The Prevalance of Congenital Optic Disc Anomalies in Turkey: A Hospital-Based Study

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Research Article

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Abstract

Purpose: The aim of this study was to investigate the prevalance of congenital optic disc (OD) anomalies in Turkey.

Methods: The 11149 eyes of 5583 patients were screened for OD anomalies. All patients were underwent a complete ophthalmic examination including best corrected visual acuity, refraction, spherical equivalent, slit lamp biomicroscopy, intraocular pressure measurements, dilated stereoscopic fundus examination. Data analyses were performed by using SPSS for Windows, version 22.0 (SPSS Inc., Chicago, IL, United States).

Results: 11149 eyes of 5583 participants were screened. Of the 5583 participants who underwent OD examination, 186 (3,3%) were found to be abnormal. 266 of 11149 (2,38%) eyes were found OD anomalies. 98 (52,7%) were female, 88 (47,3%) were male and the mean of age was 44,05±15,73 years. The prevalence of all congenital OD anomalies was found 3,3%. The tilted disc was the most common anomaly and was found at least one eye in 46 patients (75 eyes) and 0.82% of all screened patients. Peripapillary myelinated nerve fibres was the second common anomaly and was found at least one eye in 29 subjects (35 eyes) and 0,51% of all screened subjects. Peripapillary atrophy was the third common anormality, and was found in at least one eye in 24 patients (37 eyes) and 0,42% of all screened subjects.

Conclusion: To our knowledge, this is the first study that the prevalences of all congenital optic disc anomalies from Turkey. The prevalence of congenital optic disc anomalies is higher than in other countries.

Introduction

The optic nerve (ON) is one of the most important cranial nerves and the axons of the ON constitute 38% of all the axons entering and leaving the central nervous system. It courses from the glob to the brain and it is divided into four segments: intraocular, intraorbital, intracanalicular, and intracranial [1]. The optic nerve head (ONH) is the visible portion of the ON during ophthalmic examination, also known as the optic disc (OD). A congenital OD anomaly may be an isolated ocular abnormality such as peripapillary myelinated nerve fibres (PMNF) and optic disc drusen (ODD) or occur as part of a systemic disorders like central nervous system development pathologies [2]. The visual acuity of impacted eye might minimally or severely be affected, depending on the extent of the lesion and poor vision may be the reason for patients to apply to the outpatient clinics [3]. Thus, making the correct diagnosis of congenital OD anomalies during ophthalmic examination is very important for both patients and ophthalmologists. Moreover, accurate diagnosis of congenital OD anomalies would help the patients with innocuous OD anomalies avoid unnecessary advanced diagnostics [4].

Thanks to the studies showing the epidemiological characteristics of a population, the prevalence of common diseases or anomalies can be determined. Prevalence is the number of cases of a disease/anomaly present in a particular population at a given time. While making differential diagnosis, by giving priority to the diseases/anomalies with a high prevalence in the population, we can easily get the correct diagnosis without wasting time, money and energy.

The aim of this study was to investigate the prevalance of congenital OD anomalies in Turkey. According to our knowledge this is the first study that determines the prevalance of congenital OD anomalies fromTurkey.

Methods

This study was a cross-sectional hospital-based study. Between January 2019- March 2020, the ophthalmic examination records of the patients with various ophthalmic problems were collected retrospectively at the Ophthalmolgy Department of Health Science University Ankara Training and Researching Hospital which is a tertiary care center in Turkey. The consecutive patients who were aged between 18-65 years were enrolled. This study was performed in line with the principles of the Declaration of Helsinki. Approval was granted by the Ethics Committee of Ankara Training and Researching Hospital (Date: 26.11.2020/ No: 490). Informed consent was obtained from all individual participants included in the study. The 11123 eyes of 5570 patients were screened for OD anomalies All optic disc anomalies were assessed by same two ophthalmologist (GO and OC) who are a neuro-ophthalmologist and a retina specialist who were blinded to the findings of the other. All patients were underwent a complete ophthalmic examination including best corrected visual acuity (BCVA) with LogMAR chart, refraction test, spherical equivalent refraction (SE), slit lamp biomicroscopy, intraocular pressure (IOP) measurements with Goldmann aplanation tonometer, dilated stereoscopic fundus examination. Patients with OD anomalies were examinated with spectral-domain optic coherens tomography (SD-OCT) (Heidelberg Engineering, Inc., Heidelberg, Germany), autofluorescence and B-scan ultrasonography imaging to improved the sharpness of the diagnoses. The images of all patients with OD anomalies were taken by using Zeiss FF 450 IR fundus camera.

