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Abstract Topic: - Evolutionary and population genetics

Abstract Title: - Association of FOXP3 rs3761548 polymorphism and its reduced expression with Unexplained Recurrent Spontaneous Abortions: A South Indian Study

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Aims: - "The aim of this study is to investigate the association between the FOXP3 rs3761548 polymorphism and the serum concentrations of full-length FOXP3 protein in patients with Unexplained Recurrent Spontaneous Abortions (URSA) in Southern India"

Methods: - The study included blood samples from 150 URSA patients and 150 healthy, pregnant parous women. Polymerase Chain Reaction - Restriction Fragment Length Polymorphism was done for rs3761548 FOXP3 genotyping. Serum concentrations of full-length FOXP3 protein were estimated by Enzyme Linked Immuno Sorbent Assay.

Results: - The frequencies of mutant A allele, CA and AA genotypes of rs3761548 functional polymorphism were significantly elevated in patients compared to healthy, pregnant parous women and exhibited a two, three and two fold increased risk respectively towards URSA. Serum concentrations of full-length FOXP3 protein were high in controls compared to patients (10.14 ± 4.30 vs. 8.84 ± 1.73 ng/ml; $p<0.05$).

Conclusions: - Our results advocate an association of FOXP3 rs3761548 polymorphism and reduced expression of full length FOXP3 protein with URSA.

Keywords: - The frequencies of mutant A allele, CA and AA genotypes of rs3761548 functional polymorphism were significantly elevated in patients compared to healthy, pregnant parous women and exhibited a two, three and two fold increased risk respectively towards URSA. Serum concentrations of full-length FOXP3 protein were high in controls compared to patients (10.14 ± 4.30 vs. 8.84 ± 1.73 ng/ml; $p<0.05$).