

Giant platelets in a case of deletion 11q24-qter confirmed by fluorescence in situ hybridization

Abstract

Here we report the association of giant platelets and an increase in platelet volume in a 19-month-old black female with de novo del 11q24-qter. The deletion, which was visible on karyotype, was further confirmed and more precisely localized by fluorescence in situ hybridization studies (FISH) that showed the deletion to lie distal to the MLL gene region (11q23). Clinically, the case presented less severe symptoms than Jacobsen syndrome-the well known partial deletion of the distal end of chromosome 11. Platelet glycoproteins CD 41, CD 42a, C 42b, CD 61, and PAC-1 were also assayed and found to be normally expressed. To our knowledge, giant platelets are described for the first time in the relevant deleted region.

Laley A, Delneste D, Pradier O, Hans C, Darboux R, Ogur G. Giant platelets in a case of deletion 11q24-qter confirmed by fluorescence in situ hybridization. *Am J Med Genet.* 2002 Jun 15;110(2):170-5. doi: 10.1002/ajmg.10418. PMID: 12116256.