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DiGeorge syndrome presenting with seizures: A case report

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DiGeorge syndrome was described for the first time in 1968 as a defect affecting structures derived from the third and fourth embryonic pharyngeal arches along with absent parathyroid glands. According to the low incidence of this disease as well as a wide spectrum of symptoms, it is essential to report cases with less prevalent features. In this case report, a child has been introduced with a diagnosis of DiGeorge syndrome presenting with seizures.

The patient was a 27-day-old baby girl due to seizures admitted to hospital Imam Reza (AS), Mashhad, Iran. Hypocalcemia was observed in early clinical trials requested. The patient underwent echocardiography according to holosystolic murmur grade 3/6 auscultation, which showed a patent ductus arteriosus (PDA), tetralogy of Fallot (TOF), ventricular septal defect (VSD), atrial septal defect (ASD), and pulmonary atresia (PA). No thymus was found on chest x-ray, and evidence of previous conflicts was observed in the heart. Finally, Fluorescent in situ hybridization (FISH) was performed to check out Tuple gene deletion on chromosome 22q11.2, and the diagnosis was confirmed for Disgorge Syndrome.

Although the incidences of neurological symptoms associated with hypocalcemia suggest a wide range of diseases as a differential diagnosis, pediatrics should consider the heart disorders for DiGeorge syndrome through clinical examinations and imaging, if necessary.

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