

## Tilted Disc Syndrome Mimicking Neurological Disorder: A Rare Case Report

DrShallu Samria<sup>1</sup>, DrPriyanka Gupta<sup>2</sup>, DrSaurabh Gupta<sup>3</sup>,  
DrVipin Kumar Bakshi<sup>4</sup>

<sup>1</sup>Senior Resident, Department of Ophthalmology, Adesh Institute of Medical Sciences and Research, Bathinda

<sup>2</sup>Assistant Professor, Department of Ophthalmology, Adesh Institute of Medical Sciences and Research, Bathinda

<sup>3</sup>Assistant Professor, Department of General Surgery, Adesh Institute of Medical Sciences and Research, Bathinda

<sup>4</sup>Assistant Professor, Department of Radiodiagnosis, Adesh Institute of Medical Sciences and Research, Bathinda

Corresponding author: Dr. Priyanka Gupta

---

**Abstract:** Tilted disc syndrome is a congenital anomaly of the eye characterized by the inferior or inferonasal tilting of the optic disc, congenital conus, ectasia, situs inversus of the retinal vessels at the level of the optic disc, myopic astigmatism and visual field defects. There is also thinning of the retinal pigment epithelium and choroid in the inferior nasal fundus that may be ophthalmologically visible as fundus depigmentation. This syndrome occurs equally in men and women in 1-2% of the population. It is generally accepted that the syndrome shows no hereditary patterns. We present a case of 45 year female with tilted disc syndrome with bilateral situs inversus and inferonasal staphyloma. Typical fundus picture, optical coherence tomography, visual fields, B scan ultrasonography and MRI of the patient would be instructive to a clinician.

**Keywords:** Tilted disc syndrome, situs inversus, staphyloma

---

Date of Submission: 04-11-2019

Date of Acceptance: 20-11-2019

---

### I. Introduction

Tilted disc syndrome is a congenital anomaly of the eye characterized by the inferior or inferonasal tilting of the optic disc, congenital conus, ectasia and thinning of the choroid and retinal pigment epithelium of the inferior or inferonasal fundus, situs inversus of the retinal vessels at the level of the optic disc, myopic astigmatism and visual field defects.<sup>1-4</sup> The tilted disc syndrome was first clearly described in 1944.<sup>5</sup> Other terms used to describe discs with a similar appearance are Fuchs's coloboma, congenital crescents, conus, dysversion of the optic nerve head, and situs inversus.<sup>6</sup> The syndrome occurs similarly in men and women in 1 to 2% of the population and shows no hereditary patterns.<sup>7</sup> The aetiology of tilted disc syndrome is still controversial. Theories of the aetiology of the syndrome suggest that it may result from malclosure of the embryonic optic fissure, leading to a coloboma of the inferior fundus. The most common type of field defect in tilted disc syndrome is an upper temporal defect which does not respect the vertical midline. The spectrum of changes in tilted disc syndrome ranges from a minimal situs inversus of the vessels to a full coloboma of the inferior fundus.<sup>4</sup>

We present a case of 45 year female with tilted disc syndrome with bilateral situs inversus and inferonasal staphyloma. We believe that typical fundus picture, visual fields, B scan ultrasonography and MRI of the patient would be instructive to the clinicians.

### II. Case Report

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

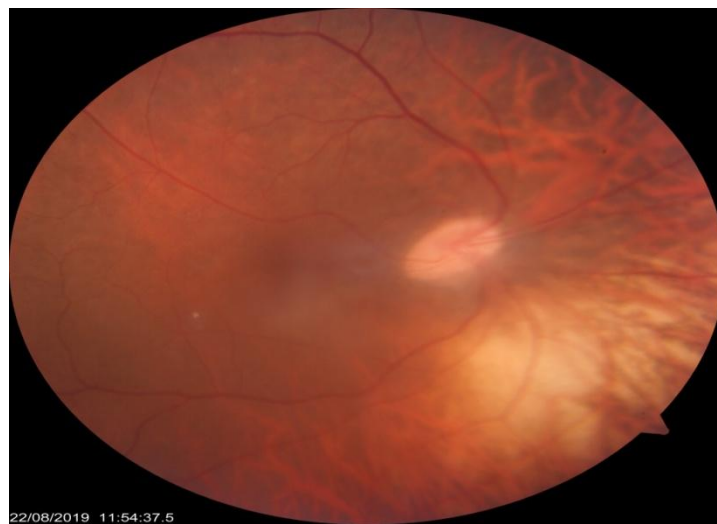
A 45 year old lady presented to ophthalmic OPD of Adesh Institute of Medical Sciences and Research, Bathinda with complaints of diminished vision more at night since childhood. She has come for the opinion of cataract surgery thinking cataract to be the cause of her diminished vision. There was no significant medical and surgical history. Significant family history of high refractive error was present. Her daughter was also high myope for which she underwent LASIK in both eyes.

