Case Review

Congenital Scoliosis and Tetralogy of Fallot With Neuro-development Delay: A Case Study and Literature Review

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Citation Ghandi Y, Karimi S, Karimi R. Congenital Scoliosis and Tetralogy of Fallot With Neurodevelopment Delay: A Case Study and Literature Review. Journal of Pediatrics Review. 2022; 10(3):247-252. http://dx.doi.org/10.32598/jpr.10.3.913.2

http://dx.doi.org/10.32598/jpr.10.3.913.2

Article info:
Received: 14 Feb 2021
First Revision: 25 Apr 2022
Accepted: 09 May 2022
Published: 01 Jul 2022

Keywords:
Congenital heart disease, DiGeorge syndrome, Scoliosis, Case report, Child

ABSTRACT

As the most common congenital heart malformation, tetralogy of Fallot (TOF) produces cyanosis. Patients with TOF suffer from a higher frequency of major noncardiac congenital disorders. Its association with congenital scoliosis influences vital and functional overcomes, restricting physical activity and lowering life expectancy. An 8-month-old female child was reported with admitted cough, fever, and ruled-out pneumonia. The child was diagnosed with heart disease at 2-month-old when cyanosis was apparent. After being admitted to a hospital, history and physical examination showed mild neurodevelopmental delays, such as an inability in rolling and crawling. Her chest x-rays revealed congenital spine abnormalities thoracic vertebral at T3-T8 levels and bilateral segmented-bar sacral vertebrae. Given that patients with TOF routinely undergo chest radiographs, physicians examining TOF patients' chest radiographs should be aware of the potential for congenital scoliosis to provide early diagnosis and referral for orthopedic evaluation and treatment.

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1. Introduction

Congenital heart disease (CHD) has a relationship with scoliosis and affects 1% of live births. This aspect induces a 10-fold enhancement in scoliosis prevalence in children with CHD in age-matched populations (1).

In addition, spine congenital deformities identified at the birth time are vertebral, a byproduct anomalous in the embryo. Minor malformations of bones occur in more than 12% of the general population and are usually not apparent. They are often identified on routine lumbar spine or chest films (2). However, congenital spinal malformations result in progressive deformities of the spine, occurring with a reported frequency of 0.5/1000 births (3).

Cases with tetralogy of Fallot (TOF) have more incidence of major noncardiac defects (i.e., scoliosis congenital with neurological abnormalities) (4). Congenital scoliosis (CS) includes congenital vertebral anomalies caused by segmentation, defects of forming, combined or contributory mechanisms, such as congenital vertebral anomalies, thoracogenic causes, and the presence of syndromes (3).

Mutations of vertebral body precursors lead to CS, which can be manifested as a formation and segmentation failure, or a combination of them. Although no specific cause of CS is known, environmental factors, genetics, vitamin deficiency, chemicals, and drugs are related to this condition (5).

Chest radiographs during infancy are repeatedly used to evaluate patients with TOF. Because of a significant correlation between CS and TOF, these cases should be screened for CS (3). In the present study, we have reported a case with CS and TOF with neurodevelopment delay.

2. Case Report

We present an 8-month-old female child, diagnosed with congenital cyanogen heart malformation (TOF), admitted because of cough and fever and ruled-out pneumonia. The child was born out of a non-consanguineous marriage. The child was detected to have heart disease on day 1 of life as the cyanosis was apparent. The cyanosis aggravated every time during cry and exertion. There was no history of spells or squatting in early childhood.

The baby was delivered vaginally at 37-week gestation with appropriated Apgar score and birth weight of 3,150 g. The mother denied consuming alcohol, smoking, or experiencing any period of illness during gestation. Clinical examination revealed cyanosis (81%) and a 3/6 systolic ejection murmur.

The chest radiographs showed a boot-shaped heart because of right ventricular hypertrophy and clear lungs. The spine X-rays indicated thoracolumbar scoliosis with a right convex thoracic curve. Also, the chest x-ray revealed butterfly vertebrae at Th3 and Th8, as well as bilateral segmented-bar sacral vertebrae. The "S" thoracolumbar scoliosis with convex thoracic right curve peaked at T5 (Figure 1).