Diagnostic criteria for congenital OD anomalies was given below. PMNF was defined as feather-like whiteish noninflammatory abnormalities in the superficial layer of the retina. The optic disc hypoplasia (ODH) was defined as the greyish appearance or pale in color and the optic nerve head was abnormally small. The morning glory disc anomaly was defined as a funnel-shaped excavation of the posterior globe that surrounds and incorporated the optic disc, which was located in the center. Optic disc colobomas (ODC) was defined as an enlarged optic disc contains a sharply delineated, white, bowl-shaped excavation optic disc pits (ODP) was defined as round or oval, gray, white, or yellowish depressions in the optic disc that was usually located temporally but can occur anywhere on the optic disc The tilted disc was defined as small ONH with an oblique orientation and oval disc shape without signs of pathology that the superotemporal optic disc was elevated and the inferonasal disc was posteriorly displaced. Also inferior or nasal disc tilting were excepted the diagnostic criterion for a tilted disc appearance.

Prepapillary vascular loops (PVL) was defined as retinal arterial anomalies that project from the optic disc cavity into the vitreous then return to the optic disc. Optic disc drusen (ODD) was defined as the deposition of hyalinelike, calcified material within the substance of the nerve. OD pigmentation was defined as the melanin deposition on the optic disc. Bergmeister papilla was defined as a small stalk of fibrous tissue arising from the optic disc. Epipapillary glial tissue was defined as greyish white fibrous tissue on the optic disc surface. Peripapillary atrophy (PPA) was defined as chorioretinal thinning and disruption of the retinal pigment epithelium (RPE) in the area surrounding the optic disc. Staphyloma of ON was defined as protrusion of the posterior shell of the eye globe. The morning glory disc anomaly was defined as a funnel-shaped excavation of the posterior globe that surrounds and incorporated the optic disc, which was located in the center.

Patients who had a history of glaucoma, ocular surgery, any disease affecting the anterior segment, retina, posterior segment or optic nerve other than congenital optic disc anomalies were excluded. Only patients with refractive errors were included in the study.

Data analyses were performed by using SPSS for Windows, version 22.0 (SPSS Inc., Chicago, IL, United States). Continuous data were described as mean ± SD.

Categorical data were described as number of cases (%).

Results

11123 eyes of 5570 participants were screened. 52% (n:2894) of participants were female and 48% (n:2676) were male. The mean age was 43.70 ± 15.5 years. The general characteristics of all subjects are shown in Table 1.

Table 1
Demographic findings of all screened participants

	Total
Number of all participants	5570
Number of eyes	11123
Mean age, years	43.70 ± 15.50
Male, number, %	2676, 48%
Female, number, %	2894, 52%

Of the 5570 participants who underwent OD examination, 174 (3.12%, 95% CIs 2.66–3.58%) were found to be abnormal. 246 of 11123 (2.21%, 95% CIs 1.93–2.48%) eyes had OD anomalies. 92 (52.9%) were female, 82 (47.1%) were male and the mean of age was 44.25±15.67 years. 71 (41%) patients were bilateral, 103 (59%) patients were unilateral involvement.

The prevalence of all congenital OD anomalies was found 3.12%. The general characteristics of patients who detected OD anomalies are shown in Table 2 and clinical signs are shown in Table 3.

Table 2
Demographic findings of patients with optic disc anomalies

	Patients with OD anomalies
Age, years	44.25±15.67
Male, number, %	82, 47.1%
Female, number, %	92, 52.9%
Unilateral, number, %	103 (59%)
Bilateral, number, %	71 (41%)
Number of OD Anomalies (%, 95% CI)	174 (3.12%, 2.66–3.58%)
Number of eyes (%, 95% CI)	246 of 11123(2.21%, 1.93–2.48%)

