei F. A novel pathogenic variant of ARMC5 in a patient with primary bilateral macronodular adrenal hyperplasia: a case report. BMC Endocr Disord. 2022 Aug 22;22(1):211. doi: 10.1186/s12902-022-01128-x. PMID: 35996143; PMCID: PMC9396781.

2.Wang W, Wei F, Li RH, Tian JH. A case report of idiopathic hyperaldosteronism characterized by bilateral adrenal adenoma. Medicine 2019;98:43(e17418).

Biography

Wei Wang, Chief physician, with master's degree, MD candidate, master's degree supervisor. International clinical bone mineral density evaluator, and ICSD lecturer, mainly engaged in research on endocrine and metabolic diseases; She presided over and participated in many clinical scientific research teaching work and published academic papers. Familiar with the standardized diagnosis and treatment of various common endocrine and metabolic diseases. In 2019, she studied in the Clinical Medical Center for Endocrine and Metabolic Diseases, Ruijin Hospital Affiliated to Shanghai Jiaotong University, majoring in obesity and weight loss as well as the diagnosis and treatment of difficult and rare pituitary-adrenal diseases.

vvhappy521@163.com