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Title: Matrix metalloproteinases gene polymorphisms in the pathogenesis of primary spontaneous pneumothorax

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Body: Primary spontaneous pneumothorax (SP) is one of the common thoracic surgical conditions requiring hospital admission. Screening and prevention SP have not been developed so far, because the lung bullae pathogenesis is not known. One of the possible directions is the study of matrix metalloproteinases (MMP) that provide the catabolism of the extracellular matrix components of connective tissue. DNA samples from 148 patients with primary SP and 152 volunteers were studied. The frequencies of genes polymorphisms MMP1 (1607insG), MMP9 (C-1562T), MMP12 (A-82G), TIMP1 (S536T) were identified. With sample isolated DNA was performed two parallel amplification reaction with two pairs of allele-specific primers. For the analysis of mixtures of restriction applied by horizontal electrophoresis in 3% agarose gel. For gene MMP1 (-1607insG) reported a significant decrease in the frequency of normal homozygous genotype G/G, which was associated with a lower risk of the primary SP (OR=0,27). GG allele carrier status was significantly more frequent in patients with primary SP and was associated with a twofold increase in risk (OR=2,27). In patients with primary SP significantly rarer normal genotype C/C gene MMP9. Heterozygous genotype C/T is dominant and met with primary SP twice as often. Predisposition to primary SP associated with carriage of the heterozygous genotype C/T (OR=2,43), the homozygous genotype T/T (OR=4,38) and T allele (OR=2,78). For polymorphic loci and genes MMP12 and TIMP1 were no statistically significant difference between groups. Our data confirm that mutant alleles of polymorphic loci MMP genes may be responsible for increased risk of SP.