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Hypoimmunoglobulinemia and Protein C Deficiency in a Girl with Jacobsen Syndrome: A Case Report

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Abstract

Jacobsen syndrome is a rare contiguous gene syndrome caused by partial deletion of the long arm of chromosome 11. The typical clinical manifestations include physical growth retardation, mental retardation, facial dysmorphisms, congenital heart disease, thrombocytopenia, or pancytopenia.

A Thai-Australian girl was born with multiple abnormalities. Typical features and her karyotype, 46, XX, del(11) (q23-qter), confirmed Jacobson syndrome. She had many uncommon findings including upslanting palpebral fissures, tortuousity of retinal vessels and hypogammaglobulinemia. In addition, this case also presented with protein C deficiency, which has not been reported previously in Jacobsen syndrome. The patient was treated with phototherapy, intravenous antibiotic injection, and platelet transfusion in neonatal period. Cranioplasty was performed for prevention of the increased intracranial pressure at three months of age. Surgical correction for strabismus was in the treatment plan.

Keywords: Jacobsen syndrome, Deletion 11q, 11q deletion syndrome, Hypoimmunoglobulinemia, Immunodeficiency, Immune deficiency

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