

A preliminary study on TP53 tumour suppressor gene mutations in breast cancer patients of Sri Lanka

TP53 tumour suppressor gene is the most frequently found mutated gene and accounts for more than 50% of sporadic and inherited cancers. Worldwide TP53 mutations account for approximately 30% of breast cancer. Germline mutations of the TP 53 gene is associated with Li-Fraumeni Syndrome, that leads to the development of breast cancer and other neoplasms. Furthermore, somatic mutations of the TP53 mutations enables early detection and effective treatment of breast cancer. The presence of a TP 53 gene mutation is by far the signal most predictive indicator for recurrence and death. Therefore an attempt has been made to identify both somatic and genetic mutations of the TP53 gene hot spot mutations in the Sri Lankan breast cancer patients.

Presence of mutations for TP53 gene was studied in highly conserved regions of exon 5, 6, 7 and 8 where 90% of acquired mutations have been reported. We have analyzed blood and breast tissue DNA obtained from breast cancer patients before commencing treatment using PCR and DNA sequencing.

Of the 15 patients studied to date mutations in the TP 53 gene were seen in 6 patients. 4 patients showed somatic mutations with a single nucleotide mutation in their tissue DNA at codon 249 in exon 7, resulting from G to T transversion (n=2) or total G deletions (n=2). germline mutations were observed in 2 patients with total G deletion at the last base of the codon 249. The prevalence of codon 249 mutation for breast cancer has been reported to be only 0.3% worldwide and is the 5th most frequent mutation for breast cancer.