Echocardiography showed situs solitus, a large subaortic ventricular septal defect with overriding of aorta < 50%, severe valvular pulmonic stenosis, and sub-valvular infundibular dynamic obstruction. The gradients across the pulmonic valve were 80 mmHg. The pulmonic valve was doming and the patient was categorized as syndromic TOF.

The abdominal ultrasound was performed and the liver, kidney, and spleen were normal. In the spinal examination, mild scoliosis without kyphosis was found. Upper and lower limp, reflex, and neuromotor examinations were normal, and the patient had a mild neurodevelopmental delay, while early development is marked by enormous gains in gross motor abilities.

The O2 systemic saturation determined by pulse oximeter was SpO2=76%. While inspecting the spine, the deviation of the axis of the thoracolumbar spine was noticed in the frontal plane, with a right convex thoracic curve; this is not corrected by lateral bending, and the right side gibbous costal deformity pronounced by movements of flexion of the spine, symmetry of the shoulders, and pelvic symmetry with apparent inequality of limb without limb neurological disorders. On laboratory findings, blood tests and liver function tests were within normal limits.

The electrocardiography showed sinus rhythm, heart rate frequency of 120/min, QRS axis deviated to the right, right ventricular hypertrophy, narrowed QRS complex, and symmetrical, sharp negative T-waves in V1-V3. The 22q11.2 deletion was done and the patient was diagnosed with a mutation.

The patient consulted with orthopedic and neurologic and neurosurgery specialists. The spinal com-

puterized tomography (CT) and magnetic resonance imaging (MRI) was not done. As there was no neurologically significant finding, brace devices were proposed to start walking.

3. Discussion and Literature Review

One of the most common deformities of the spine is scoliosis with more than 10 degrees of the lateral curve. The most common scoliosis type is idiopathic scoliosis (IS) which comprises approximately 80% of patients with scoliosis. The other 20% are cases of CS; scoliosis related to syndromes and neuromuscular scoliosis. The cause of IS is unknown and it is diagnosed by excluding the causes of other types of scoliosis (6, 7). The CS is characterized by the failure of vertebral formation or segmentation during embryonic development. The CS is frequently associated with other spinal cord abnormalities, genitourinary system, bowel, auditory, and heart (7, 8). The relationship between CHD and scoliosis has been recognized for many years.

CHD in CS cases have a 30% prevalence (6). The prevalence of CHD has increased based on several reports, including atrial septal defect, mitral valve prolapse, ventricular septal defect, mitral valve dysplasia, and tricuspid regurgitation in IS cases (9).

The prevalence of CS is 0.5 to 1 in 1000 live births. Previous studies have demonstrated that the prevalence of CS and IS has increased in CHD, including TOF (7, 9). However, studies correlating specific CHD, such as TOF and CS are scarce.

The particular importance of rapid spine growth during the first 5 years and decreased trunk and stature height associated with CS increases the risk of thoracic insufficiency syndrome. Patients with early progressive deformities, such as a unilateral bar with a contralateral hemivertebrae may benefit the most from an early diagnosis of CS. New treatment options, such as growth rods, and vertical expandable prosthetic titanium rib (VEPTR) devices, for children from the age of 1 year make early detection of CS even more imperative (4, 10).

Previous studies have demonstrated that CS is associated with spine and other organ abnormalities. Spine abnormalities as congenital deformities of the spine range from 15% to 38% (11, 12). Some other studies have demonstrated the incidence of such abnormalities was related to spinal deformity type and the hemivertebrae level (13). Basu et al. mentioned the prevalence of 26% of CHD in patients with congenital spinal deformity (14). Bollini et al. reported CHD as 8% of 75 cases with hemivertebrae. No studies have documented the anomalies in patients with CS in Chinese cases (12).

Shen et al. observed that 43% of 226 Chinese surgical patients with CS had intraspinal abnormalities and diastematomyelia as the most common anomalies. The incidence of an intraspinal anomaly in patients with failures of segmentation and mixed defects was significantly higher compared to patients with failures of formation. Abnormal findings on physical examination results were not significantly associated with intraspinal abnormality (11).

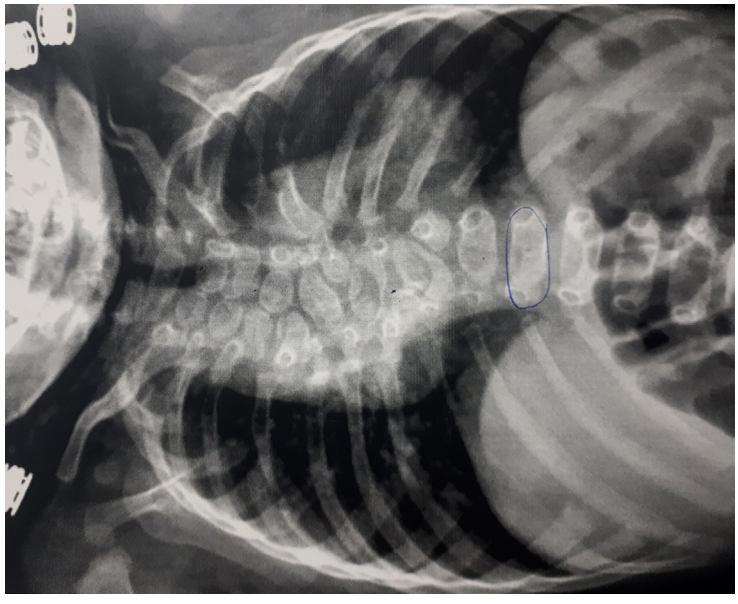

Figure 1. Radiograph showing congenital multiple malformed vertebral at thoracic level Th3 to Th8

Saifi et al. reviewed the medical records and chest radiographs of 562 patients who underwent corrective surgery for TOF. Of the 364 patients examined in the study, 12 patients (3.3%), including 6 girls and 6 boys, had a positive diagnosis of CS confirmed by the radiographic examinations (5). Jelle et al. confirmed that clinicians should be aware that scoliosis is highly prevalent (48%-49%) in association with 22q11.2DS, irrespective of other clinical features. Furthermore, 22q11.2DS may provide insights into the causes of adolescent IS. They showed that the presence of scoliosis within 22q11.2DS is not associated with the presence of CHD. In addition, they demonstrated that the majority of the scoliotic curve patterns in patients with 22q11.2DS is a typical, idiopathic-like, curve patterns (15). The 22q11.2 deletion syndrome is the most common microdeletion syndrome in humans. It is characterized by wide phenotypic variability, including CHD, immunodeficiency, and scoliosis (16, 17).

Dirkjan et al. showed that adult patients with genetically confirmed 22q11.2 DS had a higher risk of all-cause and cardiovascular death, independent of age, sex, and CHD. No differences were seen in risk for cardiac complications, including pulmonary valve replacement ventricular arrhythmias (VA), pacemaker implantation (PM), Implantable cardioverter-defibrillator (ICD). Genetic analysis is indicated in all patients, as 22q11.2DS is a risk factor for important outcomes (18).

Jelle et al. observed that scoliosis prevalence in CHD cases (without 22q11.2 deletion) approximates the general population. However, in the CHD population with a 22q11.2 deletion, the prevalence of scoliosis approximates the population with 22q11.2DS. The pediatric surgical methods and CHD severity were weaker irrespective of 22q11.2 deletion. The results support the importance of genetic diagnosis of 22q11.2DS to developing scoliosis risk in CHD cases. The 22q11.2 deletion may represent a common etiopathogenetic pathway for both CHD and scoliosis, possibly involving early laterality mechanisms (19).

TOF cases routinely have chest radiographs; in addition, based on the rapid growth of infants, scoliosis progression induces curve tends during this period. Severe curve progression causing significant compromise of the thoracic cavity may occur, leading ultimately to respiratory failure and death. Accordingly, the importance of this study has been cleared.

4. Conclusions

Based on our case report, a chest x-ray should be performed in any patient with cyanosis and especially TOF, and patients with gross motor disabilities, such as rolling, should be suspected of spinal disorder with other central nervous system complications. Therefore, TOF cases routinely undergo chest radiographs, and physicians should be aware of the potential for CS to provide early diagnosis and referral for orthopedic evaluation and treatment.

Ethical Considerations

Compliance with ethical guidelines

The study was conducted upon agreement of the Ethics Committee of Arak University of Medical Sciences and the informed consent of the patients is available upon request.

Funding

This research did not receive any grant from funding agencies in the public, commercial, or non-profit sectors.

Authors' contributions

All authors equally contributed to preparing this article.

Conflicts of interest

The authors declared no conflict of interest.

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