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Identification of single nucleotide polymorphisms within the OCT2 gene in the South African black population

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ABSTRACT

The Organic Cation Transporter 2 (*OCT2*) gene is responsible for facilitating the transport of cationic compounds, which include both endogenous substrates and clinical drugs. Single nucleotide polymorphisms (SNPs) within this gene were extensively explored in the South African black population as little research has been conducted on these individuals so far. We sequenced the *OCT2* promoter region of 10 DNA samples from the South African black population and identified four SNPs and one INDEL. We performed a luciferase assay to determine their effects on gene expression and we found two variants (rs59695691 and rs138765638) that showed a statistically significant change in luciferase expression suggesting that they may be associated with a change in *OCT2* regulatory function. We also indentified thirteen SNPs and two INDELs within the *OCT2* promoter region, and nine SNPs within the *OCT2* coding region through analysing various South African population studies. These variations could affect both gene expression and protein function. These findings help contribute to filling the gap pertaining to OCT variation in South African populations.