· Case report ·

Bilateral Brown's syndrome associated with nanophthalmos and generalized joint stiffness

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Abstract

• Brown's syndrome is characterized by absence or severe limitation of elevation in adduction with a positive forced duction test and minimal elevation deficit in abduction and primary position. Nanophthalmos is an uncommon congenital ocular malformation characterized by an extremely small eye. In this report, a case with bilateral Brown's syndrome and nanophthalmos combined with generalized joint stiffness was presented.

• KEYWORDS: Brown's syndrome; nanophthalmos; microphthalmos; generalized joint stiffness

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INTRODUCTION

Nanophthalmos is a part of heterogeneous small eye phenotypes varying from the extreme of anophthalmos to simple microphthalmos [1,2]. Representing a pure form of microphthalmos, nanophthalmos is characterized by a small eye without additional gross developmental ocular anomalies and is associated any neurological or other somatic abnormalities[1-6]. Nanophthalmos is diagnosis by a small cornea, a shallow anterior chamber, narrow anterior chamber angle, short axial length, axial hyperopia, thickened choroid and sclera. As the small nanophthalmic eyes are deeply set in orbits, the palpebral fissures are narrow [1,6].

Brown's syndrome, a form of anatomical strabismus consists of active and passive restriction of upward gaze in adduction, presents in both monocular and binocular motility [7-10]. Minimal elevation deficiency or normal elevation in abduction and primary position and a positive force duction test on elevation in adduction are main features of Brown's syndrome. The condition was explained by multiple factors such as abnormality of the superior oblique (SO) trochlea/tendon complex, short SO tendon, inferior orbital mechanical restriction and iatrogenic or other acquired factors such as trauma[11-15].

In this article, a patient had bilateral congenital Brown's syndrome and nanophthalmos associated with generalized joint stiffness was presented. According to our knowledge, presence of nanophthalmos and Brown's Syndrome in same patient had not been published before.

CASE REPORTS

A 9 year-old boy, a child of a consanguineous couple was first examined when he was 3. 5 years old. The best-corrected visual acuities (BCVA) were 0. 5 with +8. 50 +1. 50 × 80° in the right eye (RE) and 0. 2 with +9. 00 +0. 75 × 95° in the left eye LE by Snellen Chard. Suppression was presented on his(LE) on both Bagolini glasses and Worth four dot test (WFDT) and he did not have stereopsis neither with Lang nor with TNO test. His eyes were small deeply set in the orbital cavities and the palpebral fissures were narrow.

A slit lamp examination revealed a shallow anterior chamber in both eyes with anatomically normal iris and lens. Funduscopic examinations performed with both +90 diopter (D) and +78D that relieved normal optic nerve, retinal vessels and other structures.

He was orthotropic in the primary position without anomalous head posturing. However, he had no excursion from midline in adduction. Severely (-4) restrictions of elevation were observed in adduction, minimal restriction of (-1 out of 4) elevation in primary position and normal elevation in abduction on both eyes were observed.

According to his medical history, risk factors for muscle restriction such as trauma, ocular and systemic inflammatory diseases were not noticed. Nevertheless, he had generalized joint stiffness mainly on achilles tendon that cause walking difficulties. Result of his systemic evaluation he had significant restriction of extension all phalanxes, both knees and wrists. Rather than generalized joint stiffness and extension difficulties of extremities, none of other systemic developmental and laboratory abnormalities was found. He has normal height and weight and normal motor development according to his age.

During the follow up period, with the help of occlusion therapy for amblyopia, his BCVA were improved to 8/10 for both eyes with +8. 00 and +8. 00 +0. 50 × 80° in RE and LE respectively at the age of seven. He exhibited a fusional response on both Bagolini glasses and Worth four light testing (WFLT) at near and distant vision. He showed developed 200 arc/sec on Landot test and 240 arc/sec on

TNO test. However, his ocular motility disorder had not changed during follow up period. When he became old enough to perform forced duction test under topical anesthesia, significant restriction on elevation in adduction but normal elevation both primer position and abduction on his both eyes were observed.

