



## Cardiac Abnormalities Related to 10q26 Chromosomal Deletion in Neonate

Ehsan Aghaei Moghadam<sup>1</sup>, Maryam Nikoufar<sup>2,\*</sup>, Azin Ghamari<sup>3</sup> and Mohammad Taghi Majnoun<sup>2</sup>

<sup>1</sup>Assistant Professor of Pediatric Cardiology, Tehran University of Medical Sciences, Thran, IR Iran

<sup>2</sup>Pediatric Cardiology Fellow, Tehran University of Medical Sciences, Thran, IR Iran

<sup>3</sup>Medical Student, Tehran University of Medical Sciences, Thran, IR Iran

\*Corresponding author: Maryam Nikoufar, Pediatric Cardiology Fellow, Tehran University of Medical Sciences, Thran, IR Iran, E-mail: mary\_nikoofar@yahoo.com

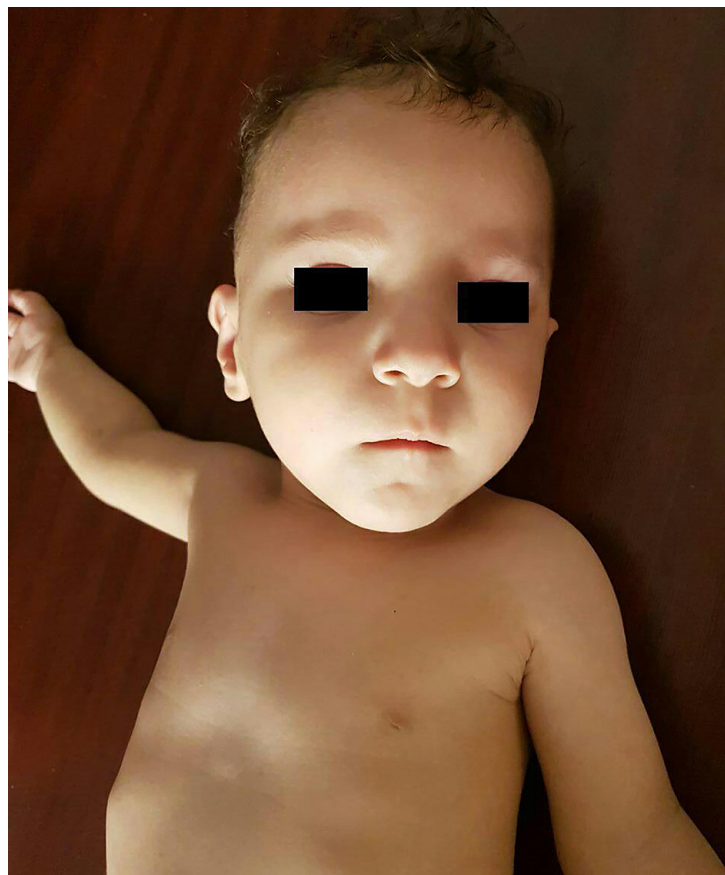
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Chromosomal microdeletions are usually accompanied with medical and developmental concerns. Although same microdeletions obviously are not symptomatic during childhood, some other deletions are related to clinical manifestations in neonates such as growth and neurodevelopmental delay, feeding difficulties, hypotonia, and motor developmental defects (1). Cardiovascular anomalies have been revealed to affect up to half of all neonates born with a 10q26 microdeletion (2). The described case was a female neonate (11 month, 15 days) with the birth weight of 2400 g that was born through cesarean section from a 25-years old healthy mother (G3P2Ab1L1). The patient was the second child of the family with no abnormal condition in first child. She was evaluated from birth because of appearing cardiac murmurs that was assessed by echocardiography leading the diagnosis of Ventricular septal defect plus pulmonary stenosis. At the age of 6 months, the patient was revisited due to pulmonary infection and admitted to neonatal intensive care unit but without requiring mechanical ventilation. Concurrently, the patient was worked-up because of septic arthritis of the hip joint simultaneously with neurodevelopmental delay. In this regard, according to the chromosomal study, deletion in the location of 10q26 was detected. Liver function test and other metabolic work-ups were all normal. Although patient could sit with help in the month of 9, she was unable to stand without helping. In physical examination, she seemed to be hypotonic and flaccid with the weight of 6500 g, and head circumference of 39.5 cm. In echocardiography assessment, the following findings were evident: perimembranous VSD size of 4.5 mm, a left to right shunt, severe supravalvular and valvular pulmonary stenosis, with a gradient of 75 mmHg and confluent good size of pulmonary artery branches. A bicuspid pulmonary valve with the size of 9.2 mm was also detected. The diameters of the left and right pulmonary arteries were also estimated to be 8mm and 7 mm respectively. Because of the pointed

evidences, the patient underwent open heart repairing surgery with the aim of total VSD closure, PDA ligation, and pulmonary valvulotomy with supra-annular patch. After surgical correction and recovery, the patient was discharge with a clinical good condition with recommending neurological follow-up for neurodevelopmental delay. 10q26 microdeletion syndrome was initially described by Lewandowski et al. (3). It has been recently showed that the specific change in chromosome 10 related to heart defects included a de novo approximately 12.5 Mb terminal deletion 10q26.12 -> qter (4). The most frequent conditions observed in the affected children including persistence of prenatal cardiac structures such as PDA followed by VSD and ASD that commonly no needing surgical intervention and thus can be self-limited (5). It can be concluded that in neonates with multiple cardiac and non-cardiac defects, chromosomal assessment by specific tests such as FISH test may result in discovering some deletions or structural polymorphisms led to accurately diagnosis of the source of syndrome and thus to better management of patient.

### References

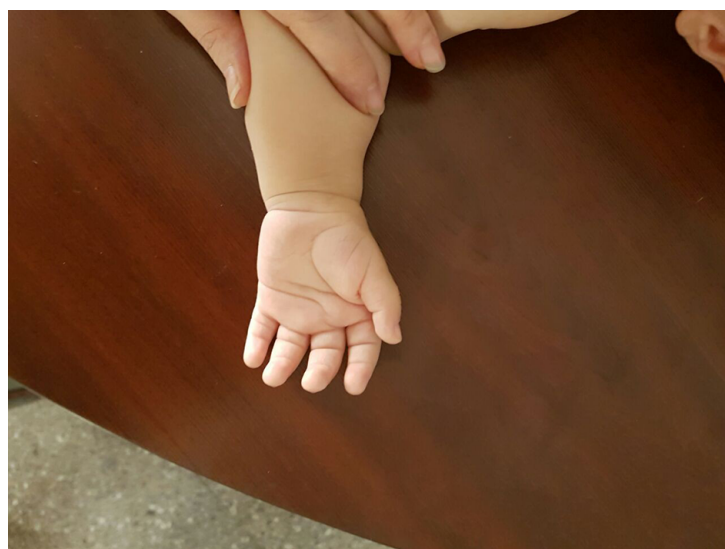
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**Figure 1.** A general picture of the neonate suffered from heart defects related to 10q26 chromosomal deletion

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**Figure 2.** A picture of the hand in the neonate suffered from heart defects related to 10q26 chromosomal deletion

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