

Association between Catechol-O-Methyltransferase Gene Variant and Bipolar Disorder

N.H.V. Kutumbarao, Arundhuti Das and V. Lakshmi

Department of Human Genetics, Andhra University, Visakhapatnam, Andhra Pradesh, India

KEYWORDS Bipolar Disorder. Catechol-O-Methyltransferase (COMT) Gene Variants. Disease Association

ABSTRACT Emotional dysregulation is a core characteristic of many psychiatric diseases. Bipolar disorder is a brain disorder that causes unusual shifts in mood, energy level, activity level and ability to carry day to day work. Catechol-O-methyltransferase (COMT) is involved in the metabolism of dopamine and epinephrine. To a large extent it is responsible in maintaining human cognitive functioning. Polymorphisms in the COMT gene lead to various dysregulations. For the study we have considered single nucleotide polymorphism (SNP) rs 4680 (val158met), a G to A transition mutation due to which valine is substituted by methionine at codon 158, which reduces the enzyme activity by four folds. The aim of the current study is to check whether the low activity allele has any association with bipolar disorder. The study included 50 unrelated patients, diagnosed suffering from bipolar disorder. Age and sex matched control samples were taken. Blood samples were collected from all the individuals after taking informed consent. SNP was genotyped with a PCR-based restriction fragment length polymorphism analysis using the restriction enzyme NlaIII. Amongst the 50 patients, 29 were homozygous for G allele, 18 were heterozygous and 3 were homozygous for A allele. In the control population only 6 individuals were found to be heterozygous for the variation. The met allele frequency is 0.24 in patients and 0.06 in control group. Chi square value is 12.08 (df=2) and p value is 0.002. Significant association was observed between the catechol-O-methyltransferase val158met polymorphism and bipolar affective disorder.