

POSTER PRESENTATION

Open Access

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

Usha Gupta<sup>1\*</sup>, Avshesh Mishra<sup>1</sup>, Saurabh S. Rathore<sup>1</sup>, Snober S Mir<sup>2</sup>, SK Agarwal<sup>3</sup>, Naveen Garg<sup>4</sup>, Balraj Mittal<sup>1</sup>

From International Conference on Human Genetics and 39th Annual Meeting of the Indian Society of Human Genetics (ISHG)  
Ahmadabad, India. 23-25 January 2013

## 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.

## Authors' details

<sup>1</sup>Department of Genetics, Sanjay Gandhi Postgraduate Institute of Medical Sciences (SGPGIMS), Lucknow, UP, India. <sup>2</sup>Department of Biotechnology, Integral University, Lucknow, UP, India. <sup>3</sup>Department of Cardiovascular and Thoracic Surgery, Sanjay Gandhi Postgraduate Institute of Medical Sciences (SGPGIMS), Lucknow, UP, India. <sup>4</sup>Department of Cardiology, Sanjay Gandhi Postgraduate Institute of Medical Sciences (SGPGIMS), Lucknow, UP, India.

Published: 21 January 2014

doi:10.1186/1755-8166-7-S1-P110

Cite this article as: Gupta et al.: Association of Genetic Polymorphisms in *STAT 3*, *STAT 5b* and GWAS Identified *PTPN22* Gene with Rheumatic Heart Disease. *Molecular Cytogenetics* 2014 **7**(Suppl 1):P110.

Submit your next manuscript to BioMed Central  
and take full advantage of:

- Convenient online submission
- Thorough peer review
- No space constraints or color figure charges
- Immediate publication on acceptance
- Inclusion in PubMed, CAS, Scopus and Google Scholar
- Research which is freely available for redistribution

Submit your manuscript at  
[www.biomedcentral.com/submit](http://www.biomedcentral.com/submit)



<sup>1</sup>Department of Genetics, Sanjay Gandhi Postgraduate Institute of Medical Sciences (SGPGIMS), Lucknow, UP, India  
Full list of author information is available at the end of the article