عنوان مقاله:

Analysis of pathogenic SNPs of NOTCH۱ gene using bioinformatics tools

محل انتشار:

کنفرانس بین المللی ژنتیک و ژنومیکس انسانی (سال: 1400)

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خلاصه مقاله:

Backgrounds: T-cell acute lymphoblastic leukemia (T-ALL) is an aggressive cancer that affectschildren and adults. More than ۵۰% of T-ALLs is caused by activating mutations in NOTCHsignaling pathway, and this has made NOTCH۱ the most prominent T-ALL specific oncogene.NOTCH۱ gene encodes a Class-I membrane receptor that is crucial for differentiation ofprogenitor pluripotent cells to committed T-cells. NOTCH۱ receptor is a ligand-activatedtranscription factor and so it directly transmits the information of extracellular signals to thenucleus and changes the gene expression. Also, this protein has two subunits of NEC and NTMthat ligands bind to NEC subunit.Materials and Methods: Information about nucleic acid and amino acid sequences of NOTCH۱were taken from NCBI database. From variation viewer section of this database and limiting theresults to pathogenic and missense mutations, ۱۱ cases of SNPs were obtained. Using databasessuch as PATHER, POLYPHEN, PhD SNP, PROVEAN and SNPs&GO, the pathogenesis ofSNPs was investigated. The effect of SNPs on protein stability was also investigated by I-Mutantdatabase.Results: According to the results, the SNPs were predicted by ۴ databases as pathogenicmutations and were identified by one database as neutral. The results of the I-Mutant databasealso showed a decrease in protein stability due to mutations.Conclusion: In this study, the effect of ۱۱ cases of SNPs on NOTCH۱ protein was investigatedand it was found that these SNPs were pathogenic and reduced protein stability. So, it can be saidthat .these SNPs are directly related to the incidence of T-ALL in children

کلمات کلیدی:

NOTCH۱, SNP, Bioinformatics

لینک ثابت مقاله در پایگاه سیویلیکا: