Case Report

Unilateral Exotropic Type II Duane's Retraction Syndrome Associated with Tetralogy of Fallot: A Case Report

Hajar Farvardin[1], Shahla Hoseini[2], Mehrnoosh Maalhagh[3], Majid Farvardin[4]
[1-4]Poostchi Ophthalmology Research Center, Department of Ophthalmology, School of Medicine, Shiraz University of Medical Sciences, Shiraz, Iran

PJO – Official Journal of Ophthalmological Society of Pakistan
Volume2023;39(4):361-363.
Doi: 10.36351/pjo.v39i4.1696

ABSTRACT

Duane's retraction syndrome is a rare congenital cranial disorder and special form of strabismus that manifests with the limitation of ocular movements in horizontal gaze, globe retraction, and narrowing of the palpebral fissure in adduction. Its cause is a teratogenic cranial innervational defect at the 4th-8th weeks of gestation. It is estimated that up to one third of these patients may have other congenital non-ocular anomalies. In this case, we present an association of unilateral exotropic type II Duane's retraction syndrome and tetralogy of Fallot in an eleven years old boy. He had no other systemic disease or congenital anomaly, and his family history was negative for strabismus and congenital heart disease. Duane's retraction syndrome can be associated with tetralogy of Fallot due to the matching time of gestational development of these two conditions, although this may be coincidental.

Key Words: Congenital anomalies, Duane's retraction syndrome, Tetralogy of Fallot.

How to Cite this Article: Farvardin H, Hoeini S, Maalhagh M, Farvardin M. Unilateral Exotropic Type II Duane's Retraction Syndrome Associated with Tetralogy of Fallot: A Case Report.

Correspondence: Majid Farvardin
Poostchi Ophthalmology Research Center, Department of Ophthalmology, School of Medicine, Shiraz University of Medical Sciences, Shiraz, Iran
Email: majidfarvardinjahromi@gmail.com

Received: July 20, 2023
Accepted: August 16, 2023

INTRODUCTION

Duane's retraction syndrome is a special form of strabismus that manifests with limitation of ocular movements in abduction and or adduction, globe retraction and narrowing of the palpebral fissure in adduction. It is categorized as a type of congenital cranial dys innervational disorders. The incidence of Duane's retraction syndrome is 0.1% in general population and it includes 1 – 4% of all cases of strabismus.[1] This syndrome is more common in females. Majority of the cases are sporadic. Few cases with autosomal dominant inheritance and incomplete penetrance have been reported.[1, 2]

Less than half of the patients with Duane's retraction syndrome may have other congenital ocular or systemic malformations.[1] Systemic anomalies have been reported in 6 – 33% of these patients and are more common in type I disorder. Frequent non-ocular anomalies in association with Duane's retraction syndrome is associated with bony skeleton, central nervous system, head anomalies, deafness, facial asymmetry, cleft palate and preauricular skin tags.[2, 3]

We present a case unilateral exotropic type II Duane's retraction syndrome with tetralogy of Fallot in an eleven years old boy. This case report is presented in accordance with the Declaration of Helsinki. Informed consent was taken from parent of the patient.

CASE REPORT

This eleven years old boy was born by cesarean

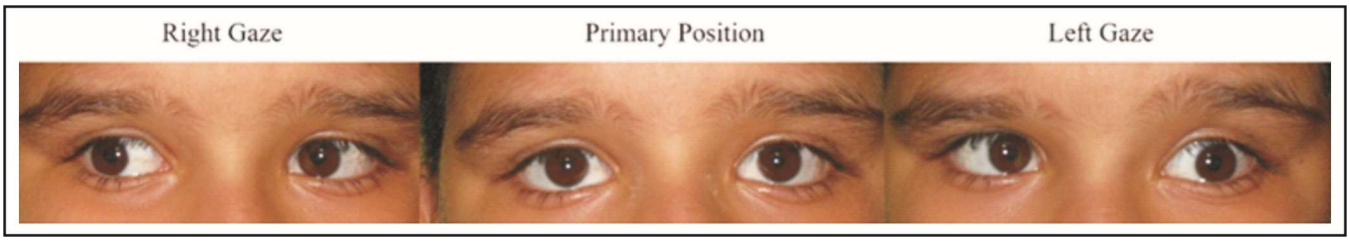

Figure 1: Picture of the patient was taken after the strabismus surgery (Bilateral lateral rectus recession). Extraocular movements of the patient showing limitations of abduction and adduction in the left eye. Deviation in the primary position and palpebral fissure narrowing were successfully corrected.

