GENETIC VARIATIONS: ORIGIN, EFFECTS AND PREDICTION

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Each individual has millions of genetic variations in their genome. This information can now be efficiently revealed with next generation sequencing techniques. As some variations are associated with diseases it would be important to be able to identify tehm. Due to the large amount of data this is not possible with experimental methods. Computational approaches are needed to study and analyze variations and to prioritize likely pathogenic or neutral cases. The origin of genetic variations and mechanisms behind them will be discussed. These concepts are needed to be able to systematically classify variations and their effects. Variation Ontology (VariO) is developed to facilitate systematic variation effect description. Many existing methods are devoted to recognizing harmful variations. This is an important prediction task, which can be extended to study of consequences and effects of harmful variants. Performance of certain prediction tools will be discussed based on large scale tests of experimentally verified cases. Our experiences from the use and development of prediction tools as well as variation database curation and development will be discussed with view for the future.