ISSN 0972-3757
INTERNATIONAL JOURNALOF HUMAN GENETICS

PRINT: ISSN 0972-3757 ONLINE: 2456-6360
Int J Hum Genet, 5(3): 183-186 (2005)
DOI: 10.31901/24566330.2005/05.03.04

PCR-Based Detection of Parental Origin of Extra Chromosome 21 in Down Syndrome

S.K. Dey and Sujoy Ghosh

Molecular Cytogenetics Laboratory, Department of Zoology, Presidency College, 86/1 College Street, Kolkata 700 073, West Bengal, India

KEYWORDS Down syndrome; trisomy 21; parental origin; PCR; microsatellite markers

ABSTRACT Down syndrome (DS) is one of the most frequent chromosomal abnormalities in humans and is associated with mental retardation .It is usually caused by the presence of an extra chromosome 21.The parental origin of the extra chromosome has been studied in twenty families each with a clinically suspected trisomy 21 proband.The trisomy 21 was confirmed by both chromosomal and molecular analyses.Molecular analysis was carried out by PCR based method, using polymorphic microsatellite markers D21S11 and D21S2055 situated on the long arm of the chromosome 21 at 21q21 and 21q22 respectively. The amplified products were subjected to polyacrylamide gel electrophoresis and alleles were scored by staining with ethidium bromide. Trisomy 21 Down syndrome was detected by the presence of three distinct alleles and transmission of alleles from parents to the offspring was determined in all but six families .Parental origin was found to be maternal in nine families and paternal in five families .The mean maternal and paternal age of our subjects were 29±6.9 and 35±6.9 years respectively. Our results further demonstrate the usefulness of highly polymorphic microsatellite markers for the accurate determination of parental origin of the extra chromosome 21 in Down syndrome and also emphasize the fact that the trisomy 21 was due to meiotic errors in maternal chromosomes.