

# CONGENITAL ANOMALIES OF OPTIC DISC: CLINICAL EVALUATION AND EFFECT ON VISUAL ACUITY

## Ophthalmology

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### Abstract:

**Aim:** Determine the prevalence, ocular association and effect on visual acuity of various congenital anomalies of optic disc.

**Study design:** Prospective observational study.

**Material and methods:** Study was conducted at a tertiary centre. In all patients slit lamp biomicroscopy, ophthalmoscopy, fundus photography, refraction, tonometry was performed

**Result:** Congenital anomalies of optic disc were detected in 94 patients with overall prevalence of 0.94% with maximum number of cases was of tilted disc. Overall the most common ocular association was refractive error. Overall 148 eyes of 94 cases were affected. Best corrected visual acuity within the range of 6/18 or better was found in 30.4 %, less than 6/18 to 6/60 was found in 29.7 %, less than 6/60 to 3/60 was found in 10.8 %, less than 3/60 to 1/60 was found in 14.2% and less than 1/60 in 14.9 %. All patients of myelinated nerve fibers, optic disc drusen and megalopapilla had a best corrected visual acuity 6/18 or better. The least visual acuity of <1/60 to PL was most commonly seen in optic nerve hypoplasia (40%), optic disc coloboma (22.8%) and tilted disc (13.3%) and in case of morning glory syndrome.

**Conclusion:** Congenital anomalies of optic disc some time mimic other acquired disorders. It is important to recognize even the relatively benign lesions in order to differentiate them from other more threatening lesions which they may clinically resemble. Accurate diagnosis and good management are essential to eliminate visual loss due to amblyopia or secondary to other complications. Benign lesions detected incidentally must be closely followed for complications and promptly addressed.

**Key words:** Congenital optic disc anomalies, tilted disc, myelinated nerve fibers, optic disc drusen. Coloboma.

### Introduction:

Congenital anomalies of the optic disc are fortunately rare and the etiology of most of them is unknown.<sup>1</sup> These conditions may be unilateral or bilateral and may underlie many cases of decreased vision, strabismus and nystagmus in childhood or

even present later in life.<sup>3</sup>

Some severe anomalies can lead to poor visual acuity while others are predominantly benign with none or a latent risk of visual loss and the type and extent of lesions dictate the visual acuity of the affected eye.<sup>2</sup>

Optic disc abnormalities may be associated with other congenital disorders of the eye and other systemic disorders often central nervous system malformations.<sup>3</sup> They may be complicated by retinal detachment, retinoblastoma, macular edema, choroidal neovascularisation and lipid exudation.<sup>4</sup>

Some of these conditions may be found on routine ophthalmic check-up but few of them often require a comprehensive evaluation and ancillary investigations like fluorescein angiography and ultrasonography which may be crucial to discover them.<sup>5</sup>

These anomalies also mimic other acquired disorders like glaucoma, papilloedema etc.<sup>2</sup> It is important to be able to recognize even the relatively benign lesions in order to differentiate them from other more threatening lesions or disease processes which they may clinically resemble.

The principal congenital abnormalities of the optic disc that can significantly impair visual function are optic nerve hypoplasia, optic disc coloboma and morning glory syndrome.<sup>6</sup> Optic disc pit and tilted disc syndrome have variable loss of visual acuity depending on the serous macular detachment.<sup>6</sup> Optic disc drusen and myelinated nerve fibers rarely present with poor vision.<sup>7</sup> Visual field changes of various types are seen in tilted disc syndrome, optic disc drusen, optic disc coloboma, optic disc pit, optic nerve hypoplasia etc. Systemic associations, predominantly CNS malformations are frequently associated with optic nerve hypoplasia, Aicardi syndrome, morning glory syndrome and optic disc coloboma.<sup>7</sup>

Accurate diagnosis and good management are essential to eliminate visual loss due to amblyopia or secondary to other complications.<sup>1</sup> Moreover benign lesions detected incidentally must be closely followed for complications and must be promptly addressed if they occur at any stage.<sup>1</sup>

## Material And Methods:

### Study Design

This study was a hospital based observational study, conducted between August 2012 and September 2014.

