Tilted Disc Syndrome in Two Siblings of Different Sexes: Case Report

Farklı Cinsiyetteki İki Kardeşte Tilted Disk Sendromu

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ABSTRACT Tilted disc syndrome (TDS) is a congenital anomaly occurring from embryonic dysgenesis, characterized by a variable ophthalmoscopic appearance and numerous associated visual deficits and complications. The syndrome occcurs similarly in men and women in 1 to 2% of the population. It is generally accepted that, the syndrome shows no hereditary patterns. We present TDS with similar laterality and features of the findings in a sister and brother of 8 person family. Full ophthalmic examination including cover tests and cranial-orbital magnetic rezonans imaging was performed. We have detected similarity of clinical and radiological findings, and laterality of findings such as lower visual acuity, higher astigmatism and exotropia to right eye in both siblings.

Key Words: Optic nerve diseases; optic disk

ÖZET Tilted disk sendromu (TDS) embriyonik disgenezi sonucu ortaya çıkan çeşitli oftalmoskopik görünüm, çok sayıda ilişkili görme alanı defekti ve komplikasyonlarla karakterize konjenital bir anomalidir. Kadın ve erkeklerde eşit sıklıkta olup, toplumda görülme oranı %1 ile %2 arasında değişmektedir. Sendromun herediter bir patern göstermediği genel olarak kabul görmektedir. Biz bulguların benzer lateralite gösterdiği, 8 kişilik bir ailenin farklı cinsiyetteki 2 kardeşinde görülen TDS olgularını sunuyoruz. Kapama testleri ve kranial-orbital magnetik rezonans görüntülemeyi içeren tam göz muayenesi yapıldı. Klinik ve radyolojik bulgularda benzerlik ve düşük görme keskinliği, yüksek astigmatizma ve ekzotropya gibi bulguların her iki kardeşte de sağ göze lateralize olduğu tespit edildi.

Anahtar Kelimeler: Optik sinir hastalıkları; optik disk

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Tilted disc syndrome (TDS) is a congenital anomaly characterized by a variable ophthalmoscopic appearance and numerous associated visual deficits and complications.[1] The most common findings in TDS include a tilted appearance of the disc, situs inversus, hypoplasia of the retina, retina pigment epithelium and choroid, and conus of the inferior or inferior nasal aspect of the disc with posterior ectasia and staphyloma of this area. The syndrome occurs similarly in men and women in 1 to 2% of the population and shows no hereditary patterns.[2] We present TDS with similar laterality and features of the findings in a sister and brother of 8 member family.

CASE REPORTS

Written informed consent was obtained from each patient before full examination.

CASE 1

In full ophthalmic examination of 24 years old male, the best corrected visual acuity (BCVA) was 20/200 in the right, 20/50 in the left eye. The manifest refraction was -1.0 (-2.50 x 140) in the right and -0.50 (-2.0 x 110) in the left. Fundus examination showed vertical-temporal tilted disc bilaterally (Figure 1a, 1b). The retinal vessels were deviated towards the crescent. The reduction of retinal pigment was shown in the posterior pole. There was nystagmus and right exotropia in primary position. Cover tests showed approximately 15-20 PD right exotropia (Figure 1c) and dissociated vertical deviation. Ocular movements were free in all directions. The cranial magnetic resonance imaging (MRI) was normal. Orbital MRI showed abnormal glob curvature at the posterior region in each eye (Figure 1d).

CASE 2

In full ophthalmic examination of 29 years old female, the BCVA was 20/200 in the right, 20/40 in the left eye. The manifest refraction was -0.25 (-3.0 x 160) in the right, and -0.50 (-1.50 x 150) in the left. Fundus examination showed vertical-temporal tilted disc bilaterally (Figure 2a, 2b).

The retinal vessels were deviated towards the crescent. The reduction of retinal pigment was shown in the posterior pole. There was nystagmus and right exotropia in primary position. Cover tests with prisma showed approximately 40-45 PD right exotropia (Figure 2c) and dissociated vertical deviation. Ocular movements were restricted -2 to nasal side at the right eye and, free in other directions. Cranial MRI was normal. Orbital MRI showed abnormal glob curvature at the posterior region in each eye (Figure 2d).

