Single nucleotide polymorphisms of ESR1 gene and the risk of breast cancer in Sudanese women

Khalid Mohamed Adam*

Department of Medical Lab Sciences, Faculty of Applied Medical Sciences, University of Bisha- P.O.Box 551, Bisha - KSA

***Corresponding author:** dr.aboalbasher@gmail.com

Introduction: The prolonged exposure to estrogen is well established risk factor for breast cancer which is the most common malignancy among Sudanese women. The effect of estrogen hormone on target tissues is mediated by its alpha receptor, through the binding of the hormone to the receptor which in turn promote the proliferation and differentiation of mammary tissues. The estrogen receptor alpha is coded by polymorphic *ESR1* gene.

Objective: To evaluate the association between rs2234693, rs9340799 and rs1801132 single nucleotide polymorphisms on ESR1 gene and breast cancer risk, type and the receptor status.

Methodology: Three single nucleotide polymorphisms were genotyped in 139 breast cancer cases and 139 age-matched cancer-free controls. Leucocytes DNA was extracted and genotyped by PCR-RFLP method. The estrogen receptor status for 65 cases were determined on a freshly obtained breast tissues according to standard histological procedures.

Results: The results of breast cancer cases showed a young mean age 46.5±10.4SD years, high frequencies of married women 97.1%, and parity 89.9%. The family history of breast or other type of female cancers in Sudanese women, proved to be the strongest risk factor, with 12 folds increase in the risk of developing breast cancer (OR=11.8, 95% CI:4-34.2, *P-0).* Of all the three studied SNPs, rs9340799 (XbaI) was the only one to show statistically significant association with the risk of breast cancer (P-0.03), where unadjusted logistic regression of the genotypic variants of this SNP showed that Xx genotype had (OR=1.6, 95% CI:0.85-3.04, *P-0),* and xx genotype (OR=2.6, 95% CI:1.23-5.46, *P-0.01).* The association of genotypic variants of different SNPs with estrogen receptor status, showed a statistically significant association between Xx variant of (XbaI) with positive receptor status (OR= 5.3, 95% CI:1.8-15.8, *P-0.002).* The other two SNPs (PvuII and *HinfI)* showed statistically significant association only with mammographic density (P-0.0 and *0.01* respectively).

Discussion: The family history of female cancers and SNPs, rs9340799 (XbaI) are possible risk factors affecting breast cancer in Sudanese women.