In our study, a total of 10,000 subjects those were attending in outpatient department for visual problems were examined ophthalmoscopically, of these only 93 patients

showed optic disc abnormalities.

A detailed comprehensive examination was carried out in all patients. Fundus examination done after full mydriasis for detecting the type and clinical pattern of various congenital anomalies of optic disc and for various complications associated with these anomalies. Stereoscopic fundus photograph was taken in all these patients by zeiss fundus camera. Special investigation: whenever needed:

- A. Visual field was analysed by Humphrey field analyser using 30-2 SITA fast strategy.
- B. B-Scan was done by (EZ-550 Sonomed) in cases like optic disc drusen to confirm the diagnosis.
- C. A Scan for axial length measurement.

Refractive status of eye analysed with automated refractometer.

The study was approved by the ethical committee of the institute and it is in accordance with the ethical standards of the responsible committee on human experimentation (institutional or regional) and with the Helsinki Declaration of 1975 that was revised in 2000.

## Results:

In this study out of 10,000 patients who were examined ophthalmoscopically in OPD, congenital anomalies of optic disc were detected in 94 patients with overall prevalence of 0.94%.

**Table-1** shows the percentage of each anomaly. Among the various optic disc anomalies studied, the highest prevalence was of tilted optic disc (0.46%).

**Table -2** shows the gender distribution in various congenital optic disc anomalies. Overall 46.80 % (44/94) cases were males and 53.2 % (50/94) cases were females. There was no sex preponderance in our study.

**Table-3** shows laterality wise distribution of cases. Bilateral involvement was found in 57.4 % (54/94) cases. All cases of optic disc drusen and megalopapilla and majority cases of optic nerve hypoplasia (66.6%) and tilted disc (63%) were bilateral. All cases of optic disc pit, morning glory

syndrome and majority (63.6%) cases of myelinated nerve fibers were unilateral.

Table 4 shows the BCVA of affected eye in various congenital anomalies of optic disc. 148 eyes of 94 cases were affected. Best corrected visual acuity within the range of 6/18 or better was found in 30.4 % (45/148 eyes), less than 6/18 to 6/60 was found in 29.7 % (44/148), less than 6/60 to 3/60 was found in 10.8 % (16/148), less than 3/60 to 1/60 was found in 14.2% (21/148) and less than 1/60 in 14.9 % ( 22/148). All patients of myelinated nerve fibers, optic disc drusen and megalopapilla had a best corrected visual acuity 6/18 or better.

The visual acuity of <1/60 to PL was most commonly seen in optic nerve hypoplasia (40%), optic disc coloboma (22.8%) and tilted disc (13.3%) and in case of morning glory syndrome.

**Table-5a** shows the ocular associations with tilted disc. Most common ocular association with tilted disc was myopia which was found in 65.2% (30/46). Refractive power of eye ranges from -1.5D to -14D spherical and -0.50 D to -5.50 D cylindrical power. Anisometropia was found in 36.9% (17/46).

Exotropia was detected in 13% (6/46) cases and all these cases were unilateral. Nystagmus was detected in 6.5% (3/46) cases and associated with bilateral involvement of tilted disc. One of these 3 cases had toxoplasmic patch at macula which was an incidental finding. One patient had optic pit in association with tilted disc. Fig.1 (a-b) show tilted disc with associated ocular anomalies.

**Table-5b** shows Optic disc coloboma was associated with coloboma of other parts of eye. Choroidal coloboma was found in 90.9% of cases, iris coloboma in 40.9% and lens coloboma was found in 9.0%. Myopia was found in 77.2%. Refractive error was ranged from -1.5 to -11 dioptre sphere and -1.25 to -4 dioptre cylinder with mean spherical equivalent was -6D. Nystagmus was detected in 31.8% all were bilateral. Strabismus was detected in 2.2% (6/22). Exotropia was present in 4 cases in which 3 with unilateral and 1 with bilateral involvement of coloboma. Bilateral case showed alternating exotropia. Esotropia was found in 2 cases both were bilateral and associated with microphthalmos and microcornea with nystagmus. Microcornea with microphthalmos was present in 18.2% (4/22) cases with 3 were bilateral and associated with

lens coloboma and nystagmus. One patient had retinitis pigmentosa. Coloboma and associated ocular anomalies are shown in fig. 1(a-c).

