The association of rs2277923 polymorphism in *NKX2-5* gene with congenital heart diseases (CHDs): A systematic review and meta-analysis

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Introduction: The *NKX2-5* gene is one of the key transcriptional factors that regulates the normal cardiac development in an embryo. Thus a single nucleotide polymorphism (SNP) rs2277923 can disturb the gene function and ultimately perturb the normal cardiac function and morphogenesis. To date, many studies have been published regarding the rs2277923 polymorphism in the *NKX2-5* gene with CHDs, but the results remained uncertain. Thus, it is the need of the hour to critically analyze all the literature to find the role of rs2277923 SNP in CHDs.

Methodology: We followed the standard PRISMA 2009 guidelines for this study. By using various electronic databases we searched the literature published in the English language till March 30, 2021. Finally, all the relevant information was extracted and analyzed by using the MetaGenyo Program.

Results: In the final analysis, we included 17 studies, and pooled odds ratio was used to calculate the association of selected SNP with CHDs. The pooled odds ratios in different tested genetic models suggested the non-significant association between rs2277923 SNP and congenital heart defects. Moreover, the results showed no publication bias existed in this meta-analysis.

Conclusions: Our results suggested that the association between rs2277923 in the *NKX2-5* gene and CHDs were non- significant. However, in the future more studies with a larger sample size are required that may provide us more definite conclusions.

