A Case Report: Jacobsen Syndrome Complicated by Paris-Trousseau Syndrome and Shone's Complex

Abstract

A preterm infant presenting with a congenital cardiac malformation and thrombocytopenia was found to have a karyotype showing a terminal deletion of the long arm of chromosome 11 of the segment 11q24.1-11qter consistent with Jacobsen syndrome. The infant was later diagnosed with Paris-Trousseau syndrome, commonly associated with Jacobsen syndrome. Because children with cardiac malformations often require high-risk surgical procedures in the early neonatal period, those with platelet dysfunction require prompt identification at birth.

Malia LA, Wolkoff LI, Mnayer L, Tucker JW, Parikh NS. A Case Report: Jacobsen Syndrome Complicated by Paris-Trousseau Syndrome and Shone's Complex. J Pediatr Hematol Oncol. 2015 Oct;37(7):e429-32. doi: 10.1097/MPH.000000000000372. PMID: 26056793.