ISSN 0972-3757
International Journal of
HUMAN GENETICS

PRINT: ISSN 0972-3757 ONLINE: 2456-6360
Int J Hum Genet, 11(2): 111-115 (2011)
DOI: 10.31901/24566330.2011/11.02.07

5HTT Promoter Polymorphism in Idiopathic Pulmonary Arterial Hypertension

V. Shivani[1], K.Sujana[1], B. K. S. Sastry[2] and Pratibha Nallari[1]

1. Department of Genetics, OsmaniaUniversity, Hyderabad 500 007, Andhra Pradesh, India
2. Department of Cardiology, CARE Hospital, The Institute of Medical Sciences, Nampally, Hyderabad 500 001, Andhra Pradesh, India

KEYWORDS IPAH. Serotonin. 5HTT Promoter-Polymorphism. Frequency. Mean Pulmonary Artery Systolic Pressure. Hyperplasia

ABSTRACT A 44bp insertion/deletion in promoter of 5HTT (5Hydroxy Tryptamine Transporter) exists as a polymorphism in general population resulting in 3 genotypes LL (528 base pair), LS and SS (484 base pair) .The transcriptional efficiency of the L allele is 2-3 fold higher than the S allele. The L-allelic variant of the 5-HTT gene promoter is associated with 5-HTT overexpression. This higher expression of the transporter leads to higher uptake of serotonin, resulting in activation of mitogenic pathways thereby inducing smooth muscle hyperplasia. The study focuses on the possible association of 5HTT insertion/deletion promoter polymorphism with Idiopathic Pulmonary Arterial Hypertension (IPAH) patients and distribution of the polymorphism among the control individuals from Indian population. In the present study, 65 IPAH cases and 100 controls were considered for comparative analysis. DNA samples from controls and patients were amplified using Polymerase Chain reaction and the products were genotyped on 2% agarose gel stained with ethidium bromide. Frequency of L allele was found to be much higher in IPAH (0.538) as compared to controls (0.335). The LL and LS genotypes were found to be at an higher risk for IPAH as compared to SS genotype (OR - 3.117, CI -1.293 ,7.579 and OR-5.250 , CI- 2.186 - 12.776). The mPASP (mean pulmonary artery systolic pressure) was also significantly higher among the LL and LS genotypes when compared to the SS genotype in IPAH patients.The L allele could be a possible risk factor for IPAH and play a significant role in disease progression