Myelinated nerve fibers were found to be associated with hypermetropia which was found in 26.7% (4/15) eyes. Fig.3 shows extensive myelination of optic disc. One case of optic nerve hypoplasia was associated with aniridaia with lens coloboma. (fig.7)

Other congenital optic disc anomalies which were studied were not associated with significant other ocular abnormalities.

## Discussion:

The present study is an observational study, conducted with the purpose of determining the prevalence and ocular association with the congenital anomalies of optic disc and their effect on visual acuity. The study was conducted in Department of Ophthalmology, S.S. Medical College and Gandhi Memorial Hospital, Rewa, M.P., during the Period August 2012 to September 2014.

In our study overall prevalence of congenital anomalies of optic disc was 0.94% which is our hospital based observational study for the cases directly attending the eye OPD or referred cases from other departments and peripheral health centres. We also calculated the prevalence of each anomaly and found that prevalence of tilted disc was (0.46%), optic disc coloboma (0.22%), myelinated nerve fibers(0.11%), optic pit (0.05%), optic disc drusen (0.11%), megalopapilla(0.03%), optic nerve hypoplasia(0.03%), and morning glory syndrome (0.01%).

In this study there was no sex predominance. Sonja Cekic et al<sup>4</sup> examined 22 patients with various congenital anomalies of optic disc out of which 11 were males and 11 were females.

The prevalence of tilted optic disc was varied from study to study as diagnostic criteria were different in different studies. In our study majority of patients were diagnosed as tilted disc with prevalence of 0.46%.

You QS *et al*<sup>9</sup> found 0.36% prevalence of tilted optic disc. In this study, small discs with an oblique orientation without signs of pathology and without high myopia (defined as

>-8 D) were included. The study by Vongphanit *Jet al*<sup>8</sup> listed 'inferior or nasal tilting' as an inclusion criterion and found prevalence of tilted disc 1.6%. Alicia CS *et al*<sup>11</sup> found prevalence of tilted optic disc as 3.5%.

In our study we found optic disc coloboma in 22 cases with prevalence of 0.22%. Nakamura KM *et al*<sup>14</sup> found prevalence of disc coloboma as 0.05%.

Myelinated nerve fiber was detected in 0.11%. Majority of cases in our study were unilateral. Kodama *et al*<sup>15</sup> found 0.57% and Nangia V *et al*<sup>17</sup> found 0.58% cases of myelinated nerve fibers. You QS *et al*<sup>16</sup> found a 0.7% prevalence of myelinated retinal nerve.

The prevalence of optic disc pit in our study was 0.05%. In a study conducted by Wanq Y *et al*<sup>20</sup> the prevalence of optic disc pit was 0.02%. Prevalence of optic disc drusen is 0.03% in this study. You QS *et al*<sup>21</sup> found prevalence rate of optic disc drusen as 0.2%.

Megalopapilla, in our study was seen in 0.03% of cases. In our study we found 3 cases of optic nerve hypoplasia with prevalence of 0.03%. Mohny BG *et al*<sup>25</sup> found annual incidence of 2.4/100,000. Morning glory syndrome is a very rare anomaly. We found only one case of morning glory syndrome with prevalence 0.01%.

In this study bilateral involvement was found in all cases of optic disc drusen (100%), megalopapilla (100%), and majority cases of tilted disc (63%) and optic nerve hypoplasia (66.6%), whereas all cases of optic disc pit (100%), and majority patients of myelinated nerve fibers (63.6%) were unilateral. We found unilateral and bilateral optic disc coloboma with almost equal frequency and only one case of morning glory syndrome with unilateral involvement.