DISCUSSION

Tilted disc syndrome is a congenital optic nerve coloboma occurring from embryonic dysgenesis. It is strongly suspected to result from incomplete closure of the fetal fissure at 6 weeks gestation with the formation of a typical coloboma of the disc, peripapillary retina, RPE, and choroid.[2] It is important to know the clinical characteristics of TDS for differential diagnosis and to avoid unnecessary neuro-radiologic imaging.[3]

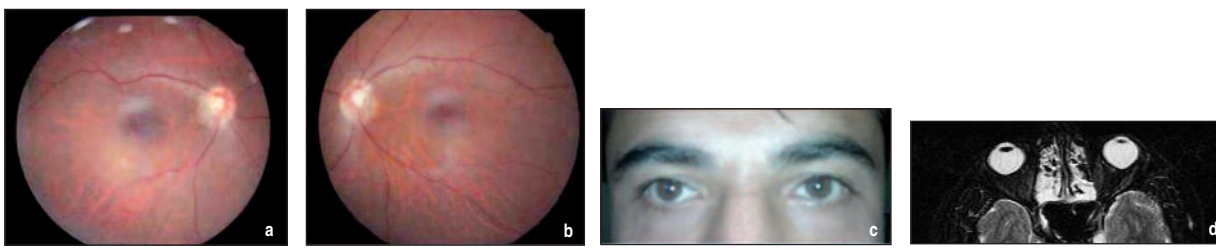

FIGURE 1: a, b) Appearance of tilted disc in right and left eyes of Case 1, **c)** Right exotropia, **d)** Abnormal glob curvature at the posterior region of glob in magnetic resonance imaging.
(See color figure at http://www.turkiyeklinikleri.com/journal/oftalmoloji-dergisi/1300-0365/)

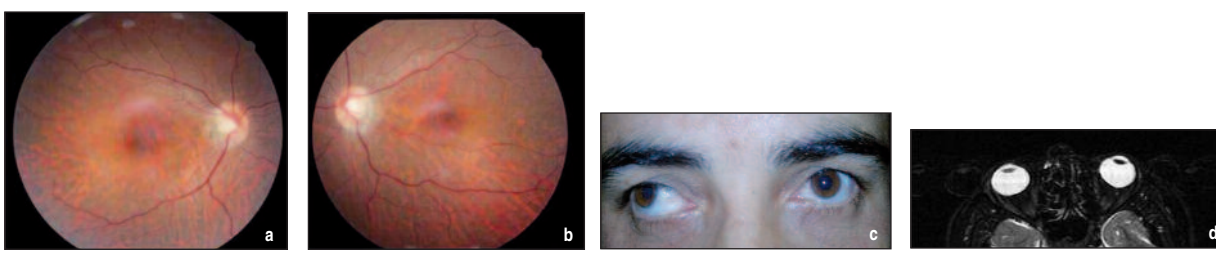

FIGURE 2: a, b) Appearance of tilted disc in right and left eyes of Case 2, **c)** Right exotropia, **d)** Abnormal glob curvature at the posterior region of glob in magnetic resonance imaging.
(See color figure at http://www.turkiyeklinikleri.com/journal/oftalmoloji-dergisi/1300-0365/)

It is generally accepted that, the syndrome shows no hereditary patterns.[2] The heredity of the conditions is not yet established, perhaps because it may easily be missed in family members of affected individuals, hence the lack of reports of the familial nature of the syndrome. Single clinical features like inferior crescent or transposition of the optic disc have been reported as genetically determined with various inheritance patterns.[4] On the other hand , Riise believes that the tilted disc syndrome can be familial, with a polymeric mode of inheritance similar to that refraction anomalies.[4] Bottoni et al. reported on a family with TDS characterized by inferonasal retinal ectasia, with linear-like lacquer cracks in three consecutive generations.[5] They demonstrated that the presence of the trait in three consecutive generations suggests an autosomal dominant mode of inheritence, although in the patients with variable expression. There are a few reports of X-linked TDS in the past. Hittner et al. identified a family with x-linked recessive congenital stationary night blindness, myopia, tilted discs, decreased vision, nystagmus, strabismus, visual field defects, abnormal fundus appearance, and abnormal electrophysiological tests.[6] Heckenlively et al. reported 10 patients with typical findings of congenital stationary night blindness including congenital nonprogressive nyctalopia, full visual fields consistent with myopia, low vision, strabismus, abnormal electroretinogram, temporal tilted disc.[7]

In our report, we have detected similarity of clinical and radiological findings, and laterality of findings such as lower visual acuity, higher astigmatism and exotropia to right eye in both siblings. X-linked inheritance was not considered because of different sexes of siblings. In summary, we believe that this may be the first report of TDS occurring in two siblings of different sexes having similar features. It is also noteworthy and meaningful for genetic inheretence. We think it will be helpful as a data to further genetic investigations in the future.

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