Her complete ophthalmic examination was done. Her uncorrected distance visual acuity on Snellen's chart was 20/120 P in the right eye and 20/200 in the left eye. Her vision improved to 20/60 in both eyes with -7.00 D spherical in the right eye and -7.00 D spherical /-1.50 D cylinder at 60 degrees. Colour vision was

normal. Visual field examination was performed using Humphrey visual field testing both with correction and without correction. First visual fields without correction showed superotemporal field defects not respecting the vertical midline which corresponded with the inferonasal staphyloma. As these visual fields were mimicking neurological disorders hence MRI Brain was advised which came out to be normal. Second visual fields with correction were also done which nullified the prior visual field changes.

The pupils were dilated with tropicamide 0.5%. Her anterior segment examination revealed minimal cataractous changes in both eyes in the form of nuclear sclerosis grade 1. Hence dilated fundus examination was done to look for the cause of diminished vision. Fundus examination showed superotemporal tilted discs in both eyes. Superotemporal part of disc was elevated and inferonasal region was depressed. Typical situs inversus was present. The direction of emergence of retinal vessels was typically nasalward followed by an acute temporal bend. There was thinning of the retinal pigment epithelium and choroid in the inferior nasal fundus. Inferonasal staphyloma was present in both eyes. [Fig 1 and 2]

**FIGURE 1** and 2 shows fundus photo of both right and left eye respectively showing bilateral superotemporal tilted discs with inferonasal hypopigmentation and staphyloma. Situs inversus is also well noted in both eyes.



**FIGURE 2**



Optical Coherence Tomography (OCT) derived RNFL thickness was significantly thinned in inferonasal quadrants of this patient. Foveal contour was maintained. Figure 3 shows the OCT pictures of macula. Patient was then sent to the radiology department for B scan ultrasonography. B scan showed oblique insertion of the optic nerve and inferonasal staphyloma. (Figure 4,5)

FIGURE 3 shows OCT macula pictures of both left and right eye respectively. These pictures show normal foveal contour and thickness but abnormal alignment of macula in both eyes.