section (birth weight: 2700 grams) and had mild cyanosis at birth that progressed over time. He had no neurodevelopmental delay. His parents were not relative and had negative family history for any congenital anomaly and strabismus. Episodes of Tet spells developed at age of 1 year and cyanosis became constant. Pediatric cardiologists considered the diagnosis of tetralogy of Fallot according to the symptoms, chest radiography, electrocardiography and echocardiography. Successful cardiac surgery was done at the age of 2-years and his cardiac symptoms including cyanosis disappeared. His parents noticed ocular deviation later on and referred him to the strabismus clinic at 6 years of age. He had a compensatory face turn to the right side. Ocular motility examination showed 25 prism diopters exotropia in the primary position, limitation in adduction and narrowing of palpebral fissure in adduction (all in the left eye). The patient was classified as an exotropic type II Duane's retraction syndrome. Bilateral lateral rectus recession was done for the patient at age of 6 years. The last ocular examination was done at the age of 11years. Best-corrected visual acuity was 20/20 in the right eye and 20/30 in the affected left eye. Cycloplegic refraction was -0.75*115 diopters in the right eye and -0.5*55 diopters in the left eye. There was no face turn. He had mild limitations in adduction and abduction in the left eye (Figure 1). The result of dilated fundus examination was unremarkable in both eyes.

DISCUSSION

Duane's retraction syndrome is rarely associated with congenital cardiac anomalies. Few cases of dextrocardia, left sided aortic arch, patent ductus arteriosus, ventricular septal defect and atrial septal defect have been reported in association with Duane's retraction syndrome.[3-5] Tredici and von Noorden reported an association of Duane's retraction syndrome with two cases of tetralogy of Fallot but findings of the patients and type of Duane's retraction syndrome were not reported in their article.[6] Zare et al, reported an association of bilateral orthotropic Duane's retraction syndrome with bilateral iris-retinal coloboma, sensory hearing loss, inguinal hernia, and tetralogy of Fallot in a 10-year-old girl.[7] Both of these patients had multiple congenital anomalies including bilateral ocular anomalies. The patient presented here, however, had no other congenital anomaly besides Duane's retraction syndrome and tetralogy of Fallot.

Tetralogy of Fallot is one of the most common congenital heart diseases that lead to cyanosis due to its specific anatomic features: pulmonary outflow obstruction, ventricular septal defect, overriding the root of the aorta and hypertrophy of the right ventricle. Its prevalence is 3 out of 10000 live births and its etiology is multifactorial.[8]It is associated with maternal intake of retinoic acid, untreated maternal diabetes, phenylketonuria and some chromosomal anomalies including trisomy 21, 18 and 13 and microdeletions of chromosome22. It may be associated with noncardiac anomalies including forked ribs and scoliosis.[8]

The most accepted theory for pathophysiology of Duane's retraction syndrome is the absence of the abducent nucleus and abnormal innervation of the lateral rectus muscle. Also, there is abnormal innervation of the lateral rectus muscle by a branch of the oculomotor nerve which leads to globe retraction and narrowing of the palpebral fissure in adduction.[1]

The oculomotor cranial nerves and nuclei 3, 4 and 6 develop during the 5th-8th week and extraocular muscle innervation develops during the 4th-6th gestational weeks.[1,2] Pathologic process related to tetralogy of Fallot and other congenital heart disorders occurs during critical embryogenesis time of the 3rd to 6th gestational weeks.[9] Duane's retraction syndrome can be associated with tetralogy of Fallot due to the

matching time of gestational development of these two conditions. The same environmental or genetic teratogenic factor may lead to an association of Duane's retraction syndrome and tetralogy of Fallot in the same patient, although this may be coincidental.

Duane's retraction syndrome and tetralogy of Fallot may be associated even in the absence of other congenital anomalies. By reporting this case, we hope to increase the knowledge of pediatricians, pediatric cardiologists and ophthalmologists about this association. This will alert them for better examination of their patients to ensure on-time treatment.

Conflict of Interest: Authors declared no conflict of interest.

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Authors' Designation and Contribution

Hajar Farvardin; Resident: *Literature Search, Data Acquisition, Data Analysis, Manuscript Preparation, Manuscript Editing, Manuscript Review.*

Shahla Hoseini; Resident: *Literature Search, Data Acquisition, Manuscript Preparation.*

Mehrnoosh Maalhagh; Consultant Ophthalmologist: *Literature Search, Data Acquisition, Manuscript Preparation.*

Majid Farvardin; Professor: *Concepts, Design, Data Acquisition, Manuscript Editing, Manuscript Review.*