Congenital superior oblique palsy in a patient with VACTERL association

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Abstract:

Different types of ophthalmological defects have been reported to be accompanying vertebral anomalies, anal atresia, cardiac malformations, tracheoesophageal fistula, renal anomalies, and limb anomalies (VACTERL) association. A 7-year-old girl with a history of VACTERL association presented with upward drifting of the left eye and anomalous head posture to the right side and was diagnosed with congenital fourth nerve palsy. We report the first case representing a combination of congenital fourth nerve palsy with VACTERL association.

Keywords:

Congenital superior oblique palsy, strabismus, VACTERL

Introduction

Congenital superior oblique palsy (SOP) is characterized by ipsilateral hypertropia and contralateral head tilt. Despite the fact that this disorder is typically an isolated clinical entity without accompanying systemic diseases, congenital SOP has been reported in patients with PHACE syndrome,[1] olfactory hypoplasia,[2] and Holt–Oram syndrome.[3]

The VATER association was first identified in 1973 as the coexistence of five defects, i.e. vertebral anomalies (V), anal atresia (A), esophageal atresia and/or tracheaesophageal fistula, and radial and renal anomalies (R). The acronym VACTERL association was later introduced when cardiovascular anomalies (C) and limb defects (L) were also added.[4,5]

In this case report, we aimed to present a patient with VACTERL association concurrent with congenital SOP.

Case Report

The guardians of the patient provided informed consent for the publication of this case report and any related images. This case report complies with the tenets of the Declaration of Helsinki.

A 7-year-old girl was referred to the Strabismus Department at the Haydarpasa Numune Education and Research Hospital for evaluation of strabismus. Her prior medical history was relevant to the VACTERL association. She had multiple anomalies including esophageal atresia with tracheoesophageal fistula, anal atresia, scoliosis, syringomyelia, renal dysplasia, and cardiac transposition of great arteries. She had previously undergone two balloon dilatations for the esophageal and anal atresia and finally a laparoscopic Nissen fundoplication for her gastroesophageal reflux. Her parents noticed the abnormal head posture as early as the 1st year of life and denied any history of prior head trauma or a family history of strabismus.

On examination, she showed a constant head tilt to the right and a mild facial asymmetry existed. Although hemifacial microsomia has been reported to be associated with VACTERL anomaly, we considered it was not the case for our patient who showed no sign of ear or facial skeleton hypoplasia clinically or radiologically. Therefore, we related the mild facial asymmetry to the head posture resulting from the superior oblique paralysis. Uncorrected visual acuity was 20/20 in both the eyes and cycloplegic refraction

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showed + 1.75 D OD + 2.00 OS. The intraocular pressure was 16 mmHg bilaterally, and the pupils were equal and reactive. A relative afferent pupil defect was not observed. Anterior segment and dilated fundus examination were unremarkable in both the eyes. Objective fundus torsion could not be documented and subjective torsion could not be detected on the double Maddox rod test. Ocular motility examination revealed a left hypertropia of 8Δ in all primary up, down, and left gaze positions, 14Δ on the right gaze, and 8Δ esotropia at distance and near. The titmus fly test and the random dot stereotest were not reliable as the child could not cooperate enough, but we assumed that there was some form of fusion as the child had a compensatory head tilt. Ductions and versions revealed mild overelevation in adduction in the left eye, whereas fixating with the fellow eye indicated inferior oblique overaction [Figure 1]. A slight increase in hypertropia measured as 20Δ was noticed on the left head tilt, indicating a positive Bielschowsky sign. The child did not complain of diplopia and vertical vergence amplitudes were found 14Δ.

Axial T2-weighted magnetic resonance imaging was obtained with 0.25-mm thickness for the trochlear nerve and trochlear nerve and extraocular muscle sizes were viewed as normal on both the sides.

The patient underwent an uneventful strabismus surgery with left inferior oblique anteriorization. Postoperatively, there was a total correction of the hypertropia and the patient was 8Δ esotropic in the primary position. The anomalous head position was corrected [Figure 2].

