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Genetic Analysis of Barter's and Giltelman's Syndromes in Saudi Patients

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Background

Bartter and Gitelman syndromes are rare autosomal recessive disorders characterized by hypokalemia, metabolic alkalosis, and normal to low blood pressure. They can be clinically divided into antenatal and classical Bartter's syndrome and Giltelman's syndrome. On the other hand, they can be classified into five subtypes (I to V) based on the underlying mutant genes. Furthermore, the role of gene mutation in Bartter's and Giltelman's syndromes has not been previously studied on the Saudi population.

Methods

Four unrelated Saudi patients were screened for genetic mutations. DNA was extracted from whole blood using the Gentra Pure gene DNA purification blood Core Kit C. Primers were designed to include exon and intron boundaries for the following genes: *SLC12A1*, *KCNJ1*, *CLCNKB*, *BSND* and *SLC12A3*. Polymerase chain reaction was performed through PTC200 Thermal Cycler and was checked on 2% agarose gel. Mega BACE DNA analysis system was used to screen for mutations, and the data was analyzed by Lasergene Software. Biochemical data was extracted from patients' files.

Results

Among the three studied genes, 3 novel mutations and a reported one were discovered. Two mutations were identified in *NKCC2* gene in 2 patients, the novel c.1216 G>C (p.406 Asp>His) and the reported c.1942G>A (p.648Asp>Asn). Moreover, the other 2 novel mutations c.1325A>C (p.442Asn>Thr) and c.1685T>C (p.562Asn>Thr) in SLC12A3 and CLCKB respectively, were discovered in the other 2 patients.

Conclusion

In this study, we identified three nonsynonymous novel mutations, in addition to a known one in 4 unrelated Bartter and Gitelman syndromes patients. Further research is warranted in order to facilitate the early diagnosis of such genetic syndromes.

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