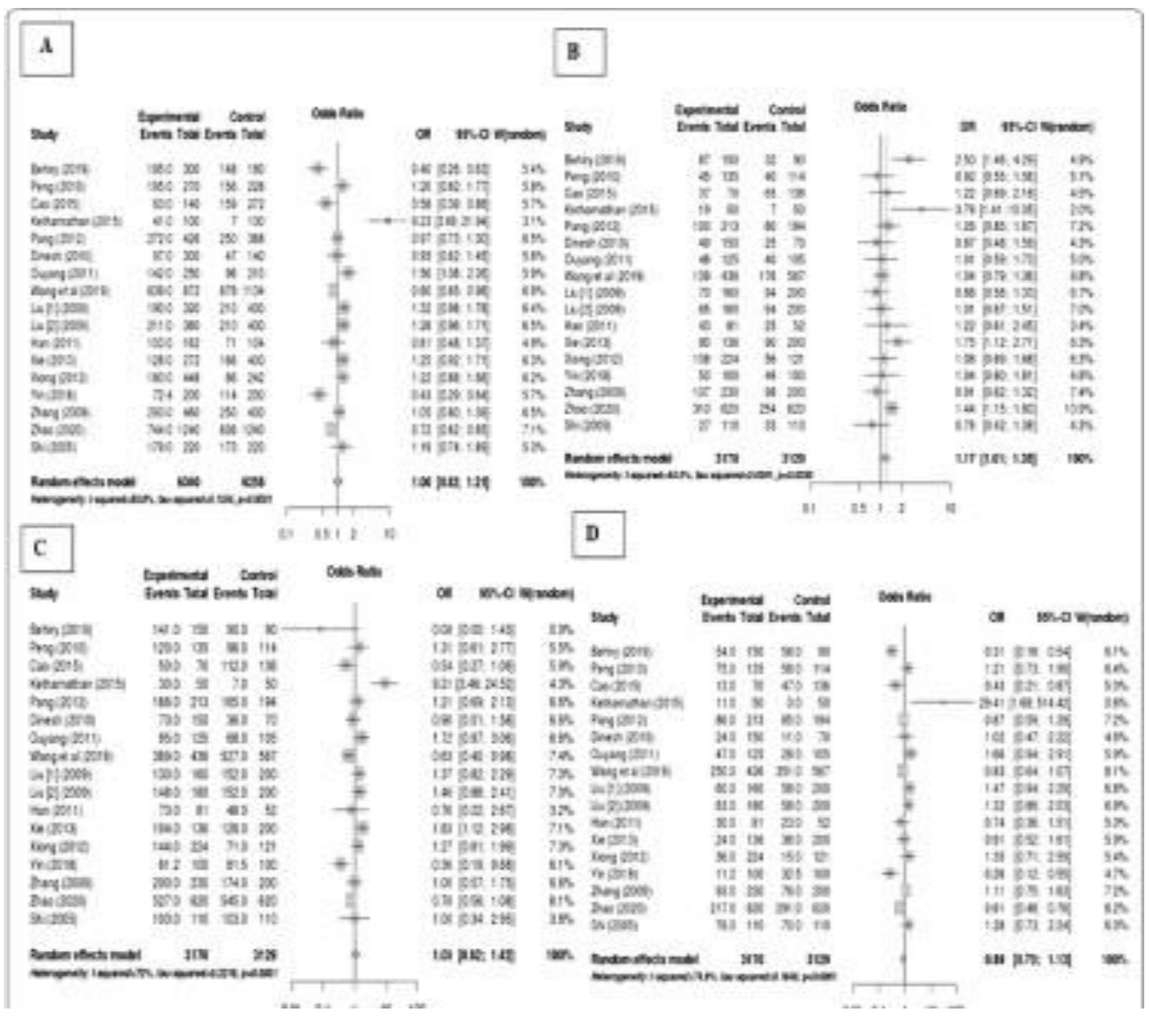
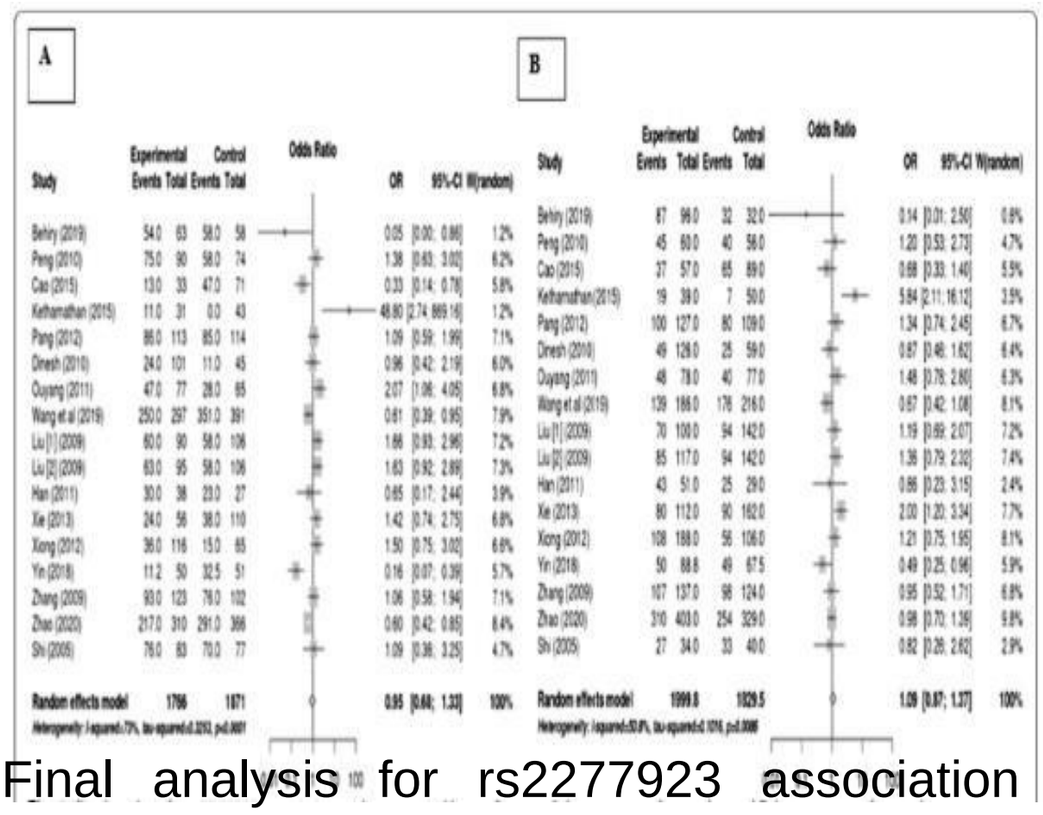


Fig 1. Final data analysis for rs2277923 association with congenital heart defects and the forest plot for different genetic models showing results as a allelic, b over dominant, c dominant, and d recessive, respectively (



Final analysis for rs2277923 association with congenital heart diseases A: homozygote forest plot and B: heterozygote forest plot

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