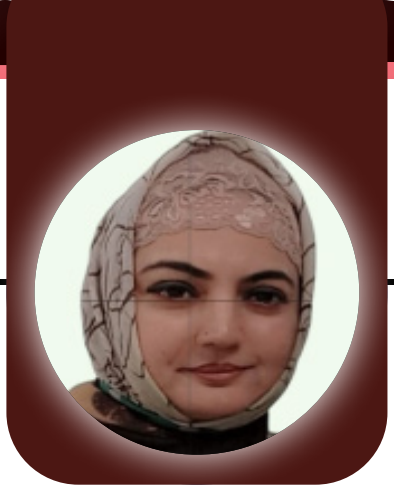

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Rare and heterogeneous manifestations of leucocyte adhesion deficiency type 1: report of two cases with diagnostic dilemmas and novel ITGB2 mutation

Abstract:

Primary immunodeficiency disorders (PID) are rare disorders with heterogeneous manifestations, overlapping with other diseases such as autoimmunity, malignancy, and infections. This makes the diagnosis very challenging and delays management. Leucocyte adhesion defects (LAD) are a group of PIDs in which patients lack adhesion molecules on leukocytes needed for their emigration through blood vessels to the site of infection. Patients with LAD can present with diverse clinical features including severe and life-threatening infections, early in life, and the absence of pus formation around infection or inflammation. There is often delayed umbilical cord separation, omphalitis, late wound healing, and a high white blood cell count. If not recognized and managed early, can lead to life-threatening complications and death.

Case Presentation: LAD 1 is characterized by homozygous pathogenic variants in the integrin subunit beta 2 (ITGB2) gene. We report two cases of LAD1 with unusual presentations (post-circumcision excessive bleeding and chronic inflammation of the right eye) which were confirmed by flow cytometric analysis and genetic testing. We found two disease-causing ITGB2 pathogenic variants in both cases.

Conclusions: These cases highlight the importance of a multidisciplinary approach to recognizing clues in patients with uncommon manifestations of a rare disease. This approach initiates a proper diagnostic workup of primary immunodeficiency disorder leading to a better understanding of the disease, and appropriate patient counseling, and helps clinicians to be better equipped to deal with complications.

Biography

Sabahat Sarfaraz is an Assistant Professor of Pathology at Dow International Medical College, and Dow University of Health Sciences. She has a keen interest in allergy diagnosis and testing. Besides she runs a transplant immunology lab at DUHS.