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Abstract Topic: - Molecular and cytogenetic diagnostics

Abstract Title: - Investigation of VEGFR1-710 C/T promotor Polymorphism in Male Diabetic Retinopathy Patients.

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Aims: - Diabetes is a chronic lifelong condition defined by excessive circulating glucose in blood. It is caused by the body's inability to create or use insulin to its full capacity. Diabetes is becoming more common and has become one of the most significant problems to modern health care. Diabetic Retinopathy (DR) is classified clinically into two types: nonproliferative DR (NPDR) and proliferative DR (PDR). The two most common causes of vision impairment and blindness in DR patients are diabetic macular edema (DME) and retinal neovascularisation. Retinopathy, neuropathy, nephropathy, peripheral artery disease, coronary artery disease, cerebrovascular disease, diabetic cardiomyopathy and diabetic foot are the complications of Diabetes. Angiogenesis is the process through which new vascular networks emerge from established vessels. Angiogenesis is regulated by cellular signaling mediated by vascular endothelial growth factor (VEGF) and its receptors. VEGF triggers it's signalling via VEGFR1, VEGFR2 and VEGF3 receptors. VEGFR1 has a high affinity for VEGF and acts as an endogenous inhibitor of angiogenesis by sequestering VEGF .VEGFR1 has been demonstrated to be a positive modulator of pathological angiogenesis in experimental models of certain primary malignancies and wet age-related macular degeneration. Several single nucleotide polymorphisms (SNPs) have been reported in different region of VEGFR1. The VEGFR1-710C/T promoter polymorphism has been studied in different diseases, like breast cancer, bronchopulmonary dysplasia and till date there is no study on diabetic retinopathy. The aim of this case control study was to investigate the association of VEGFR1 -710C/T promoter polymorphism in male diabetic retinopathy patients from Punjab, North-West India.

Methods: - The genomic DNA was extracted from blood samples of 52 male diabetic retinopathy patients and 52 age matched healthy unrelated male controls using standard phenol chloroform method. Quality and quantity of DNA samples were checked on 1% ethidium bromide stained agarose gel. The DNA samples were screened for VEGFR1-710C/T promoter polymorphism using PCR-RFLP method. The region of VEGFR1 containing -710C/T polymorphism was amplified using specific primers. PCR products were digested with NspI restriction enzyme. Genotyping was done on the basis of fragments obtained after digestion.

Results: - The mean age of patients and controls were 58.46±8.92 and 58.88±11.12 years respectively. The frequency of CC genotype was 96.15% vs 94.23% and CT genotype was 3.85% vs 5.77% in patients

and controls respectively. The frequency of C allele was 98.07% vs 97.11% and T allele was 1.92% vs 2.88% in patients and controls respectively. None of the patients and controls had TT genotype.

Conclusions: - There was no association of VEGFR1-710C/T polymorphism with diabetic retinopathy risk in the studied patients .

Keywords: - The mean age of patients and controls were 58.46±8.92 and 58.88±11.12 years respectively. The frequency of CC genotype was 96.15% vs 94.23% and CT genotype was 3.85% vs 5.77% in patients and controls respectively. The frequency of C allele was 98.07% vs 97.11% and T allele was 1.92% vs 2.88% in patients and controls respectively. None of the patients and controls had TT genotype.