Discussion

The exact pathogenesis remains unknown in the majority of cases with VATER/VACTERL association.[6,7] Although there has been one report showing mitochondrial cytopathy in one patient, VACTERL anomaly is commonly accepted as sporadic.[6]

Patients with the VACTERL association may have accompanying ophthalmic problems. These problems include ptosis, strabismus, myopia, hypermetropia, amblyopia, anisometropia, cloudy corneas, microcornea, congenital corneal anesthesia (i.e. recurrent erosions, keratitis, ulcers, and leukomas), anisocoria and heterochromia iridis, cataracts, and lacrimal system abnormalities.[8-10] Usually, the retina and posterior portion remain unaffected. A case of Coats' disease presence with the VATER association has been documented.[11] More recently, there has been one case report presenting a case

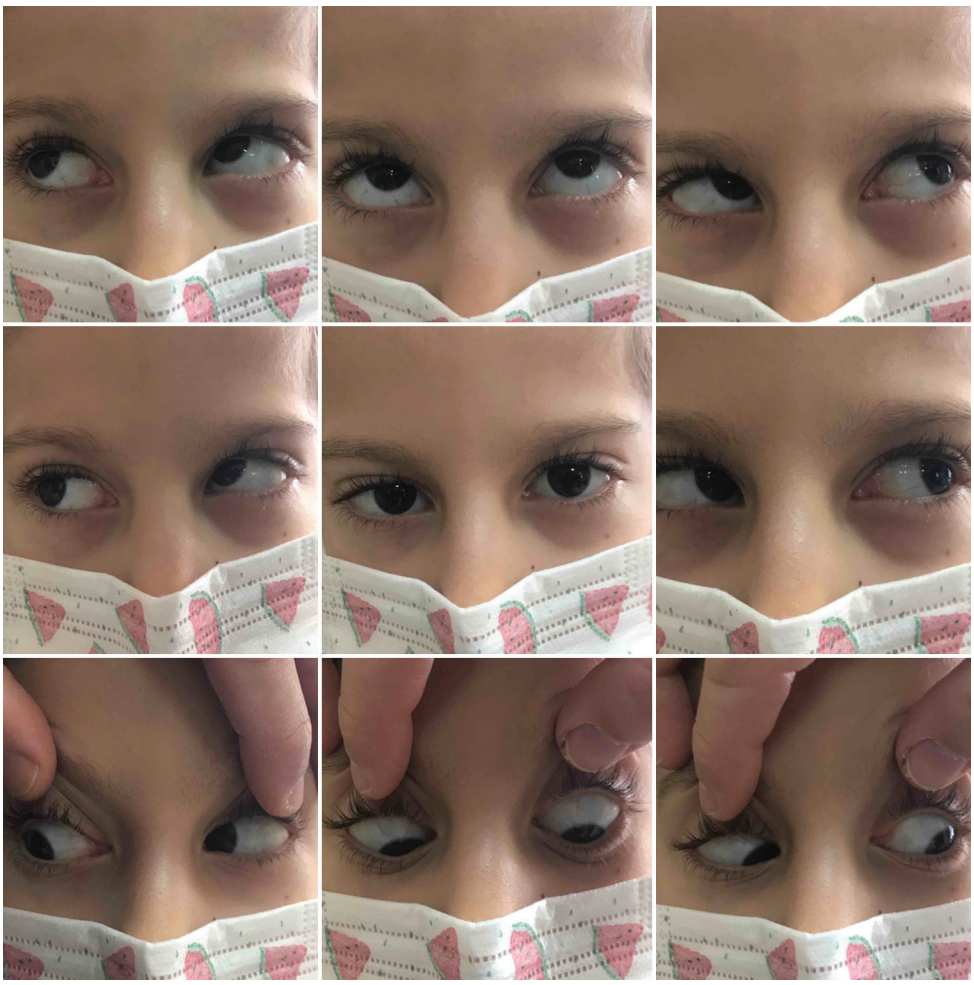

Figure 1: Preoperative photos of the patient in 9 positions of gaze

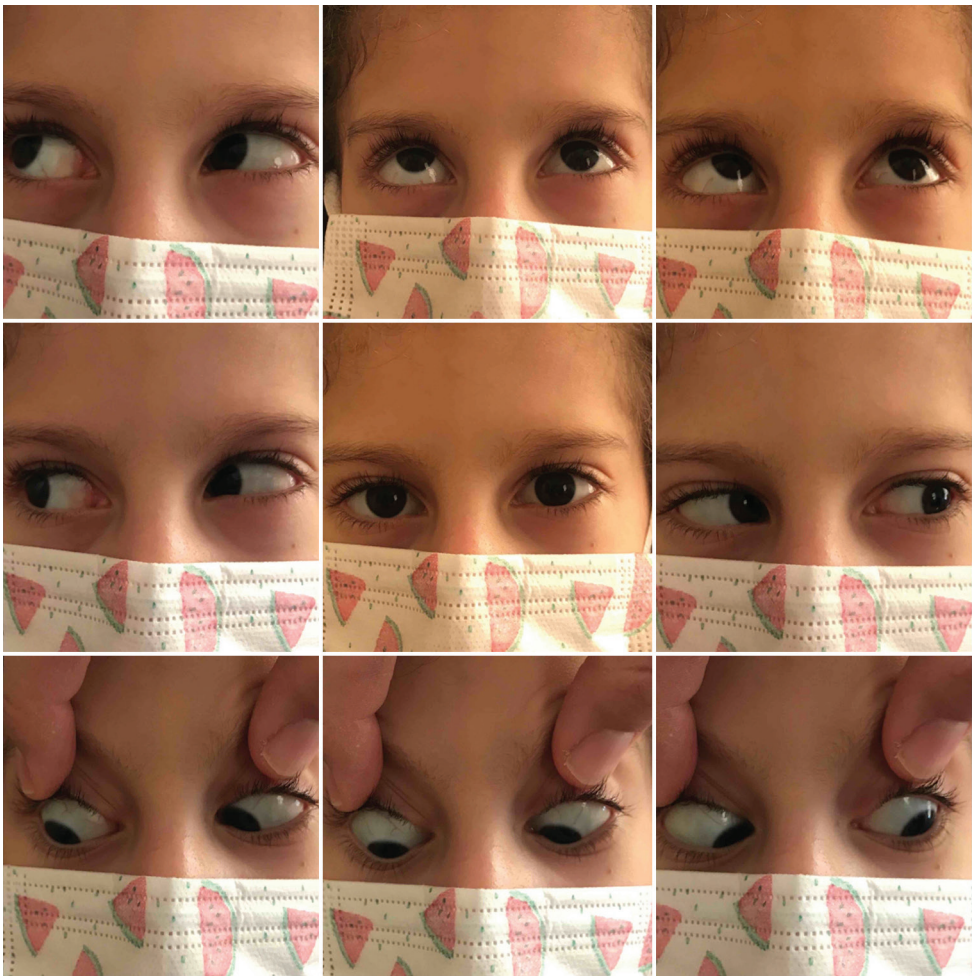

Figure 2: Postoperative 1st month photos of the patient in 9 positions of gaze

of Duane's retraction syndrome concurrent with VACTERL association.[12]

SOP has been reported in patients with PHACE syndrome and Holt–Oram syndrome,[1,3] but different forms of concurrent congenital cranial dysinnervation disorders such as Duane retraction syndrome, Brown syndrome, or congenital fibrosis of extraocular muscles have also been reported.[13-15]

Herein, we present a patient with VACTERL association who showed concurrent SOP which has not been previously documented. We believe that this case may make a contribution to what we know about the spectrum of ocular anomalies coexisting with the VACTERL association. In conclusion, it is important to consider the probability of numerous ocular and systemic disorders when assessing these patients.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the legal guardian has given his consent for images and other clinical information to be reported in the journal. The guardian understands that names and initials will not be published and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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