

عنوان مقاله:

In Silico Approach for Prediction of the Structural and Functional Impacts of ۱۸۴(T>C) Missense Mutation Identified in BRCA1 Gene in a Syrian Breast Cancer Patient

محل انتشار:

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خلاصه مقاله:

Background: The classification of genetic variations depending on their clinical impacts is highly relevant for clinical decision-making. Therefore, predicting the effects of missense mutations using in silico tools has become a frequently employed approach. The objective of this study was to analyze the impacts of a previously detected BRCA1 missense mutation using an in silico prediction tool in the context of invasive breast cancer. **Methods:** In this bioinformatics study, application of the in silico combination of tools Phyre2 to ۱۸۴(T>C), a BRCA1 missense mutation previously characterized by Khalil et al. in ۲۰۱۸, we used to predict its clinical impacts. **Results:** Incidence of the missense mutation caused a disorder in the zinc binding RING finger functional domain of BRCA1 protein. This incidence was considered to be a major contributor to the interaction of this protein with other proteins in signal transduction pathway in the mechanism of cellular response to DNA damage. The functional analysis also revealed that the detected missense mutation might significantly affect the function of both the protein and phenotype of the living organism. **Conclusion:** In silico prediction confirmed the detrimental impact of the identified missense mutation in the exon ۲ of BRCA1 gene on both the structural and functional properties of the generated BRCA1 protein.

کلمات کلیدی:

In silico, Missense, ۱۸۴(T>C), Syria, Phyre2

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