Table 3
Clinical signs of patients with optic disc anomalies

	N (%, %95 CI) of patiens	N (%, %95 CI) of eyes	Unilateral/Bilateral	BCVA, LoGMAR	SE, D
Tilted disk	46 (0.83, 0.59 to 1.06)	75 (0.67, 0.52 to 0.83)	17/29	0.13±0.23	-3.85±3.13
PMNF	31 (0.56, 0.36 to 0.75)	35 (0.31, 0.21 to 0.42)	27/4	0.03±0.1	0.17±0.74
PPA	24 (0.43, 0.26 to 0.60)	37 (0.33, 0.23 to 0.44)	11/13	0.22±0.36	-3.76±5.70
OD Pigmentation	16 (0.29, 0.15 to 0.43)	18 (0.16, 0.09 to 0.24)	14/2	0.01±0.02	-0.32±1.18
ODD	15 (0.27, 0.13 to 0.41)	28 (0.25, 0.16 to 0.34)	2/13	0.08±0.26	-0.20±1.77
Glial	12 (0.22, 0.09 to 0.34)	14 (0.13, 0.06 to 0.19)	10/2	0.05±0.19	-0.31±0.84
Bergmeister papilla	11 (0.20, 0.08 to 0.31)	14 (0.13, 0.06 to 0.19)	8/3	0.00±0.00	-0.10±1.17
PVL	6 (0.11, 0.02 to 0.19)	6 (0.05, 0.01 to 0.10)	6/0	0.00±0.00	-0.38±0.63
ODH	5 (0.09, 0.01 to 0.17)	7 (0.06, 0.02 to 0.11)	3/2	0.32±0.47	1.30±5.56
OD Staphyloma	4 (0.07, 0.00 to 0.14)	5 (0.04, 0.01 to 0.08)	3/1	0.8±0.71	-15.20±10.35
Megalopapilla	1 (0.02, 0 to 0.05)	2 (0.02 ,0 to 0.04)	0/1	0.00±0.00	-10.00±0.00
ODP	1 (0.02, 0 to 0.05)	1 (0.01 ,0 to 0.03)	1/0	0.52±0.00	0.5±0.00
Morning glory	1 (0.02, 0 to 0.05)	2 (0.02 ,0 to 0.04)	0/1	0.00±0.00	0.00±0.00
ODC	1 (0.02, 0 to 0.05)	2 (0.02 ,0 to 0.04)	0/1	1.00±0.00	-12.00±0.00
TOTAL	174 (3.12, 2.66 to 3.58)	246 (2.21, 1.93 to 2.38)	102/72		

BCVA; best corrected visual acuity

SE; spherical equivalent

D; dioptries

PMNF; peripapillary myelinated nerve fibres

	N (%, %95 CI) of patiens	N (%, %95 CI) of eyes	Unilateral/Bilateral	BCVA, LoGMAR	SE, D
PPA; Peripapillary atrophy					
OD; Optic disc					
ODD; optic disc drusen					
ODH; optic disc hypoplasia					
ODC; optic disc colobomas					
PVL; Prepapillary vascular loops					
ODP: Optic disc pit					

The tilted disc (Figure 1) was the most common anomaly and was found at least one eye in 46 patients (75 eyes) and 0.83% of all screened patients. Seventeen patients had unilateral, 29 patients had bilateral involvement. The mean SE power was -3.85 ± -3.13D and the mean BCVA was 0.13 ± 0.23 LogMAR in patients with tilted disc anomalies. Inferionazal tilting (54.3%) was the most common direction; other types of tilting direction were inferior (34.2%), inferiotemporal (8.6%), nasal (2.9%) respectively.

PMNF (Figure 2) was the second common anomaly and was found at least one eye in 31 subjects (35 eyes) and 0.56% of all screened subjects. In patients with PMNF, the mean SE power was 0.17 ± 0.74 D and the mean of BCVA was 0.03 ± 0.1 LogMAR. Twenty-seven patients had unilateral, 4 patients had bilteral involvement. The most often location of PMNF was located at the superiotemporal region of ON (31.25%); The other frequent locations of PMNF were superior (26.1%), inferior (18.75%), and temporal region of ON (18.75%), (12.5%). All of PMNF anomalies were continous with ONH.

PPA (Figure 3) was the third common anomaly, and was found in at least one eye in 24 patients (37 eyes) and 0.43% of all screened subjects. The mean SE power was -3.76 ± 5.7 D and the mean of BCVA was 0.22 ± 0.36 LogMAR in patients with PPA. 11 patients had unilateral and 13 patients had bilateral involvement. Seven patients (11 eyes) with PPA had no myopic refractive error.

Other anomalities were ODD (Figure 4), epipapillary glial tissue (Figure 5), pigmentation, PVL (Figure 6), Bergmeister papilla (Figure 7), ODH (Figure 8), staphyloma of ON, ODP, ODC, megalopapilla (Figure 9) and morning glory disc anomaly.

