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综述

上皮细胞钠通道ENaC及其基因研究现状

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摘 要 人类上皮细胞钠通道(hENaC)由α、β、γ三个亚单位组成,分别由SCNN1A、SCNN1B、SCNN1G基因所编码。ENaC负责钠离子的限速重吸收，对于维持钠的自身平衡、细胞外液量和血压起重要作用。功能获得性ENaC基因突变可引起一种罕见的遗传性高血压—liddle综合征；而功能丧失性ENaC基因突变可引起一种遗传性低血压—假性低醛固酮血症；原发性高血压是受遗传因素和环境因素共同影响的复杂性疾病，由于ENaC的维持钠的自身平衡和血压的重要作用，因此ENaC基因作为原发性高血压的候选基因而倍受关注。

关 键 词 上皮细胞钠通道 ； 上皮钠通道基因 ； 高血压

分类号

Present condition of the studys of the epithelial Na channel(ENaC) and its genes

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Abstract The human epithelial Na channel is composed of three subunits: α、β、γ,Which were encoded by SCNN1A、SCNN1B、SCNN1G genes respectively. ENaC is responsible for the rate-limiting step of sodium reabsorption and thus plays an important role in the maintenance of sodium balance, extracellular fluid volume and blood pressure. Gain-of-function mutations in ENaC genes cause an infrequent hereditary hypertension —Liddle syndrome; Loss-of-function mutations in ENaC genes cause an hereditary hypotension—Pseudohypoaldosteronism; Essential hypertension is a kind of disease cooperated by genetic factors and environmental factors, ENaC genes are given more attention as candidate gene of essential hypertension beasease of its important role of maintenance of sodium homeostasis and blood pressure .

Key words The epithelial Na channel ； ENaC gene ； Hypertension

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