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Value of predictive bioinformatics in inherited metabolic diseases

David J Timson

Abstract

Typically, inherited metabolic diseases arise from point mutations in genes encoding metabolic enzymes. Although some of these mutations directly affect amino acid residues in the active sites of these enzymes, the majority do not. It is now well accepted that the majority of these disease-associated mutations exert their effects through alteration of protein stability, which causes a reduction in enzymatic activity. This finding suggests a way to predict the severity of newly discovered mutations. *In silico* prediction of the effects of amino acid sequence alterations on protein stability often correlates with disease severity. However, no stability prediction tool is perfect and, in general, better results are obtained if the predictions from a variety of tools are combined and then interpreted. In addition to predicted alterations to stability, the degree of conservation of a particular residue can also be

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