# Role and diagnostic value of gene variants in assessing the risk of chronic obstructive pulmonary disease

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**ABSTRACT.** Meta-analyses have revealed many positive associations between gene variants and susceptibility to chronic obstructive pulmonary disease (COPD). However, some of those positive results may be false positives. Therefore, we investigated the genetic polymorphisms associated with COPD risk and determined their diagnostic value. We extracted the odds ratio (OR) and 95% confidence interval for each polymorphism from published meta-analyses concerning gene variants and COPD susceptibility in October 2014, subsequently we calculated false-positive report probabilities (FPRPs) for statistically significant associations (P value < 0.05). We determined the diagnostic value of the true positive polymorphisms of COPD using the Meta-DiSc software. Twenty-five gene polymorphisms were significantly associated with COPD risk. The FPRP test results were as follows: 1) when the

 

prior probability was 0.001 and the OR was 1.5, *ADAM33* rs612709, *CHRNA3/5* rs1051730, *CHRNA3/5* rs8034191, *CHRNA3/5* rs16969968, and *TGFB1* rs1800470 were truly associated with COPD risk (FPRP < 0.2); 2) when the prior probability was 0.000001 and the OR was 1.5, all the variants except *TGFB1* rs1800470 remained noteworthy; and 3) when the probability was 0.000001 and the OR was 1.2, *ADAM33* rs612709 and *CHRNA3/5* rs1051730 remained true positives. Unfortunately, the results of the diagnostic accuracy meta-analyses suggested that none of the variants had high value for COPD diagnosis.

**Key words:** COPD; Polymorphism; Susceptibility; FPRP

 