A RARE CASE OF LIDDLE SYNDROME DUE TO A NOVEL MUTATION IN EPITHELIAL SODIUM CHANNEL α-SUBUNIT (SCNN1A) IN A NORMOTENSIVE ADULT : A CASE REPORT

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ABSTRACT

Introduction

Liddle syndrome is a genetically heterogenous autosomal dominant disorder. The key clinical characteristics of this syndrome are early onset salt-sensitive hypertension with hypokalemia, metabolic alkalosis, inhibition of renin activity and aldosterone secretion. Liddle syndrome is caused by mutations (missense or frameshift) in the genes of epithelial sodium channel (ENaC) subunits.

Case report

The patient is a 58-year male, diabetic for 4 years, normotensive, known chronic kidney disease for 4 years, presented to our OPD with symptoms of generalized weakness especially lower limbs. Routine testing revealed hypokalemia, hypomagnesemia, anemia. Patient had persistent hypokalemia which didn't respond to oral potassium supplements. After ruling out the common causes of persistent hypokalemia and based on the findings of hypokalemic metabolic alkalosis, hypomagnesemia, urinary K and TTKG suggestive of renal potassium loss, a presumptive clinical diagnosis of a channelopathy (Bartter/Gitelman syndrome) was made. The genetic panel test however made a revelation of Liddle syndrome due to a novel mutation in α-subunit of epithelial sodium channel (ENaC). This is the first case of Liddle syndrome to be reported from our Centre and one of the few from our country.

The patient was put on Amiloride 5 mg along with oral potassium supplements. Subsequently there was an improvement in muscle power. One month later, patient was off potassium supplements and maintained normal electrolytes with Tab Amiloride.

Conclusion

Our results expand the mutational spectrum of Liddle syndrome, this being the second α-subunit mutation to be reported in literature. The role of genetic testing in appropriate clinical setting cannot be overemphasized.

Keywords: Liddle syndrome, ENaC, SCNN1A