عنوان مقاله:

The germline Pathogenic Variants in a Patient with Familial Squamous Cell Carcinoma of Breast

محل انتشار:

چهاردهمین کنگره بین المللی سرطان پستان (سال: 1397)

تعداد صفحات اصل مقاله: 1

نویسندگان:

Mina Amin - *Department of biotechnology, faculty of Chemistry , university of Kashan, kashan ,Iran*

Mehrdad Zeinalian - *Department of Genetics and Molecular Biology, School of Medicine, Isfahan University of Medical Sciences, Isfahan, Iran*

Sina Narrei - *Ala Cancer Prevention and Control Center (MACSA), Isfahan, Iran*

Elaheh Mahmoodi khaledi - *Department of biotechnology, faculty of Chemistry , university of Kashan, kashan ,Iran*

خلاصه مقاله:

Introduction & Aim: Squamous cell carcinomas (SCCs) represent the most frequent human solid tumors as a major cause of cancer mortality. Squamous cell carcinoma in the breast is a very rare disease that it is not widely studied. The estrogen and progesterone receptors are not typically expressed in these cancers, so the endocrine therapy is not effective on them. The deleterious mutations in genes like TP63, TP53, EP300, RECQL4 could lead to different types of SCC. Recognition of involved genetic factors in this disease can lead to finding of its molecular mechanisms and identifying the specific predisposing biomarkers for screening and early detection.Methods: In this study we selected a person with SCC of breast and positive family history for SCC. We tried to identify the novel pathogenic variants for familial SCC by next-generation sequencing (whole exome) method. Results & Conclusion :We identified two heterozygous pathogenic variants in EP300(c.3143-4del T), RECQL4(c.3104-3105 ins A) genes that they may play an important role in initiating and progressing of the disease according to their function in other types of SCC. .Further cases or the population based studies are recommended to confirm the pathogenicity of the noted variants

کلمات کلیدی: