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Structure-based modeling of the effects of missense mutations associated with human disorders

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Human DNA sequence differs among individuals and the most common variations are known as single nucleotide polymorphisms, or SNPs. Studies have shown that non-synonymous coding SNPs (nsSNPs - SNPs occurring in protein coding regions which lead to amino acid substitutions) can be responsible for many human diseases or cause the natural differences among the individuals by affecting the structure, function, interactions and other properties of expressed proteins. Of particular interest are mono-genetic diseases resulting from missense mutations affecting the wild type characteristics of a specific protein. Using various cases of disorders, it is demonstrated that almost always the mutations do not directly affect the functional properties of the corresponding protein, but rather indirectly alter its wild type characteristics. This provides an opportunity the disease-causing effect to be tackled with small molecule binding. In addition, it is demonstrated that disease-causing mutations do not necessary destabilize protein stability or protein-protein interactions, but can be stabilizing and still be harmful. Overall, a detailed computational analysis combined with an analysis of the corresponding biological function is needed to make reasonable prediction of the disease association of missense mutations.

Biography

Emil Alexov has received MS in Physics from Sofia University, Bulgaria in 1983 and PhD in Physics in 1990. Since then he got various positions in academia and in research institutions as Sofia University, Bulgaria; RIKEN Institute, Japan; City College of New York and Columbia University, New York. Since 2005 he moved to Clemson University, SC where he currently is Professor of Physics. His lab work on various projects the main being maintenance and further development of DelPhi and modeling effects of human DNA variations with respect to human diseases. He co-authored more than hundred peer reviewed papers, serves on several Editorial Boards and reviews for various journals. He has co-organized and will co-organize scientific meeting including the Gordon Research Conference of "Human Single Nucleotide Polymorphism and Disease" August 3-8, 2014.

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