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Rheumatic heart disease: Genetic susceptibility and resistance

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Rheumatic heart disease (RHD) is a complex autoimmune disease caused by repeated infection of Streptococcuspyogenes. RHD is a major public health problem in developing countries like India. Overall prevalence of this disease is about 2/1000 in all age groups, suggests that there are about 2.0 to 2.5 million patients of RHD in the country. The familial clustering and high concordance of RHD among homozygous twins suggests the involvement of genetic factors in RHD.

In order to establish the role of innate and adaptive immune genes in RHD, we have sequenced all exons and UTRs of the following genes: *MASP2, MASP1, FCGR2A, IL23A, FCN2, IL12A, IL12B, IL2, IL4, 1L6, CTLA4, FOXP3, TLR2, TNF-alpha, TNF-beta, IL1-beta, IL1RN, IL17A* and *MBL2* in 87 RHD patients and 25 healthy controls by ion-torrent PGM and Sanger sequencing methods. We found a few variations in MBL2 and MASP2 genes, which are significantly associated with RHD. We have also analysed serum MBL2 level of 58 RHD patients and 29 healthy controls by ELISA and found significantly lower MBL2 concentration in RHD patients compare to healthy control. Interestingly, we found two non- synonymous variation (p.V377A, p=0.0010 and p.Y371D, p=0.0005) in exon 10of MASP2 gene, which are negatively associated with RHD.

A detailed clinical, genetic and biochemical results would be discussed during the presentation.