Abstract

Urate level in human bloodstream is influenced by purins degradation in organism and its elimination. Enviromental factors are important for this balance, but heredity has a key role. Disbalance of processes increase the risk of deseases caused by patalogical blood urate levels.

Healthy people have the most of generated urate reabsorbed and returned back to bloodstream *via* proximal tubules of the kidneys. The various membrane transporters participate on urate handling in human kidneys. Their intact function is necessary for transcellular transport. In its different isoforms, membrane transpoter GLUT9 (*SLC2A9*) is located on the apical and basolateral membrane of proximal tubule cells. His function in transmission of urate was detected just recently. GLUT9 is probably the one of most important proteins from regulators of blood urate levels in humans.

We have an extensive set of individuals with different serum urate levels and *SLC2A9* gene polymorfism from the Czech population. In this thesis there are results from the expression of human GLUT9 allelic variants performed on *Xenopus laevis* oocytes and functional studies of these proteins uptaking [14C] radiolabelled urate.

We have detected significant changes of urate transmission through V169M, D281H, P350L, R294H allelic variants of GLUT9. Simultaneously these variants protein were detected on the plasma membrane Xenopus oocytes.