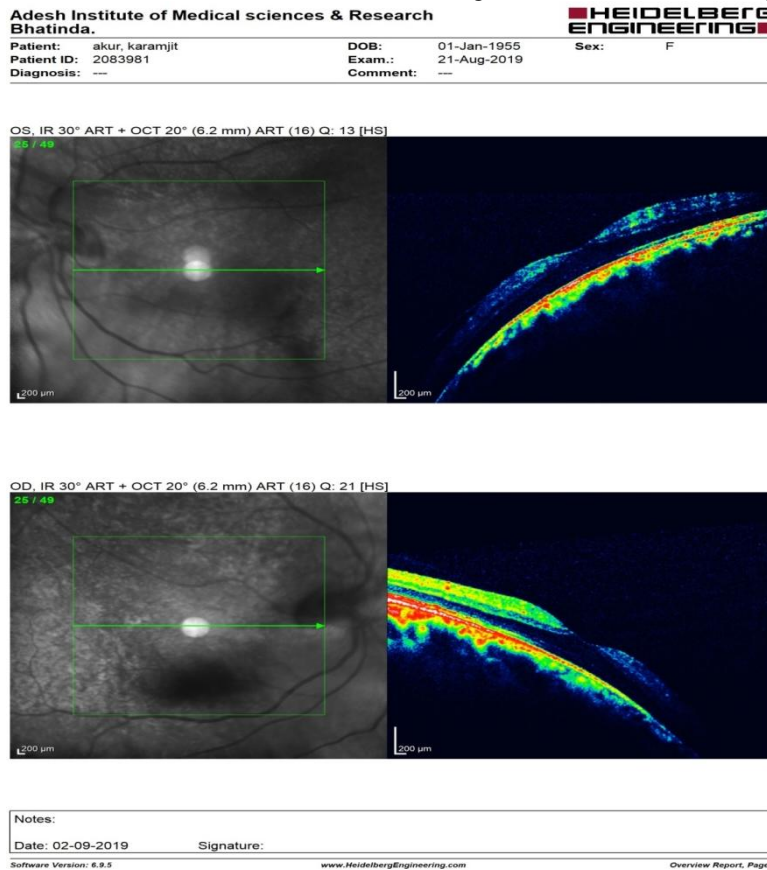
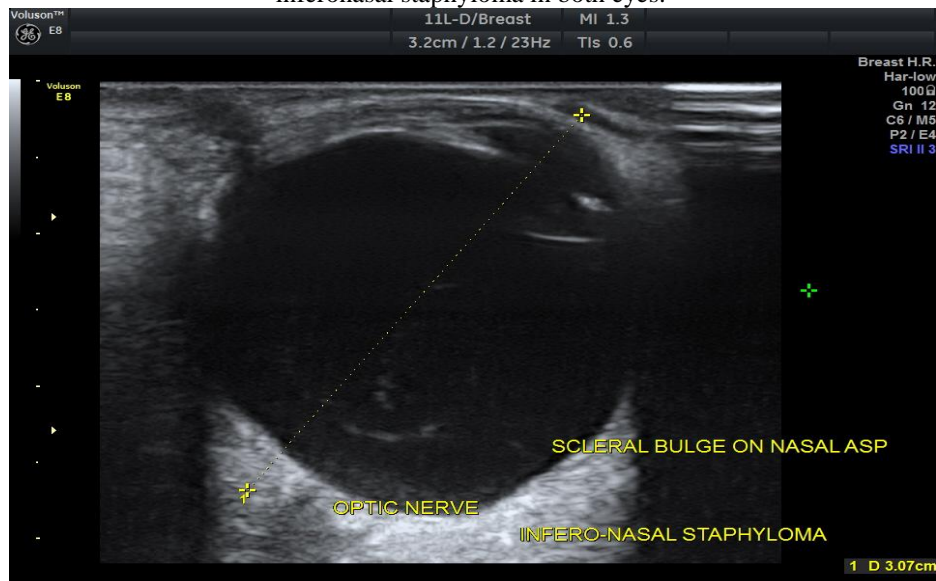
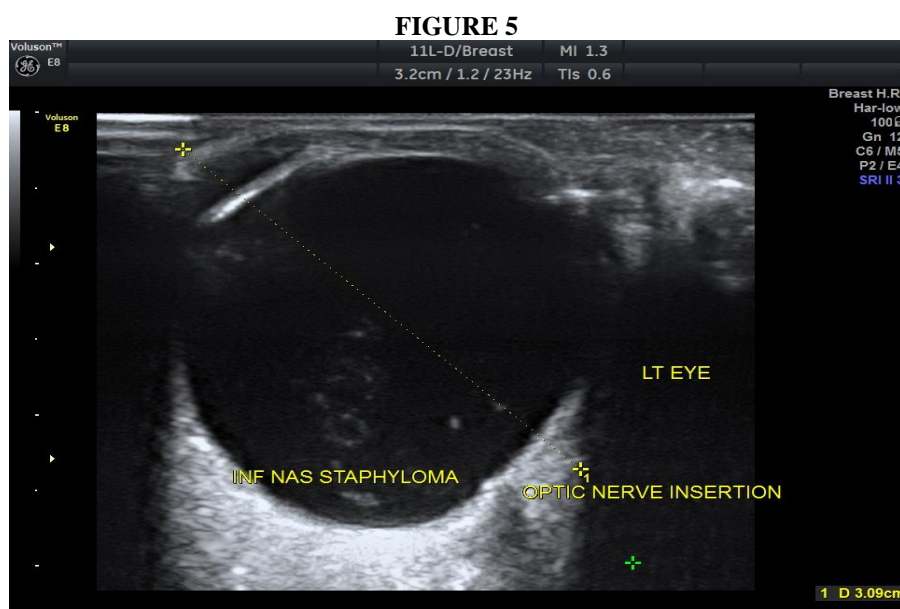


FIGURE 4 and 5 shows B scan of right eye and left eye respectively with oblique insertion of optic nerve and inferonasal staphyloma in both eyes.





