

Polymorphisms in healthy individuals

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Abstract

Tight junction is the area between epithelial cells that controls the fluid between cells. It consists of three main groups of proteins, transmembrane proteins claudin and occludin, accessory proteins zonula occludence 1, 2, 3 and cytoskeletal protein Actin. Although their main function is to prevent leakage and diffusion of molecules across epithelial barriers they also contribute to the adherence of cells. In malignant tumors, tight junctions frequently show structural and functional abnormalities. Our aim in this study was to see the frequency of genotypes rs6174821 and rs9869263 in claudin 1, CLDN1 and rs4562, rs3744400 of claudin 7, CLDN7 among the healthy population in Sweden. 265 samples were collected from blood and plasma donors. DNA was extracted and mutational screening for CLDN 1 SNP rs6174821 and rs9869263 and CLDN 7 SNP rs4562 and rs3744400 was performed using pyrosequencing. The Hardy-Weinberg formula was used to calculate the genotype. The results show that there is a difference in the frequency of polymorphisms between men and women for several of the genotypes as well as differences compared to HapMap. In CLDN 1 SNP rs9869263 for men and women together the frequency for TT was 0.1959 for TC 0.385 and for CC 0.593 while the HapMap frequency for both men and women was for TT 0.035, for TC 0.230 and for CC 0.735. CLDN 7 SNP rs3744400 for men and women together genotype TT 1.931 and for TG 0.066, whereas GG were 0. When we compare to HapMap frequency for both men and women genotype TT 0.991, TG 0.009 and GG were 0. For men and women together the frequency for SNP rs4562 CC was 0.71, for TT 0.323 and for TC 0.97. The HapMap frequency for men and women was 0.292 for CC, 0.185 for TT and 0.523 for TC.

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Biography

Victoria Hahn-Stromberg is a versatile writer and researcher that published extensively in Hematopathology.

She is functioning as Associate Professor in the Department of Medical Cell Biology at Uppsala University.