Breast cancer gene 1 (brca 1) mutation

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Breast cancer is the most common cancer among women in industrialised countries ,it is the leading cause of cancer death (.Approximately one in ten women will develop breast cancer during her lifetime. Currently more than 4000 new breast cancer cases are diagnosed yearly and the number is increasing,Many risk factors (hormonal, environmental) for breast cancer are known but most of the genetic background and molecular mechanisms still remain to be elucidated. family history being the most important factor for determining breast cancer risk .In this study, we aimed at detection of mutation in BRCA1 gene its relation with family history in Female patients attending surgery Department, Benha University Hospital. All subjects included in this study subjected to the following: Full history taking and clinical examination. For breast cancer group:• Radiological investigations: Soft tissue mammography.• Breast tissue Biopsy. Blood samples were collected placed immediately into EDTA tubes. After centrifugation at 3500 rpm, plasma samples were collected and stored at – 70oC until further processing . Genomic DNA will be extracted from whole blood DNA concentration in extracted samples using the UV spectrophotometer (absorbance at 260 nm).Our results suggested that high frequency of exon 11 (5382insc) , exon 5, Low frequency of exon 2 (185del).Identification of BRCA1 and BRCA2 mutation carriers is an important focus in prevention and early detection of breast and ovarian cancer risk Diagnosing breast cancer in younger women (under 40 years old) is more difficult because their breast tissue is generally more dense than the breast tissue in older women. By the time a lump in a younger woman’s breast can be felt, the cancer often is advanced.A woman’s risk of breast cancer is higher if her mother, sister, or daughter had breast cancer. The risk is higher if her family member got breast cancer before age 40. Having other relatives with breast cancer (in either her mother’s or father’s family) may also increase risk.CONCLUSIONSWe concluded from this study that high mutation frequency of exon 11 (5382insc) , exon 5 of brca1 gene and Low mutation frequency of exon 2 (185del) .That seems apparent from the study’s finding that women with a strong family history were still at higher risk for developing the disease.