Anterior chamber depths and axial lengths were measured with A mode Ultrasound. The anterior chamber depths were 2.59mm in RE and 2.57mm in LE, lens thickness was 3.99mm in RE and 3.96mm in LE. Axial lengths were 18.51mm in RE and 18.87mm in LE. Horizontal corneal diameters were 9.5mm in RE and 9.5mm in LE measured by autokeratorefractometer. The keratometry readings were 44.00 & 46.75 in RE and 44.50& 45.50 in the LE.

B mode ultrasound was showed small globs, with normal optic nerves without other ocular anomalies on both eyes.

He was able to walk normally after he had undergone achiloplasty as an elongation operation on his both achilles tendons, in the same session, at the age of five for his walking difficulties.

He had undergone computed tomographic (CT) scanning and MRI of both orbit and brain when he was 6 years old without help of sedation. Both CT scanning and MRI (Figure 1) showed bilateral small glob size, with normal size of orbital cavities normal appearing optic nerve. On MRI evaluation normal anatomic insertion sides of both SO, normal trochlear positions on both eyes were observed. However, SO tendon horizontal widths were measured as 5mm in RE and 6mm in LE. Neither in trochlear area nor in the other part of orbit factors may cause mechanical restriction such as scar tissues, infiltrative or inflammatory process or orbital cysts were observed. No intracranial abnormalities were found in MRI.

His mother, a cousin of his father had a normal phenotype and ophthalmologic findings. However, his father had small eye deeply set in orbit. The BCVAs were 10/10 with +0.50 × 180° in the RE and +0.50 × 180° in the LE. A scan ultrasound biometry showed and axial length of 21.34mm in the RE and 21.06mm in the LE. Anterior chamber depth was 3.10mm in the RE and 2.94mm in the LE. Lens thickness was 3.81mm and 3.70mm in RE and LE respectively. The keratometry readings were 44.50 and 44.00 in RE and 44.75 and 44.50 in LE. Fusion was normal both tested with Bagolini glasses and WFLT for both near and distant visions. He had stereopsis 120 arc/sec for TNO and 100 arc/sec for Landot test. On ocular motility examination, he had neither deviations nor restrictions. He had no systemic illness particularly arthritis, inflammation and joint stiffness. According to his family history, no one had small eyes and ocular motility disorders.

DISCUSSION

Nanophthalmos is a rare ocular anomaly characterized by a small eye without any other ocular or systemic gross malformation. Clinical features included a deeply set globe in orbit, a narrow palpebral fissure, a smaller than normal corneal diameter of 11.0mm, a shallow anterior chamber, thickened choroid and sclera, a short axial length (<20mm), but normal size of lens and a high lens eye volume ratio [1,4,10].

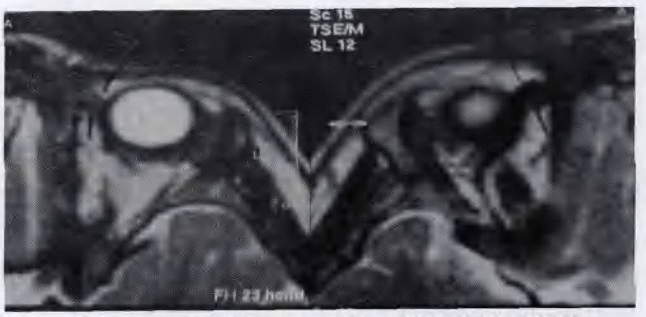

Figure 1 Increased SO tendon width of RE-LE in MRI

Although high hyperopia due to short axial length may consider a defining feature of nanophthalmos, refraction is determined by the combined refractive power of the cornea, crystalline lens and axial length. Hence, the degree of hyperopia is variable and in some patient, unusually myopia may develop [1,5,6].

Our patient had small corneal diameter, narrow anterior chamber, short axial length, deeply set in orbit and narrow palpebral fissure that are diagnostic features of nanophthalmos. Although he had short axial length that might cause high hyperopia, our patient also had high keratometric values as 44.00-46.75 in RE and 44.50-45.50 in LE, that prevents very high hyperopia in contrary to the general expectations. In nanophthalmos high keratometry as high as 51.00 diopters had been published [11]. He also had limitation of elevation in adduction with a normal elevation in abduction and positive force duction test on elevation in adduction that are a clinical definitional characteristic of Brown's Syndrome [7-9,12-15].

