Abstract

Arterial hypertension is the most prevalent cardiovascular dissease which can lead to various end organ damage. It is most commonly a primary diagnosis or so called essential hypertension with a multifactorial pathogenesis combining genetic and enviromental factors. Its prevalence is increasing with age. On the other hand there are some relatively rare monogenic forms of arterial hypertension, which are caused by a single point mutation of particular gene. These diseases can manifest in childhood and one of them is Liddle syndrome. It is a heritable form of arterial hypertension caused by mutation in gene coding of a epithelial sodium channel. This channel is responsible for sodium reabsorption in distal parts of nephrone in kidney. Mutated channel is overactive which leads to increased retention of sodium and therefore water, resulting in exapansion of intravascular volume and hypertension. Along with high blood pressure hypokalaemia and low serum aldosteron occurs is some patients as well. These disturbances can be useful leads in work-up and together with positive family history of high blood pressure may guide a physician to start thinking about more rare forms of hypertension. Correct diagnosis enables to chose the right treatment which, in case of Liddle syndrome, is epithelial channel blocker.

This work summarizes existing knowledge about epithelial sodium channel in organism under physiological conditions and its role in developement of arterial hypertension. Furthermore it elaborates own examination of a family with Liddle syndrome in which new mutation of epithelial sodium channel was discovered. There is great variability in disease phenotype between the family members which is further analyzed.

Key words: aldosterone, amiloride, arterial hypertension, epithelial sodium channel, hypokalaemia, Liddle syndrome