You QS *et al*<sup>9</sup> found bilateral tilted disc in 34.8% cases and Vongphanit *Jet et al*<sup>8</sup> in 37.5% cases. Lissa M<sup>10</sup> observed bilateral tilted disc in 58.3% cases. Unilateral optic disc coloboma was found in 67% of cases by Nakamura KM *et al*<sup>14</sup> and 76.5% cases by Gopal L *et al*<sup>12</sup>. In contrast Berk AT *et al*<sup>13</sup> documented bilateral involvement in 53.3%.

Kodama *et al*<sup>15</sup> found unilateral myelinated nerve fibers in 93.9% cases and You Q S *et al*<sup>16</sup> in 79.3% cases. Bilateral optic disc drusen was observed in 60% of cases by Thuttell M J *etal*<sup>22</sup>. Brown G.C.*et al*<sup>19</sup> noted unilateral optic pit in

86% cases and Gordon *Ret al*<sup>18</sup> in 91.6% cases. Bilateral optic nerve hypoplasia was seen in 88% by Acers T.*etal*<sup>28</sup> and in 84% of cases by Mohny BG *.et al*<sup>25</sup>. Kindler P.*et al*<sup>26</sup> reported ten cases of morning glory syndrome. All cases were unilateral.

The congenital abnormalities of the optic disc that can significantly impair visual function are optic nerve hypoplasia, optic disc coloboma.<sup>6</sup> Optic disc pit and tilted disc syndrome have variable loss of visual acuity depending on the serous macular detachment.<sup>6</sup> Optic disc drusen and myelinated nerve fibers rarely present with poor vision.<sup>7</sup>

In our study vision was severely affected in cases of optic disc coloboma, optic nerve hypoplasia and morning glory syndrome and patients with myelinated nerve fibers, optic pit, megalopapilla and optic disc drusen had good visual acuity.

Vongphanit J *et al*<sup>8</sup> noted the mean best-corrected visual acuity was 20/30 Snellen equivalent in eyes with tilted disc, or one log MAR line worse than eyes without tilted disc 20/25 Snellen equivalent. M. J. Thuttell *et al*<sup>25</sup> observed bilateral involvement in 60% cases of optic disc druses with normal visual acuity. Visual acuity better than 20/30 was reported in 86% cases of megalopapilla by Maisel JM *etal*<sup>23</sup>.

Gorden R.*et al*<sup>18</sup> found visual acuity better than 6/12 in 9/12 cases of optic pit. Giuseppe *et al*<sup>27</sup> reported 3 cases of morning glory syndrome with visual acuities of 2/200, 2/200 and perception of light only. In our study all cases of optic pit, optic disc drusen and megalopapilla had normal visual acuity.

Optic disc abnormalities may be associated with other congenital disorders of the eye.<sup>3</sup> Overall the most common ocular association with congenital anomalies of optic disc was refractive errors. We found optic disc pit and tilted disc together in the same eye of one patient.

In our study the most common ocular association with tilted disc was myopia and it was found in 77.2% cases. Other associated anomalies were strabismus (13%) and nystagmus (6.5%). One patient had toxoplasmic patch at macula which was an incidental finding. The most common cause of correctable visual impairment in tilted disc is refractive error. The amount of refractive error with tilted disk (compared to controls) varies from study

to study. Alicia CSet *al*<sup>11</sup> found significant (odds ratio: 2.77; p -0.001) association between myopia (88.5%) and tilted disc compared to normals. Vongphanit J *et al*<sup>8</sup> found myopia in 66.2% of eyes with tilted discs compared with 12.4% of eyes with a normal disc appearance. Strabismus was detected in 17%.

Optic disc coloboma in association with other ocular disorders was diagnosed in 45.4% of cases. The association of coloboma with other ocular disorders has been reported but there is little data on the prevalence of specific ocular disorders within the coloboma population.

In our study we found that most common ocular association with optic disc coloboma was myopia. Other associations were nystagmus (31.8%), strabismus (22.7%), microcornea and microphthalmos (18.2%). We also found that optic disc coloboma was associated with coloboma of other part of eye including iris, lens and choroid and one case was associated with retinitis pigmentosa. Nakamura KM *et al*<sup>14</sup> found 58% patients with other ocular disorders. In their study, 33% of patients were diagnosed with amblyopia and 30% were diagnosed with strabismus. Retinal detachment was a rare complication of coloboma patients in this study, affecting only 6% of study patients. Berk AT *et al*<sup>13</sup> found microphthalmia in 30%, microphthalmos in 20%, strabismus in 8% cases in their study on coloboma and observed that optic disc colobomas have a wide variety of presentations. Poor visual acuity was observed mostly in eyes with macular involvement and microphthalmia.