Discussion

Congenital anomalies of the OD are rare but they are crucial to diagnose accurately, as they can cause visual impairment or pseudopapilledema apperence. Accurate and precise examination of optic nerve head morphology is important to make correct diagnosis and functional assessment. Substantial interindividual and intraindividual morphological variations and factors such as race and age determine the normal range. Estimating the pervalence of these variations and anomalies is essential because it is

representative of the characteristics of the population. After determining which diseases/anomalies are common in a population, by giving priority to the diseases/anomalies with a high prevalence in the population the accurate diagnosis can be easily reached without wasting time, money and energy.

There were a few studies described the prevalences of various OD anomalies [5–10]. To our knowledge this is the first study estimating the prevalence of all congenital OD anomalies from Turkey. In this study, the prevalence of OD anomaly was 3.12%. Bassi et al found it to be 1.1% in the adult South Indian population [5]. The reasons for the difference in prevalence between our study and other studies may be due to the racial differences or age and gender distribution differences between examined population groups. In addition, our study also was a hospital-based study, namely, all patients who presenting with various ophthalmic problems were screened in this study. Bassi et al reported 63.7% were males and 36.3% were females and the mean age at presentation was 51.4±8.9 years [5]. Contrary to Bassi et al, in this study, the gender ratio was almost equal and the mean age of cases was younger. These differences may be caused by the fact that our study was a hospital-based study and there was no difference between the number of male and female patients applying to hospital or the genetic characteristics of our patients because of geographical differences. If our study was population-based rather than hospital-based, the prevalence of ON anomalies could have been lower. In following paragraphs, we separately discussed each congenital OD anomaly that we observed in Turkey.

The prevalence of tilted disc in Turkey population was found 0.83% in our study. As is known the tilted disc and PPA are the characteristic shape of myopic disc [6]. Similar to this information, in our study, SE was compatible with myopic shift in tilted disc and PPA cases. The prevalence of tilted disc was found 1.6% in Australia and 0.49% in Northern China [7, 8]. Both study used inferior or nasal disc tilting as the diagnostic criterion for a tilted disc appearance as in our study. In another study was reported that the prevalence of tilted optic discs was 3.5% in adult Chinese population in Singapour but when the definition of tilted optic discs included tort, the prevalence decreased to 2.4% [6]. They explained that this variation is resultant of diversity in definitions of tilted and torted optic discs, different inclusion criteria, and varying research methods between the studies. Also, the difference of race/ethnicity between the study groups could be explanation of these variety.

PMNF is caused by oligodendrocytes abnormally migrated into the retina, and is a relatively rare finding in a general ophthalmological clinic [9]. You et al. reported PMNF prevalence in adults 0.4% in China, Kodama et al. reported 0.57% in Japan, Bassi et al. reported 0.28% in South India [5, 9, 10]. The prevalance of PMNF was found 0.56% in our study, similar to other studies.

PPA is a non-specific finding. It can occur in both benign and pathologic conditions (glaucoma, myopia) and can also be a result of aging. It is not well understood that how to the peripapillary region is affected in these diseases [11]. Liu at al. reported that the area of PPA was associated with SE power and as SE power increased, the PPA area increased linearly [12]. Similar to literature, two-third of patients with PPA had high myopic SE power in our study.

The true congenital optic disc pigmentation is typically described as discrete, irregular and granular in appearance. Prevalence of OD pigmentation was found 0.03% in a previous study [5]. Contrarily previous study we found higher rate in our study (0.29%).

ODD is acellular deposits located in the optic nerve head which progressively become more calcified and allow them to be visible on ophthalmoscopy. The prevalence of ODD in children is about 0.4% [13]. In adults, the prevalence of ODD was found 2% in previous studies [14, 15]. In our study, ODD was seen less common in Turkish population than literatüre (0.27%).

Epipapillary glial tissue is a source for intrapapillary proliferations, as the glial tissue surrounds the area of cavernous atrophy of the optic nerve and is defined with other optic disc anomalies likewise ODP [16]. Bassi et al. found the prevalence of epipapillary glial tissue 0.28% [5]. The prevalance of epipapillary glial tissue in our study was compatible with the literatüre (0.22%).

A persistent hyaloid artery results from persistant hyaloid vascular system; a small stalk of fibrous tissue on the optic disc is also known as Bergmeister papilla. Its prevalence was reported 0.03% in South India in adults [5]. Unlike to this study, we determined the prevalence of Bergmeister papilla higher (0.2%).

