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Analysis of DNA/RNA sequence changes in 20 genes involved in the cell cycle in a Colombian population

Elizabeth Vargas, Oscar Ortega Recalde, Ludis Morales and Maria Martinez-Aguero
Universidad del Rosario, Colombia

Post-transcriptional RNA editing is a mechanism that provides high protein and phenotypic diversity in all organisms. Different systems have been identified in multicellular eukaryotes and its main form is pre-mRNA editing. Also, other editing cellular mechanisms are known in mammals. Among the most studied is the change of A-to-I, catalyzed by ADAR (adenosine deaminase acting on RNA) protein family, which is an adenosine (A) is modified in the pre-mRNA as inosine (I). This change is recognized by the translation machinery as a guanosine (G). Other editing processes have not been deciphered yet and RDD (RNA/DNA differences) suggest unknown aspects of the transcription or post-transcriptional modifications of RNA. The aim of the project was to identify RDD changes presented in two kinds of genes (proto-oncogenes and tumor suppressor genes) in a healthy population of 16 women living in Bogota. Next generation sequencing was performed on 20 genes. They were included in the analysis only the SNP variants with sequencing depth greater than 10X. As a final result, 6 RDD variants were found in 3 genes, *AKT1*, *ATM* and *P53*, in 8 of the 16 individuals. Of these variants, 33% are transversion (C-to-A, C-to-G) and 67% transition (A-to-G and C-to-T). This new paradigm represents an important unexplored aspect of the variation of eukaryotic genomes. RNA editing as mediator of the variation of organisms is a little explored area and there are no reports of the frequencies of these changes in human populations. This type of edition could explain many critical processes in cell regulation and in the future, could be an attractive option for therapeutic intervention in cancer and other diseases.

Biography

Elizabeth Vargas has completed her Master's degree in Biological Sciences at the Pontificia Universidad Javeriana. She is currently working at the Faculty of Natural Sciences and Mathematics at the Universidad del Rosario and has been involved as a Researcher in several projects in cell and molecular biology.

lizvargas211@hotmail.com

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