Exploration of association between EPHX1 and chronic obstructive pulmonary disease on the basis of combined data mining

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ABSTRACT. Chronic obstructive pulmonary disease (COPD) is an important respiratory disease with high mortality. Although smoking is the major environmental risk factor for the development of COPD, only 10% of heavy smokers develop symptomatic disease, suggesting association between genetic susceptibilities and environmental influences. In recent years, as one of the most widely studied genes including tests for associations between a genetic variant and COPD, epoxide hydrolase 1 (EPHX1) was found to be involved in the metabolism of tobacco smoke, an important risk factor of COPD. However, genetic associations with COPD identified in studies on EPHX1 are controversial. To address this issue, except for performing the meta-analysis, which specially added our current study on two polymorphisms (T337C and A416G) of EPHX1, we performed
combined data mining based on functional prediction algorithms of nonsynonymous single-nucleotide polymorphisms and gene-based variable threshold testing. Genetic variations in \textit{EPHX1} did not affect COPD in Caucasian and Eastern Asian population, which is supported by recent evidence. We found no association between \textit{EPHX1} and COPD; however, a minor effect of \textit{EPHX1} on COPD risk was not completely excluded; further replication studies with large samples are needed to confirm our findings.

**Key words:** Chronic obstructive pulmonary disease; Meta-analysis; Epoxide hydrolase 1; Nonsynonymous single-nucleotide polymorphisms; Variable threshold