Baris E. Suzek, Transcriptomics, 2:2
http://dx.doi.org/10.4172/2329-8936.S1.002

OMICS International
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International Conference on
Transcriptomics

July 27-29, 2015 Orlando, USA

Locating disease-associated single amino acid polymorphisms on proteins

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The next generation sequencing technologies are continuously improving and large sets of human mutations are becoming available within institutions and through public resources. A subset of these mutations lead to single amino acid polymorphisms (SAPs). Identifying SAPs' disease-causing impact and developing methods to identify SAPs' disease-associations are active research areas. In this work, we investigated the link between disease-associated SAPs and their protein sequence context (i.e. sequence features). We have integrated disease-associated SAPs and sequence features from several public bioinformatics resources. The SAPs and sequence features are then mapped to each other using protein resource UniProt Knowledgebase. Based on our initial results, over 39% of disease-associated SAPs are located on functional domains, topological domains and near post translational modifications. Over 19% of them are located on known secondary structures. Our findings will help in better understanding of SAPs' disease associations. In the future, the protein sequence context for SAPs can be utilized in development of more accurate computational methods to predict disease-associated SAPs.

Biography

Baris E. Suzek has completed MS in Computer Science at Johns Hopkins University and PhD in Bioinformatics and Computational Biology at George Mason University. Currently, he is an Assistant Professor in Computer Engineering Department at Mugla Sitki Kocman University in Turkey and an Adjunct Assistant Professor at Georgetown University in USA. Before 2013, he served as a Research Assistant Professor in Department of Biohemistry and Bioinformatics Team Co-lead in Protein Information Resource at Georgetown University. He has published more than 20 papers in reputed journals, holds one patent and is recipient of several national awards in USA.

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