PVL originates from a branch of either retinal artery or vein ending at the disc or surrounding optic disc [17]. The prevalence of PVL was 0.115% in Japan, and 0.15% in Africa [18, 19]. Our result was conformable to previous studies.

ODH is a morphologically and functionally abnormal small optic disc which may appear ophthalmoscopically gray or pale in color and is often surrounded by a peripapillary halo [20]. In the adult Indian population and is seen only in 0.04% [5]. The prevalence of ONH in England was reported that 10.9 per 100,000 [21]. Our study shows similar results with Asian population (0.09%).

The optic disc changes like staphyloma of OD is a common characteristic of highly myopic eyes and may be caused by scleral stretching in the peripapillary region [22]. In our study, staphyloma of OD was associated with high myopic eyes (0.02%)

ODP are focal depressions located at the bottom of the ONH that can be associated with a non-rhegmatogenous serous macular detachment [20]. Its prevalence was found 0.02% in China, and 0.01% in South India similar to our results [5, 23].

Optic disc coloboma (ODC) is a congenital malformation of the optic disc which is caused by a defective closure of the embryonic fissure that can cause severe visual impairment or blindness [20]. Skriapa Manta et all reported prevalence of ODC 0.0089% in chilhood population in Sweden, Bassi et al. reported 0.08% in adult population [5, 24]. During our study, we found the prevalence of ODC like previous studies.

Morning glory disc anomaly has a characteristic appearance and may result from abnormal development of the lamina cribrosa and posterior sclera [25]. Ceynowa et al. reported the prevalance of Morning glory disc anomaly was 0.26% [26]. Our results supportted to literature (0.02%)

Conclusion

To our knowledge, this is the first study that estimates the prevalences of all congenital optic disc anomalies from Turkey. The major limitation of this study is that it was not population-based and so our study represents only the patients going to the hospital. However, in ophthalmology practice, we think it can be valuable in terms of showing the rate of optic disc abnormalities that ophthalmologists may encounter. We found that the prevalence of congenital optic disc anomalies is higher in Turkey compared to other countries, We think that early recognition of congenital optic disc anomalies is important in the differential diagnosis of acquired diseases that can cause vision loss and thus, the use of unnecessary additional diagnostic methods will be prevented.

Declarations

Ethics

The study was approved by Ethics Committee of Ankara Training and Researching Hospital and adhered to the tenets of the Helsinki declaration. All the patients were explained the procedure in detail and an informed consent was taken from them.

Conflict of Interest Statement

None of the following authors have any proprietary interests or conflicts of interest related to this submission: None of the authors

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Author Contribution

G.O. conceived the idea, designed this study, collected data, , interpreted data, and drafted this paper. O.A.C. collected data, performed the statistical analysis. G.S. collected data and supervised this study. All of the authors read and approved the final version of this work.

Statements and Declarations

There is no conflict to disclosure.

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Author Contributions

All authors contributed to the study conception and design. Material preparation, data collection and analysis were performed by [Gozde Orman], [Ozlem Aydinoglu-Candan] and [Gulten Sungur]. The first draft of the manuscript was written by [Gozde Orman] and all authors commented on previous versions of the manuscript. All authors read and approved the final manuscript.

Ethics approval

This study was performed in line with the principles of the Declaration of Helsinki. Approval was granted by the Ethics Committee of Ankara Training and Researching Hospital (Date: 26.11.2020/ No: 490). *Informed consent was obtained from all individual participants included in the study.*

Consent to publish

The authors affirm that human research participants provided informed consent for publication of the images in all Figures.

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Figures

Figure 1

Tilted disc. (a); superotemporal region of optic disc is elevate and peripapillary atrophy is seen at inferonasal region in right eye. (b); superior optic disc is elevated and peripapillary atrophy is seen inferior at region in right eye of an another patient

Figure 2

Myelinated nerve fiber (MNF). Peripapillary MNF is surrounding of the optic disc

Figure 3

Peripapillary atrophy (PPA)

Figure 4

Optic disc drusen (ODD). (a); Fundus photograph of a ODD in right eye. (b); Fundus autoflourance of ODD in same patient. (c); Swept optic coherence tomography of ODD. Red arrow is shown lower reflectivity with hyperreflective margin of ODD

Figure 5

Glial tissue. Fundus photographs of optic disc glial tissue in two different patient's is shown with red arrows