Myelinated nerve fibers were associated with hypermetropia in 18.2% of cases in our study. Nangia V *et al*<sup>17</sup> found significant association of myelinated nerve fibers with hypermetropia.

In our study in all 3 cases of optic disc drusen, the optic disc appeared as small, elevated disc with blurred margins. The diagnosis was confirmed with B- Scan ultrasonography.

You QSet *al*<sup>1</sup> found that optic disc drusen were significantly associated with small optic discs (p < 0.001). One case of optic disc drusen in our study was associated with retinitis pigmentosa.

Megalopapilla in our study was observed in 3 cases and all of them had large optic disc with large cup disc ratio. Neuroretinal rim was normal in all cases. One case was associated with raised intraocular tension of both the eyes.

Visual field analysis was carried out in this patient but field was not reliable. Sampaolesi *Ret al*<sup>24</sup> evaluated optic nerve heads of 405 eyes and divided them into 172 normal eyes, 168 eyes with glaucoma, 30 primary congenital glaucomas and 35 megalopapillas. They examined optic nerve heads with the HRT (Heidelberg Retina Tomograph) which revealed a large optic nerve head with an increased cup, but associated with a normal rim volume in cases of megalopapilla.

We found only 3 cases of optic nerve hypoplasia. One case was associated with lens coloboma and aniridia.

Only one case of morning glory syndrome was seen in our study and which was associated with esotropia. Kindler *et al*<sup>26</sup> found strabismus in 40% cases.



Fig. 1a Bilateral tilted optic disc



Fig. 1b Tilted disc with optic pit

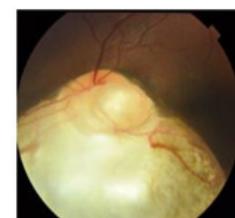


Fig. 2a Coloboma involving optic disc and choroid

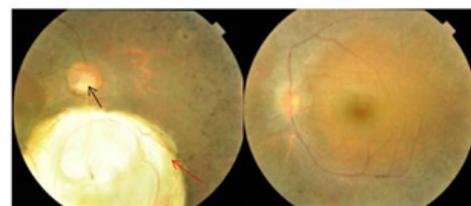


Fig. 2b Optic disc coloboma with choroidal coloboma with retinitis pigmentosa



Fig. 2c Iris coloboma, Cataract, Microcornea with lens coloboma



Fig. 3 Myelinated nerve fibers

Fig. 4 Optic pit

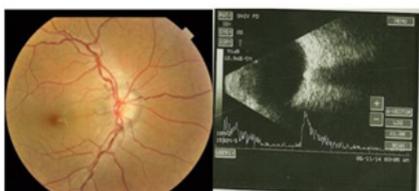


Fig. 5 Optic disc drusen with hyperchosity on B- Scan



Fig. 6 Megalopapilla

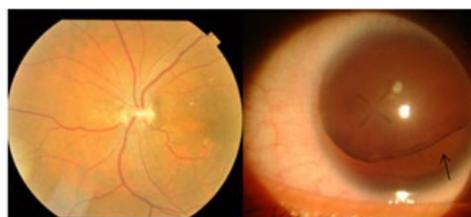


Fig. 7 Optic nerve hypoplasia with aniridia with lens coloboma

**Table-1 Distribution Of Anomalies**

S. No.	Optic Disc Anomaly	Number Of Patients	Percentage
1.	Tilted disc	46	48.9%
2.	Optic disc coloboma	22	23.4%
3.	Myelinated nerve fibers	11	11.7%
4.	Optic pit	5	5.3%
5.	Optic disc drusen	3	3.2%
6.	Megalopapilla	3	3.2%
7.	Optic nerve hypoplasia	3	3.2%
8.	Morning glory syndrome	1	1.06%
	<b>Total</b>	<b>94</b>	<b>100%</b>

Among the various optic disc anomalies studied, the highest prevalence was of tilted optic disc (0.46%).

