Refinement of SNP Mutations of Atopic Dermatitis Related Filaggrin Through Existing R Packages

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In the US alone, there are currently 17.8 million affected by Atopic Dermatitis (AD), commonly known as Eczema. It is characterized by itching and skin inflammation. AD patients are at higher risk for infections, depression, cancer, and suicide. Genetics, environment, and stress are some of the primary causes. With the rise of personalized medicine and the acceptance of gene editing technologies, as evidenced by the 2020 Nobel Prize in chemistry for CRISPR, AD - related mutations need to be identified for treatment. Genome Wide Analysis Studies (GWAS) have associated the FLG (Filaggrin) gene with AD, but not identified specific problematic mutations. This research aims to refine known Single Nucleotide Polymorphisms of FLG for gene editing technologies to target. The research utilizes the R language and its Bioconductor packages to refine data from the National Center for Biotechnology Information's (NCBI's) Variation Viewer. The research filtered by coding regions and conserved domains. It also removed synonymous variations and treated non-synonymous, frameshift, and nonsense mutations separately. In the non-synonymous mutations, the mutations were refined and ordered by the BLOSUM62 substitution matrix. Overall, the project removed 96.65% of data, which was redundant, and ordered the remaining, relevant data, by impact. Currently, the research is identifying mutations in phosphorylation sites and will soon test the impact of multiple mutations and genes together. The code for the project can also be repurposed as a tool for other diseases. This project will help solve GWAS's imprecise identification challenge. It is the first step in providing the refined databases required for gene-editing treatment.

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