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L1(CAM) Mutation in Males with Mental Retardation

M. Swarna, M. Sujatha, P. Usha Rani and P.P. Reddy

*Institute of Genetics and Hospital for Genetic Diseases, Begumpet,
Hyderabad 500 016, Andhra Pradesh, India*

KEY WORDS Mental Retardation; L1(CAM) mutation; CRASH.

ABSTRACT L1 CAM (L1 cell adhesion molecule) plays a key role in the development of nervous system. Recent studies have shown the evidence for the involvement of L1 (CAM) mutations in X-linked mental retardation syndromes. No such studies were undertaken from India and hence, an attempt was made to detect L1 (CAM) mutations in mental retardation. Eight patients from different families were selected to know the molecular basis of mental retardation in these patients. Genomic DNA from all the patients and controls was analyzed by polymerase chain reaction. PCR analysis was carried out using specific primers to screen for L1 (CAM) mutations. Among the eight patients, in two patients mutations in exon-4 and intron-4 junction were detected.

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