Primary immunodeficiency in combination with transverse upper limb defect and anal atresia in a 34-year-old patient with Jacobsen syndrome

Abstract

We describe a 34-year-old male patient with Jacobsen syndrome associated with a broad spectrum of anomalies and an increased susceptibility to infections. Features commonly seen in Jacobsen syndrome were short stature, mental retardation, congenital heart disease, cryptorchidism, strabismus, distal hypospadia glandis, and mild thrombocytopenia. Chromosome analysis disclosed a mosaic 46,XY,del(11)(q24.1)/46,XY karyotype with a very low percentage of normal cells. In addition, transverse upper limb defect, imperforate anus, and hearing impairment were noted. Cellular anomalies include functional impairment and deficiency of T-helper cells, and a low serum immunoglobulin M (IgM)-level. The presence of a transverse limb defect and primary immunodeficiency has not been reported previously in Jacobsen syndrome.

von Bubnoff D, Kreiss-Nachtsheim M, Novak N, Engels E, Engels H, Behrend C, Propping P, de la Salle H, Bieber T. Primary immunodeficiency in combination with transverse upper limb defect and anal atresia in a 34-year-old patient with Jacobsen syndrome. Am J Med Genet A. 2004 Apr 30;126A(3):293-8. doi: 10.1002/ajmg.a.20592. PMID: 15054845.