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Preliminary study on BRCA1 mutation analysis of Sri Lankan breast cancer patients

There has been a sudden increase in the number of patients diagnosed with breast cancer even in developing countries such as Sri Lanka in the recent past. Two breast cancer susceptibility genes: BRCA 1 and BRCA2 have been cloned previously. Inherited mutations in one or both of these genes are known to confer a predisposition to breast and ovarian cancer. Due to the large sizes of these two genes, it is not feasible to analyze large number of samples for all possible BRCA 1 and BRCA 2 mutations to determine population prevalence. Therefore the detection of mutations in these genes have to be

limited to commonly identified ones. Two BRCA 1 mutations have been detected relatively frequently, namely, 185delAG (exon2) and 5382ins C (exon 20).

In order to identify BRCA 1 mutation hot spots among Sri Lankan breast cancer patients we have PCR amplified exon 2 and exon 20 of 3 patients, exon 2 separately form another 9 patients and exon 20 of another 5 patients. All these patient samples were sequenced and the sequence data were computer analyzed using Gene Jockey II (Biosoft). None of the patient samples, appeared to have the two most common mutations, 185delAG and 5382ins C found in the other parts of the world. This might have been due to the low number of patient samples studied so far. Currently, work is in progress to further characterize these hot spot mutations among Sri Lankan breast cancer patients and to identify country specific mutations, if there are any.