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Association of Genetic Polymorphisms in STAT 3, STAT 5b and GWAS Identified *PTPN22* Gene with Rheumatic Heart Disease

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Background

Rheumatic heart disease (RHD) is an inflammatory, autoimmune disease, occurring as a consequence of group A streptococcal infection complicated by rheumatic fever (RF). Cytokines are important mediators of inflammatory and immune responses. JAK-STATs have been demonstrated to be critical elements in signaling by certain families of cytokines. GWAS has identified *PTPN22* SNPs as non-HLA genetic variants to be associated with susceptibility to autoimmune diseases. Based on these, we looked for association of genetic variants of *STAT 3*, *STAT 5B* and GWAs identified *PTPN22* with RHD in North Indian population.

Methods and results

This case-control study included 400 RHD patients and 200 controls. The polymorphisms were identified using RFLP/Taqman probes. Statistical analysis was performed by using SPSS. We observed that *STAT3* CG and GG genotypes were significantly associated with RHD (p=0.024 & p=0.027 respectively), *STAT5b* CT&TT genotypes were significantly associated with RHD (p=0.001 & p=0.002 respectively) while both the SNPs of *PTPN22* gene did not show any association with RHD. Further categorization of RHD patients into mitral valve disease (MVD) and combined valve disease (CVD) subgroups revealed that *STAT3* CG&GG genotypes were associated with MVD and *STAT5b* CT&TT genotypes were also associated with both MVD&CVD.

Conclusions

STAT3 & *STAT5b* gene polymorphisms may play an important role in the pathogenesis of RHD but GWAS identified *PTPN22* SNPs may not be associated with susceptibility of RHD.

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