**Table-2 Gender Distribution**

Sex	Tilted disc		Optic disc coloboma		Myelinated nerve fibers		Optic pit		Optic disc drusen		Megalopapilla		Optic nerve hypoplasia		Morning glory syndrome	
	N	%	n	%	n	%	n	%	n	%	n	%	n	%	n	%
Male	20	43.5%	10	45.5%	6	54.5%	3	60%	3	100%	1	33.3%	1	33.3%	0	0
Female	26	56.5%	12	54.5%	5	45.5%	2	40%	0	0%	2	66.7%	2	67.7%	1	100%
Total	46	100	22	100	11	100	5	100	3	100	3	100	3	100	1	100

Overall 46.80 % (44/94) cases were males and 53.2 % (50/94) cases were females.

**Table-3 Lateralitywise Distribution**

Laterality	Tilted disc		Optic disc coloboma		Myelinated nerve fibers		Optic pit		Optic disc drusen		Megalopapilla		Optic nerve hypoplasia		Morning glory syndrome	
	n	%	N	%	n	%	n	%	n	%	n	%	n	%	n	%
Unilateral	17	37%	9	41%	7	63.6%	5	100%	-	-	-	-	1	33.4%	1	100
Bilateral	29	63%	13	59%	4	36.4%	-	-	3	100%	3	100%	2	66.6%	-	-
Total	46	100%	22	100%	11	100%	5	100%	3	100%	3	100%	3	100%	1	100%

Table-3 shows laterality wise distribution of cases. Overall bilateral involvement was found in 57.4 % (54/94) cases.

**Table-4 Best Corrected Visual Acuity (BCVA) Of Affected Eye**

BCVA	Tilted disc		Optic disc coloboma		Myelinated nerve fibers		Optic pit		Optic disc drusen		Megalopapilla		Optic nerve hypoplasia		Morning glory syndrome	
	n	%	n	%	n	%	n	%	n	%	n	%	n	%	n	%
NO PL	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
<1/60-PL	10	13.3%	8	22.8%	-	-	1	20%	-	-	-	-	4	80%	1	100%
<3/60-1/60	15	20%	6	17.1%	-	-	-	-	-	-	-	-	-	-	-	-
<6/60-3/60	11	14.7%	5	14.3%	-	-	-	-	-	-	-	-	-	-	-	-
<6/18-6/60	29	38.7%	12	34.3%	-	-	-	-	-	-	-	-	1	20%	-	-
6/18 Or Better	10	13.3%	4	11.4%	15	100%	4	80%	6	100%	6	100%	-	-	-	-
<b>Total</b>	<b>75</b>	<b>100%</b>	<b>35</b>	<b>100%</b>	<b>15</b>	<b>100%</b>	<b>5</b>	<b>100%</b>	<b>6</b>	<b>100%</b>	<b>6</b>	<b>100%</b>	<b>5</b>	<b>100%</b>	<b>1</b>	<b>100%</b>

**Table -5 (A) Ocular Association With Tilted Disc**

S.No.	Associated ocular anomalies	Number of patients	Percentage
1.	Myopia	30	65.2%
2.	Anisometropia	17	36.9%
3.	Exotropia	6	13%
4.	Nystagmus	3	6.5%
5.	Optic pit	1	2.3%
6.	Toxoplasmic patch	1	2.3%

**Table-5 (B) Ocular Association With Optic Disc Coloboma**

S. No.	Ocular association	Number of cases	Percentage
1	Choroidal Coloboma	20	90.9%
2	Myopia	17	77.2%
3	Iris coloboma	9	40.9%
4	Anisometropia	9	40.9%
5	Nystagmus	7	31.8%
6	Strabismus	5	22.7%
7	Microcornea with microphthalmos	4	18.2%
8	Cataract	2	9.0%
9	Lens coloboma	2	9.0%
10	Retinitis pigmentosa	1	4.5%

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