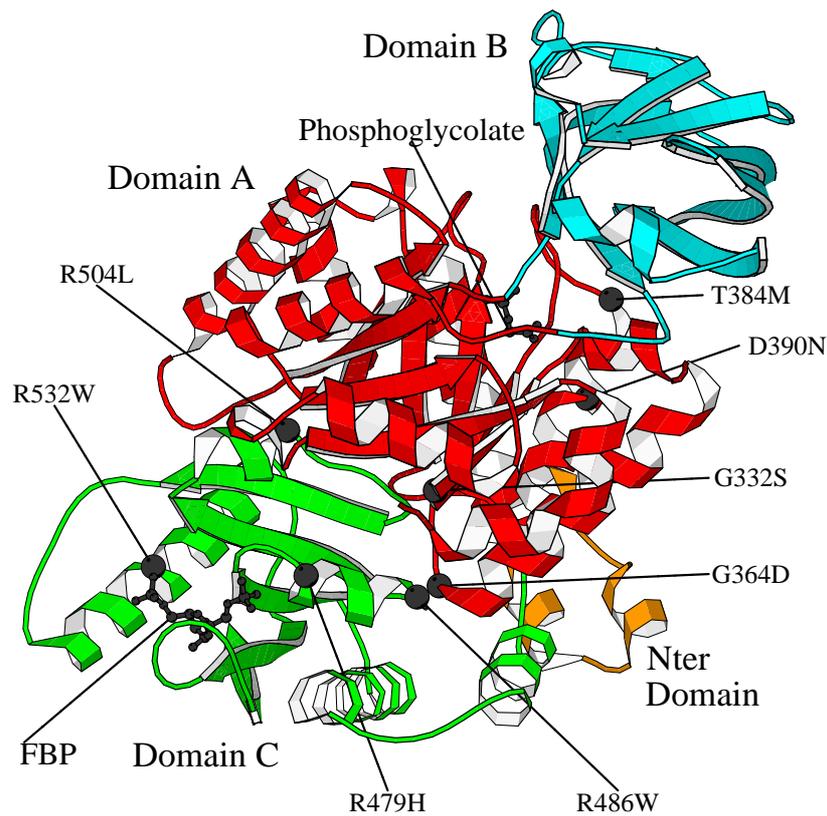


clinical symptoms with the molecular properties of the mutant enzymes. Mutations greatly impairing thermostability and/or activity are associated to severe anemia. Some mutant proteins exhibit moderate changes in the kinetic parameters, which are sufficient to cause mild-to-severe anemia, underlining the crucial role of RPK for erythrocyte metabolism.

Conclusions - Prediction of the effects of mutations is difficult since there is no relation between the nature and location of the replaced amino acid and the type of molecular perturbation. Characterization of mutant proteins may serve as a valuable tool to assist with diagnosis and genetic counseling.



Reference

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