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In silico analysis of single nucleotide polymorphism in *FBN1* gene and study of genetic interaction

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Marfan syndrome is a common autosomal dominant hereditary connective tissue disorder with variable presentations, mutations in *FBN1* gene were found to be responsible for Marfan syndrome and other relate connective tissue disorders. SNPs contributes to gene mutations and expression variations justifying phenotypic variations among patients and hence such SNPs would be potential target for identification and analysis which may help in early diagnosis of such life threatening disorder. Computational methods were used on this work focusing on analysis of SNPs in the coding regions of *FBN1* gene found as non-synonymous variants (ns-SNP) and those in the 3'un-translated regions (3'UTR) affecting miRNA binding using computational methods including SIFT and polyphen for analysis of (nsSNPs) while (3'UTR) SNPs was analyzed using PolymiRTS tool functions and Interactions of *FBN1* gene with functional similar genes was predicted using gene MANIA software. Out of 1134 ns-SNPs analyzed, 38 SNPs were found to damaging while analysis of 175 SNP in 3'UTR prove that 24 SNPs are disturbing to their target sites and 46 SNPs are creating to new target sites. Using the damaging ns-SNPs predicted on this work may be helpful in early diagnosis and on screening of *FBN1* related disorders.

Biography

Mohanad Khalid Ali Abdelrahim has completed his MBBS from University of Khartoum Faculty of Medicine in April 2015 and started working as teaching assistant at University of Khartoum Department of Human Anatomy on May 2015. He is a student of Master's of Human Anatomy at faculty of medicine University of Elneelain. He is also member of Daoud Research Group.

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