### III. Discussion

Tilted disc syndrome is a developmental abnormality in the embryonic retina which may result in hypoplasia of the retinal pigment epithelium and neural elements as well as an anomaly of the disc.<sup>8</sup> It is generally accepted that tilted disc syndrome shows no hereditary patterns.<sup>1</sup> But in our study some form of hereditary pattern was found. Both mother and daughter were high myopes. Mother was having high myopia of -7 D in both eyes with tilted disc syndrome and daughter was also high myope for which she had undergone LASIK in both the eyes. This hereditary pattern was recorded in previous studies as well. Riise believed that tilted disc syndrome can be familial with polymeric mode of inheritance.<sup>2</sup> Bottoni et al reported tilted disc syndrome in three consecutive generations of a family.<sup>9</sup> The type of field defect found in our study was superotemporal which was not respecting the vertical midline. Similar field defects were also supported in previous studies done by Riise in 1975, Giuffre in 1986, Vuori et al in 2008 and many others.<sup>(2, 4, 10)</sup>

Fundus examination of our patient revealed bilateral superotemporal tilted discs with inferonasal hypopigmentation and staphyloma. Bilateral tilted discs was also demonstrated by Ozyol et al in 2014 and Williams A et al in 2005.<sup>(7,11)</sup> Bilateral Inferonasal hypopigmentation was also seen by Young et al in 1976 in all cases with field defects.<sup>3</sup> OCT scan of our patient revealed inferonasal thinning of RNFL and abnormal alignment of macula. In contrast to RNFL thickness, contour and thickness of macula was normal in our patient. These OCT findings were supported by a study done in 2009 by Moschos et al. He showed significant RNFL thinning in patients with tilted disc syndrome and normal mean value of foveal thickness.<sup>8</sup> B scan of our patient showed tilted discs and inferonasal staphyloma in both eyes. This was supported by a study done by Phu J et al in 2017.<sup>12</sup>

### IV. Conclusion

Patients with tilted disc syndrome can present with conditions mimicking neurological disorders. Such patients should be carefully evaluated for optic nerve abnormalities with false visual field deterioration.

Visual field defects in patients with TDS can be well nullified by giving proper glasses prescription prior to perimetry contrary to no such improvement in the neurological cases. Typical fundus picture, typical visual fields, B scan ultrasonography and MRI would be instructive to a clinician.

### FINANCIAL SUPPORT AND SPONSORSHIP

Nil.

### CONFLICTS OF INTEREST

There are no conflicts of interest.

### References

- [1]. Apple DJ, Rabb MF & Walsh PM (1982): Congenital anomalies of the optic disc. *SurvOphthalmol* 27: 3–41.
- [2]. Riise D (1975): Nasal fundus ectasia. *ActaOphthalmol* 53: 3–108.
- [3]. Young SE, Walsh FB & Knox DL (1976): Tilted disc syndrome. *Am J Ophthalmol* 82: 16–23.
- [4]. Vuori ML, MantyjarviM: Tilted disc syndrome may mimic false visual field deterioration. *ActaOphthalmol*. 2008; 86: 622-625.
- [5]. Rucker CW (1944) Bitemporal defects in the visual fi elds resulting from developmental anomalies of the optic discs. *Archives of Ophthalmology*, **32**, 56–9.
- [6]. Dorrell D: The tilted disc. *British Journal of Ophthalmology*, 1978, 62, 16-20.
- [7]. Ozyol E, Ozyol P, Duman R: Tilted Disc Syndrome in Two Siblings of Different Sexes: Case Report. *TurkiyeKlinikleri J Ophthalmol* 2014; 23(1):57-9.
- [8]. Moschos MM, Triglianos A, Rotsos T, Papadimitriou S et al : Tilted disc syndrome: an OCT and mfERG study. *Doc Ophthalmol*(2009) 119:23-8.
- [9]. Bottoni FG, Eggink CA, Cruysberg JR, Verbeek AM. Dominant inherited tilted disc syndrome and lacquer cracks. *Eye (Lond)* 1990; 4(Pt 3):504-9.
- [10]. Giuffre G (1986): The spectrum of visual field defects in tilted disc syndrome. *Neuro-ophthalmology* 6: 239–246.
- [11]. Williams A, David A: The tilted disc syndrome. *Practical Neurology*,2005, 5, 54-5.
- [12]. Phu J, Khuu KS, Yapp M, Assaad N et al: The value of visual field testing in the era of advanced imaging: clinical and psychophysical perspectives. *ClinExpOptom* 2017; 100: 313- 32.

Dr. Priyanka Gupta. “Tilted Disc Syndrome Mimicking Neurological Disorder:A Rare Case Report.” *IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)*, vol. 18, no. 11, 2019, pp 23-27.