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**Monogenic susceptibility and influence of hormonal risk factors on
breast cancer: A hospital based study in Sri Lanka**

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ABSTRACT**MONOGENIC SUSCEPTIBILITY AND INFLUENCE OF HORMONAL RISK
FACTORS ON BREAST CANCER: A HOSPITAL BASED STUDY IN SRI
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Breast cancer is the commonest cancer among adult females in Sri Lanka. As it is a multifactorial disease, the aetiology is heterogeneous, therefore differences in morbidity and mortality were observed among different populations, ethnic groups etc. Germ line mutations of cancer suppressor genes such as BRCA 1, BRCA 2, or p53 are inherited in autosomal dominant pattern and express as breast cancer associated syndromes which are usually expressed before 50 years. Also reproductive and menstrual characteristics may influence the risk of having breast cancer. Therefore family history and exposure to endogenous and exogenous oestrogen is considered as main risk factors for breast cancer. Publications on monogenic susceptibility and influence of hormonal risk factors on breast cancer among Sri Lankan populations are scarce.

Therefore the present study was focused on describing the monogenic susceptibility of breast cancer and assessing the influence of hormonal risk factors on breast cancer.

A hospital based population of Sinhalese patients who were histo-pathologically confirmed of having breast cancer, was studied at oncology unit, general hospital Kandy, Sri Lanka during year 2003-2004.

Study sample included 276 females with breast cancer. They were diagnosed of having breast cancer during the period 1998-2004. All affected relatives and index cases were females. Site specific breast cancer, breast and ovarian cancer and breast cancer associated with a sarcoma were observed among them. Autosomal dominant inheritance with female preponderance was observed among the pedigrees with early onset site specific breast cancer. Bilateral breast cancer and multiple primary cancers were rare. Age specific expression of breast cancer indicated an early onset disease among majority of sporadic breast cancer patients. A significant risk of having breast cancer was associated with nulliparity although menstrual and reproductive factors and use of hormonal contraceptives conferred a lower risk to majority with sporadic breast cancer.

Monogenic susceptibility was identified in 5-6% of patients with breast cancer. In the present study and these may be explained by mutations of BRCA1 BRCA 2 and P53 genes. Shared genetic and environmental factors may have contributed to the clustering of breast cancer in few pedigrees. Early onset of sporadic breast cancer necessitates the study of receptor status and mutation analysis. Menstrual and reproductive factors and use of hormonal contraceptives do not confer a high risk to majority of the sporadic cancers although nulliparity conferred a significantly high risk for some patients.

