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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. Hypo-calcemia was observed in early clinical trials requested. The patient underwent echocardiography according to holo-systolic murmur grade 3/6 auscultation, which showed a patent ductus arteriosus, tetralogy of fallot, ventricular septal defect, atrial septal defect and pulmonary atresia. No thymus was found on chest x-ray and evidence of previous conflicts was observed in the heart. Finally, fluorescent *in situ* hybridization 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 hypo-calcemia 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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