Modelling Differences in Protein Interactions Caused by Familial Hypercholesterolemia for Personalised Medicine

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Genetic disorders are very common in the general population. These disorders are caused by abnormalities in one's genome (DNA), which in turn encodes the proteins that are expressed in one's body. To predict the changes the mutations cause, computational methods are used to approximate the protein structure and interactions. As a case study, the research looks at the common genetic disorder familial hypercholesterolemia and uses an algorithmic model on a mutation known to have a link to the disorder. A hypothesis is made about how this mutation might cause the symptoms (specifically LDL-cholesterol accumulating in the blood) as the mutated protein's ability to bind an inhibitor increases. The method may be used to explore how mutations changes interactions within the body, and how different mutations should be treated differently, even though they cause the same general symptoms.