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**Abstract Topic:** - Molecular effects of genetic variation

**Abstract Title:** - TP53 p.R72P polymorphism in infertility.

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**Aims:** - Infertility is characterized by the inability to achieve pregnancy after 12 months of unprotected sexual intercourse. One in every six couples experiences infertility, with contributing risk factors encompassing hormonal, immunological, and psychological elements. In normal spermatogenesis, TP53 a known tumor suppressor gene located on 17p13, plays an important role in controlling the number of germ cells by apoptosis of defective germ cells. As the activity of TP53 is very crucial for DNA repair, cell division and the apoptotic pathway, any single nucleotide substitution in the gene can cause gain-of-function or loss-of-function changes. One such change is the TP53 codon 72 polymorphism (rs1042522); the wild type CCC in codon 72 codes for proline, and the mutant CGC codes for arginine. It has been reported that women carrying the P/P genotype of p.R72P polymorphism were at an increased risk of recurrent pregnancy loss in Austrian and Chinese populations and with implantation failure in the Caucasian population. On the contrary in Brazilian population, the R/R genotype of p.R72P polymorphism was associated with recurrent pregnancy loss but in Brazilian males, the TP53p.R72P polymorphism was not significantly associated with idiopathic infertility. Thus few studies have reported the association of p.R72P polymorphism with infertility but the results have been inconclusive. The Arg/Arg genotype and Arg allele of TP53codon 72 have been reported to be associated with increased risk for azoospermia in Indian population.

The aim of present case-control study was to identify the association, if any, of TP53 p.R72P polymorphism with infertility in couples facing repeated IVF failure.

**Methods:** - In this study 50 infertile patients facing repeated IVF failure and 50 age-matched healthy controls from Jalandhar region of Punjab, India were screened. The mean age of patients was  $34.52 \pm 5.91$  and of controls was  $36.94 \pm 6.36$  years. Genomic DNA was isolated from blood samples using standard phenol chloroform method. Quality and quantity of DNA samples was checked on 1% ethidium bromide

stained agarose gel. The samples were screened for TP53 p.R72P polymorphism by the PCR-RFLP technique using BstUI enzyme.

**Results:** - The frequency of PP, PR and RR genotype was 26% vs 36%, 54% vs 36% and 20% vs 28% in patients and controls respectively. PR frequency was higher in cases but difference was statistically non significant. No association of TP53 p.R72P polymorphism was found with infertility (OR = 1.01, CI = 0.34-2.98, p = 0.98).

**Conclusions:** - The present study concluded that TP53 p.R72P polymorphism was not associated with infertility in infertile patients from Punjab, India.

**Keywords:** - The frequency of PP, PR and RR genotype was 26% vs 36%, 54% vs 36% and 20% vs 28% in patients and controls respectively. PR frequency was higher in cases but difference was statistically non significant. No association of TP53 p.R72P polymorphism was found with infertility (OR = 1.01, CI = 0.34-2.98, p = 0.98).