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Computational Analysis Revealed Five Novel Mutations in Human IL2RG gene Related to X-SCID

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Abstract

Background: X-linked severe combined immunodeficiency is a rare life-threatening disorder that presents in early infancy with severe recurrent opportunistic infection.

Purpose: to use bioinformatics tools for identifying effects of possible pathogenic Single Nucleotide Polymorphisms (SNPs) on structure and function of IL2RG gene as a cause of X-linked SCID.

Method: Data on the IL2RG gene was gathered from the dbSNP/NCBI database. Prediction of damaging effect was done using bioinformatics software: SIFT, Polyphen, Provean, SNAP2, SNP&GO, PHD-SNP.P-Mut, I- mutant, Project HOPE, PolymiRTS and GeneMAINIA. The effects of the mutations on the 3D structure of the human IL2RG protein were predicted using RaptorX and visualized by Chimera.

Result: In-silico prediction identifying 1479 SNPs within the IL2RG gene, 253 out of them presented at the coding region and 50 SNPs took place in the miRNA 3'UTR while 21 occurred in 5'UTR region and 921 occurred in intronic regions. Using bio informatics tools, 12 nsSNPs damaging IL2RG coding region were found. Five nsSNPs were identified as novel SNPs including: G305R, C182Y, G114D, Y105C and Y91C. Moreover, 3'UTR region analysis showed two SNPs out of 50 (rs144075871 and rs191726889) were predicted to disrupt miRNAs binding sites which affect gene expression.

Conclusions: This study revealed 5 novel nsSNPs in the IL2RG gene by using different software and two SNPs disrupting miRNAs binding sites in 3'UTR region. These SNPs might be considered as an important factor in causing diseases related to IL2RG gene mutation and could be used in the diagnosis of X-linked SCID.