

Evaluation of Brca1/2 Mutational Status among Algerian Familial Breast Cancer

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Abstract : breast and ovarian cancer are respectively the first and fourth leading causes of cancer among women in Algeria. A family story of cancer in the most important risk factor, and in most cases of families with breast and /or ovarian cancer, the pattern of cancer family can be attributed to mutation in BRCA1/2 genes. objectives: the aim of our study is to investigate the spectrum of BRCA1/2 germline mutation in familial breast and /or ovarian cancer and to determine the prevalence and the nature of BRCA1/2 mutation in Algeria. methods: we determined the prevalence of BRCA1/2 mutation within a cohort of 161 probands selected according to the Kaiser-Moser score. double stranded sanger sequencing of all coding exons of BRCA1/2 including flanking intronic region were performed. results: we identified a total of 23 distinct deleterious mutations (class 5) 12 different mutations in BRCA1 (52%) and 11 in BRCA2 (48%). 78% (18/23) were protein truncating and 22% (5/23) missense mutations. 3 novel deleterious mutations have been identified, which have not been described in public mutation database. one new mutation was found in two unrelated patients. the overall mutation detection rate in our study is 28,5% (46/161). moreover, an UVS c7783 located in BRCA2 is found in two unrelated probands and segregates in the 02 families/ conclusion: our results suggest a large spectrum of BRCA1/2 mutation in Algerian breast/ovarian cancer family. The nature and prevalence of BRCA1/2 mutation in Algerian families are ongoing in a larger study, 80 probands are today under investigation. This study which may therefore identify the genetic particularity of Algerian breast /ovarian cancer.

Keywords : BRCA1/2 mutations, hereditary breast cancer, Algerian women, prevalence

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