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Disease specific databases for personalized medicine: Fabry disease as a case of study

Next generation sequencing of all exons will become common for symptomless people in the near future. Since we all have approximately 20000 variants, differentiation among non-pathological, mild or deleterious mutations will be necessary. We will show how disease specific databases and predictive tools can be precious for personalized diagnosis and therapy. The analysis of sequence conservation among orthologous proteins, even in the absence of structural information on the human protein can be sufficient to identify severely pathological mutations, but training sets represent the weak point in the development of prediction tools. At present, more than 70000 missense mutations have been reported with seven variants per protein on average but at least 70 cases more than 100 variants are known. A quantitative phenotype can be associated to mutants measuring the residual activity or the stability of proteins expressed by transient transfection. In this case, it can be possible to predict the severity of the mutation quantitatively. This implies that in many cases it is possible to develop disease specific predictive tools. Lysosomal alpha galactosidase which is associated to Fabry disease represents a good case to test the effectiveness of specific tools that allow predictions scored according to severity. More than 380 missense mutations are known and for 305, the residual activity in cells has been assessed. For the deficiency of lysosomal alpha galactosidase, disease specific predictive tools can also be exploited to estimate responsiveness to specific drugs such as pharmacological chaperones.

Biography

Maria Vittoria Cubellis completed her graduation in Chemistry from the University of Naples and PhD in Biochemistry. She is a Professor of Biochemistry and Bioinformatics at the University of Naples Federico II. Her recent research interests are focused on “Pharmacological chaperones for the treatment of rare diseases and in particular on the prediction of mutations associated to Fabry disease which are responsive to drugs”. She is also trying to extend the approach with pharmacological chaperones to other pathologies, such as disorder of glycosylation type 1a, a disease for which there is no cure at present.

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