Surgical treatment Brown's syndrome has been advocated in case of patient have abnormal head position, manifest hypotropia and exaggerated downshoot in adduction [9,13,14]. None of those factors was evaluated in our patient. Therefore, we preferred only observation without surgical interventions for Brown's Syndrome and occlusion therapy for treatment of amblyopia. With the occlusion therapy not only his BCVAs but also his single binocular visions such as fusion and stereopsis were improved.

Both of his MRI and CT revealed small glob without associated any other anomalies such as orbital cysts.

The dysfunction of the superior oblique requires an effective interplay of multiple components, including innervational status, mechanical muscle abnormalities, such as laxity, scar tissue, infiltrative process. Insertion of SO has been shown to vary and to affect the muscle's pulling direction and this function. Since the trochlea is the functional mechanical origin of the SO, change in trochlear position in the orbit such as in craniofacial syndromes also alters the pulling direction of the muscle. Factors involve free movement in the trochlea complex including inflammations, trauma to the trochlear area and other restrictive phenomena have been presumed to compromise muscle function that cause acquired Brown's Syndrome [7,15-19]. High resolution MRI requires the patient to maintain gaze on stable for several minutes that is possible for only cooperative children. Therefore, we had to postpone MRI

examination until our patient to get mature enough. According to our patients both MRI and CT revealed short ocular axial length, small globe in normal shape and size of orbital cavity without any cystic component. We did not observe any anatomical abnormalities on trochlea including abnormal location, fibrous bands and mechanical restrictive factors such as scar tissues. But on MRI of our patient, the size of SO tendon width was measured as 6mm in RE and 5 mm in LE both of which were significantly higher than the normal adult value of 2.4 ±0.4mm that may cause mechanical restriction of muscle function in adduction [20]. Maggi and Maggi [15] reported that they observed thickening of SO tendon during the operation in the case of Brown's syndrome, but did not give exact measurement. According to their observation, they hypothesized that the thickening of the tendon could be the result of a lacking or delayed patency of the trochlea. That may prevent free sliding of tendon at the side of trochlea. Even we did not performe exploration as Maggi and Maggi [15], we observed similar findings on non-invasive method of MRI evaluation in our patient.

A possibility of a genetic defects such as X-linked, autosomal recessive or dominant penetrance or mutations in genes had been pointed out of nanophthalmos [4,21]. Even the patient is a child of consanguineous marriage, and due to presence of nanophthalmos in father of our case, it seems more likely that the penetrance of nanophthalmos in our case is autosomal dominant.

Histological anomalies in nanophthalmic eye that cause increased scleral thickening have been described by different authors. The collagen lamella has been found to be disorganized. It is postulated that collagen bundles are in some areas pack more closely and the size of the collagen fibers seems to vary more than in the normal size. Also a decreased amount of glycosaminoglycans and an elevated fibronectin content, abnormal metabolism of the glycosaminoglycans that might cause the sclera to be less elastic [2, 6, 22-25].

Not only restriction on ocular function, but also restriction in extension on all phalanxes, both knees, whist and achilles tendons on both legs may be some parts of same intrinsic disorders of connective tissues system. That may be also related abnormal histological structure of sclera in nanophthalmic eyes.

As far as we know, our patient is the first case in the literature that has bilateral Brown's syndrome, nanophthalmos and generalized joint stiffness presented together. All of these may be seen as a coincidence or may be related a part of a similar connective tissue disorder such as a syndrome.

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双眼Brown's综合征合并小眼球及系统性关节僵硬

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摘要

Brown's综合征(上斜肌腱鞘综合征)是指患眼在内转位时不能上转或上转严重受限,内转时被动牵拉试验阳性,在外转和第一眼位是上转受限程度极小。小眼球是一种不常见的先天性眼畸形,表现为眼球极小。本论文报告1例双眼Brown's综合征及小眼球合并系统性关节僵硬。

关键词:Brown's综合征;小眼球;小眼;系统性关节僵硬