

## The bioinformatical analysis Artemis defected protein in a subset of SCID patients

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**Abstract:** Artemis is a protein that in humans is encoded by the DCLRE1C (DNA cross-link repair 1C) gene. This protein is important for V(D)J recombination of the immunoglobulin and T-cell receptor genes. The mutations in DCLRE1C caused Immunodeficiency and it have variable clinical presentations including severe combined immunodeficiency (SCID) [1]. At this present to investigate of gene network and matrix families throughout the promoter region of DCLRE1C gene, GeneMANIA, PFAM and Gene2promoter softwares were used and to study evolutionary relationship of DCLRE1C in human compared to other organisms, ClustalX, GENEDOC and Treedraw softwares, respectively [2]. Regarding to phylogenetic tree achieved by analysis of DCLRE1C gene, Homo sapiens has the highest and lowest similarity to *Gorilla gorilla* and *Galeopterus variegatus*, respectively. This result mentions the importance and conservation of sequences in Evolutionary process. In addition to study physical interaction showed 7 proteins that the most significant of them was included, ATM, RAD51, MRE11 that for example a novel role for RAD51 was identified in innate immune. further more study about co-expression genes showed 5 key gene that most importance was C17orf62, that's Ubiquitous expression in spleen, lymph node and thyroid (main tissues for immune response). Analysis of DCLRE1C gene promoter showed the the presence of 27 matrix families ( $P < 0.05$ ). Among the key matrix families can be point out to ETSF (promoter region specific for protein transporters and effective in T cell activation) and RXRF (retinoid X receptor as obligate partners forms heterodimeric and thyroid-hormone receptors) and DUX4 (its binds and activates LTR elements from a class of MaLR endogenous primate retrotransposons and suppresses the innate immune response to viral infection) and NR2 (Nuclear receptor subfamily 2 factors, The first finger controls specificity of DNA-binding, the second finger controls specificity of dimerization [3]. Totally, obtained results emphasize complexity and functional importance of highly conserved Artemis protein. These genetic studies provide a novel approach for treating SCID patients by using gene therapy.

**Key words:** Artemis; SCID; gene network; phylogenetic; promoter

### References:

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