5. CONCLUSION

Breast cancer has become a serious health problem worldwide as well as in India. In spite of so much research on its epidemiology and pathogenesis the exact cause of breast cancer is unknown. It is reported that this is a complex disease and involves various factors and processes including environmental and genetic factors (Ponder, 2001). It is well reported that low penetrance genes like DNA repair and estrogen signalling may confer susceptibility to this diseases. Our study provides support that genetic polymorphisms in ESR1 and DNA repair genes may influence the susceptibility to breast cancer. Many studies have been carried out in different populations, but the results are inconsistent. Thus, large-scale association studies are required to determine the associations between these polymorphisms and breast cancer in multi-ethnic groups using high statistically powered tools.

Key Findings:

- Computational analysis revealed that nsSNPs influence the structure and function of the proteins and may be diseases associated. We found rs2307166 of XRCC1, rs5745906 of NEIL1 and rs79561451 of XRCC4 genes to be most damaging nsSNPs which are potential candidates for case-control studies that determine susceptibility to breast cancer.

- SNP rs25487 of XRCC1, rs7182283 and rs4462560 of NEIL1, rs3734091 of XRCC4, and rs9340799 and rs2234693 of ESR1 genes were found to be associated with breast cancer.

- Two SNPs (rs3798577 and rs2747648) which are reported to be present in miRNA binding site and one novel mutation were detected in 3’UTR region which is in process of submission to ClinVar database.