ISSN 0972-3757
International Journal of
HUMAN GENETICS

PRINT: ISSN 0972-3757 ONLINE: 2456-6360
Int J Hum Genet, 11(3): 177-182 (2011)
DOI: 10.31901/24566330.2011/11.03.05

Application of Fluorescence *in situ* Hybridization (FISH) Technique to Discern Complete/Partial Monosomy 21

M. Jeevan Kumar[1], R. Ashok Kumar[2], T. Jayasudha[1] and Kalpana Gowrishankar[1*]

1 Department of Medical Genetics, Kanchi Kamakoti CHILDS Trust Hospital, Chennai, Tamil Nadu, India
2 School of Bio Sciences and Technology, VIT University, Vellore, Tamil Nadu, India

KEYWORDS Partial Monosomy. GTG. Interphase. Metaphase. FISH

ABSTRACT The present study pertains to a 7 month old female infant who showed dysmorphic features and developmental delay. Conventional cytogenetic analysis by GTG banding technique was carried out and this revealed monosomy 21. Interphase FISH and metaphase FISH were employed for better delineation of the observed results. FISH analysis confirmed unbalanced X;21 translocation. This study concludes that FISH technique along with conventional cytogenetic analysis serves for better understanding and interpretation of complete/ partial autosomal monosomies.