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Digeorge Syndrome and Immune Thrombocytopenia Purpura

Amna Mohsin[1*] and Mahasin Shaheen[2]

*[*1]Lister hospital East and North Hertfordshire NHS trust, UK*

[2]Al Qassimi Hospital, Al Khezamia, Sharjah, UAE.

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ABSTRACT

Digeorge Syndrome (22q11.2 deletion syndrome) comprises of various congenital anomalies such as cardiac defects, specific facial appearances, defective T cell production, hypocalcemia etc. This syndrome is also associated with various autoimmune diseases. Here we present a case report which shows association of Immune thrombocytopenia purpura with Digeorge Syndrome. A 15 years old boy born with congenital cardiac defects, diagnosed as 22q11.2 deletion syndrome, underwent corrective cardiac surgeries after birth. He developed thrombocytopenia at this age, requiring admission and platelet transfusions. His further work up confirmed Immune thrombocyte penicpurpura and its presentation being related to the genetic disorder itself.

Keywords: Cleft palate, Congenital cardiac anomalies, 22q11.2 deletion, Digeorge syndrome, Immune thrombocytopenic purpura, Purpuric rash

Corresponding author: Amna Mohsin, Lister hospital East and North Hertfordshire NHS trust, UK, E-mail: am_rawalian@hotmail.com

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