



Relationship between vitamin D (1,25-dihydroxyvitamin D3) receptor gene polymorphisms and primary biliary cirrhosis risk: a meta-analysis

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ABSTRACT. The vitamin D (1,25-dihydroxyvitamin D3) receptor (*VDR*) gene encodes a protein that functions in the transcriptional regulation of vitamin D-responsive genes and plays a role in innate immunity and adaptive immune responses. In this study, we investigated the relationship between *VDR* polymorphisms (*BsmI*, *ApaI*, and *TaqI*) and primary biliary cirrhosis (PBC) risk. We conducted an overall meta-analysis and subgroup meta-analysis based on ethnicity that included a total of 6 eligible studies (672 cases and 1148 controls). We detected no significant PBC risk variation for all genetic models in the overall analysis and in the subgroup analysis based on ethnicity for the *BsmI* polymorphism. For the *ApaI* polymorphism, significant associations were observed in the overall analysis as well as in the Asian subgroup. Furthermore, in the subgroup analysis based on ethnicity, a significant association was observed in the Caucasian subgroup but not in the Asian subgroup for the *TaqI* polymorphism. Based on the results of our meta-analysis, the *VDR BsmI* polymorphism may not be associated

with PBC risk, while the *VDR ApaI* polymorphism is likely associated with PBC risk, particularly in Asians. The *VDR TaqI* polymorphism may be associated with PBC risk in Caucasians.

Key words: Meta-analysis; Polymorphism; Primary biliary cirrhosis risk; *VDR*