Figure 6

Vascular loop. Fundus photograph of vascular loop is shown with red arrow

Figure 7

Bergmeister papilla. Red arrow shown persistan fetal hyaloid artery which arises from the centre of the optic disc, consists of a small tuft of fibrous tissue

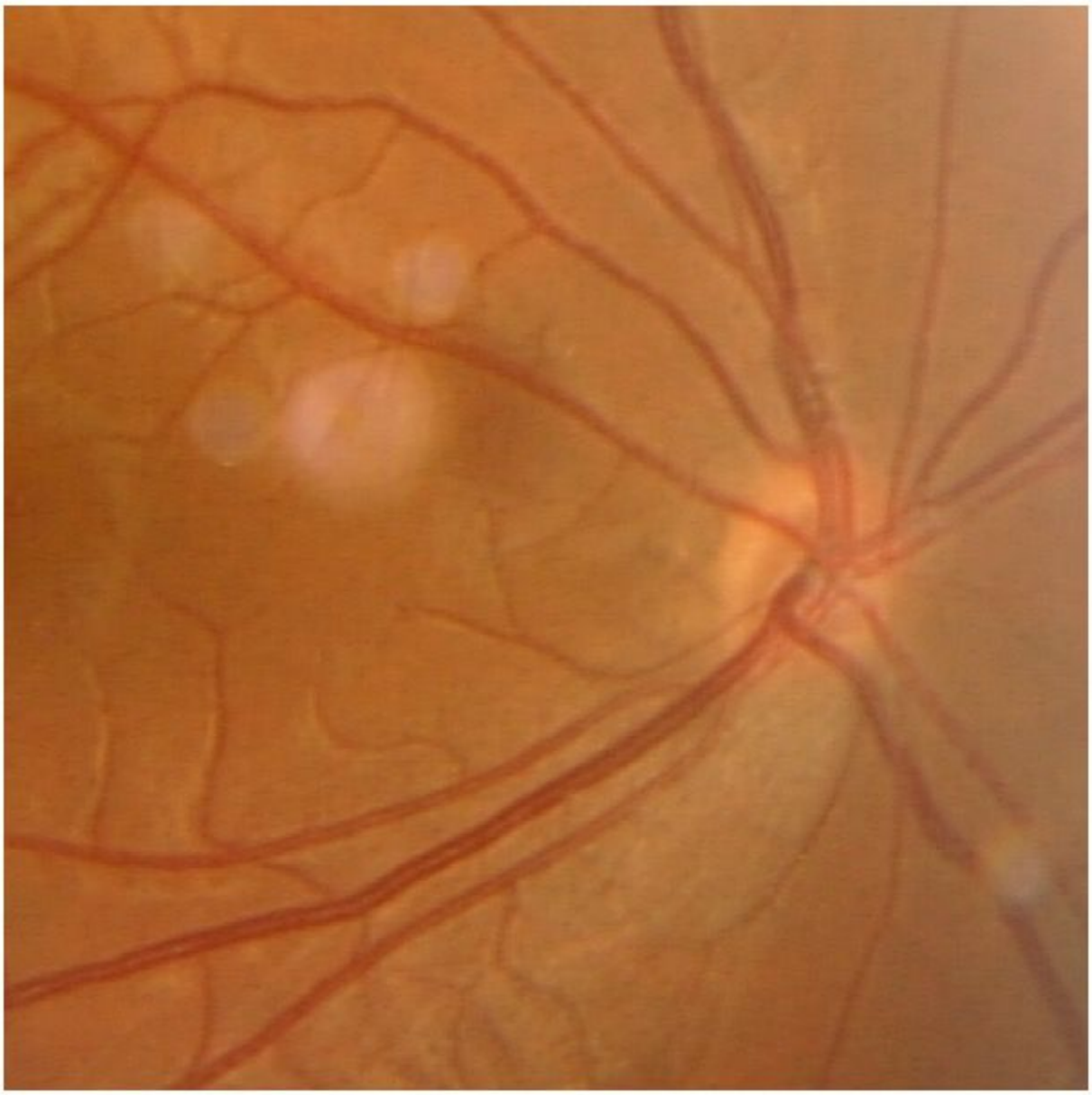

Figure 8

Optic disc hypoplasia (ODH). Fundus photograph of a patient with ODH which is seen small optic disc surrounding with peripapillary halo

Figure 9

Megalopapilla. Fundus photograph of a patient with megalopapilla which is seen a large optic disc