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Mutation characterization and heterodimer analysis of patients with leukocyte adhesion deficiency: Including one novel mutation

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Background and Aim: Leukocyte adhesion deficiency type 1 (LAD-I) is a rare, autosomal recessive disorder of neutrophil migration, characterized by severe, recurrent bacterial infections, inadequate pus formation and impaired wound healing. The *ITGB2* gene encodes the β2 integrin subunit (CD18) of the leukocyte adhesion cell molecules and mutations in this gene cause LAD-I. The aim of the current study was to investigate the mutations in patients diagnosed with LAD-I and functional studies of the impact of two previously reported and a novel mutation on the expression of the CD18/CD11a heterodimer.

Materials and Methods: Blood samples were taken from three patients who had signed the consent form. Genomic DNA was extracted and ITGB2 exons and flanking intronic regions were amplified by polymerase chain reaction. Mutation screening was performed after Sanger sequencing of PCR products. For functional studies, COS-7 cells were co-transfected with an expression vector containing cDNA encoding mutant CD18 proteins and normal CD11a. Flow cytometry analysis of CD18/CD11a expression was assessed by the dimer-specific IB4 monoclonal antibody.

Results: Two previously reported mutations and one novel mutation,p. Cys562Tyr were found. All mutations reduced CD18/CD11 heterodimer expression.

Conclusion: Our strategy recognized the p.Cys562Tyr mutation as a pathogenic alteration that does not support CD18 heterodimer formation. Therefore, it can be put into a panel of the carrier and prenatal diagnosis programs.

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