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Case Report

Morning Glory Anomaly in A Fifteen-Year-Old Saudi Girl

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Abstract

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Background: The Morning Glory anomaly is an extremely unusual birth defect affecting the optic disc. It may exist in isolation or in conjunction with other ocular or non-ocular malformations. Its distinctive appearance is characterized by a large disc containing radial arteries and a remnant of white glial tissue, which is situated within a funnel-shaped excavation of the posterior fundus.

Case Report: For the past five years, a 15-year-old Saudi girl has complained of gradual impaired vision in her right eye. Upon examination, the right eye exhibited a best corrected visual acuity of hand movement and a normal anterior segment. Furthermore, a substantial optic disc was excavated in the center, and the vessels exhibited an aberrant emergence, reminiscent of the Morning Glory anomaly. The left eye exhibited no abnormalities.

Conclusion: This optic disc defect can only be treated by first ruling out any associated general and/or ocular abnormalities. Regular follow up is necessary due to the risk of serous retinal detachment. It is crucial to check for deep amblyopia early on and then treat the condition appropriately if it is detected.

Keywords: Morning Glory Syndrome; Optic Disc Anomaly; Amblyopia; Decreased Visual Acuity.



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INTRODUCTION

Congenital optic nerve abnormality morning glory anomaly [MGA] occurs in extremely rare cases. This condition is more common among white people and among females. Unilateral manifestation is more common. The optic disc is encased and included in an excavation of the posterior globe. It causes impaired vision, and has also been associated with a number of ocular and non-ocular abnormalities [1].

The occurrence of MGA is unknown; nevertheless, a 2015 trail reports a prevalence of 2.6/100,000 in Stockholm, Sweden. By the age of two, the condition is usually diagnosed. Around the age of five years, there are instances in which late presentations are observed [2]. Most of the cases are discovered in the early childhood.

In this article, we report a case of a sporadic morning glory anomaly with otherwise normal ophthalmic examination in a 15 years old Saudi female patient referred from a paediatrician to the ophthalmology department in New Najran General Hospital. The late presentation of morning glory anomaly is unusual, and there are not enough studies reporting it.

Case Report:

A fifteen years old female patient with no pat medical or ocular history, who presented with a five-year-history of gradual deterioration of vision in the right eye.

On examination, she had no facial abnormalities and the neurologic examination was normal. No familial history of ocular illness or congenital abnormalities was present. Her unaided visual acuity was limited to hand movement in the amblyopic right eye, while the left eye exhibited a visual acuity of 1.0, does not improve by glasses. There was no nystagmus, no strabismus, normal pupillary examination, and normal ocular motility. Anterior segment examination and intraocular pressure were normal in both eyes.

Fundoscopy of the right eye showed a large excavated disc with a central whitish area of glial tissue [Figure 1].

The blood vessels fanned out from the disc in a radial fashion. No abnormalities were detected during the examination of the left eye. Using B-scan ultrasound, we know that the right eye has an axial length of 24.7 millimetres and the left eye of 23.3 millimetres. There were floating echogenic strands in the right vitreous humour, but no retinal detachment. The left eye was sonographically normal.

The right anomalous optic nerve head was identified by optical coherence tomography [OCT] of the optic nerve head. This head is characterized by an expanded disc area [7.04 mm²] in addition to cup [cup/disc ratio 0.92], as well as a denser retinal nerve fibre layer [RNFL], particularly in the superior region. The left eye had normal RNFL thickness, absence of disc oedema, and a typical appearance of the optic nerve head [Figure 2]. OCT macula was normal in both eyes [Figure 3]

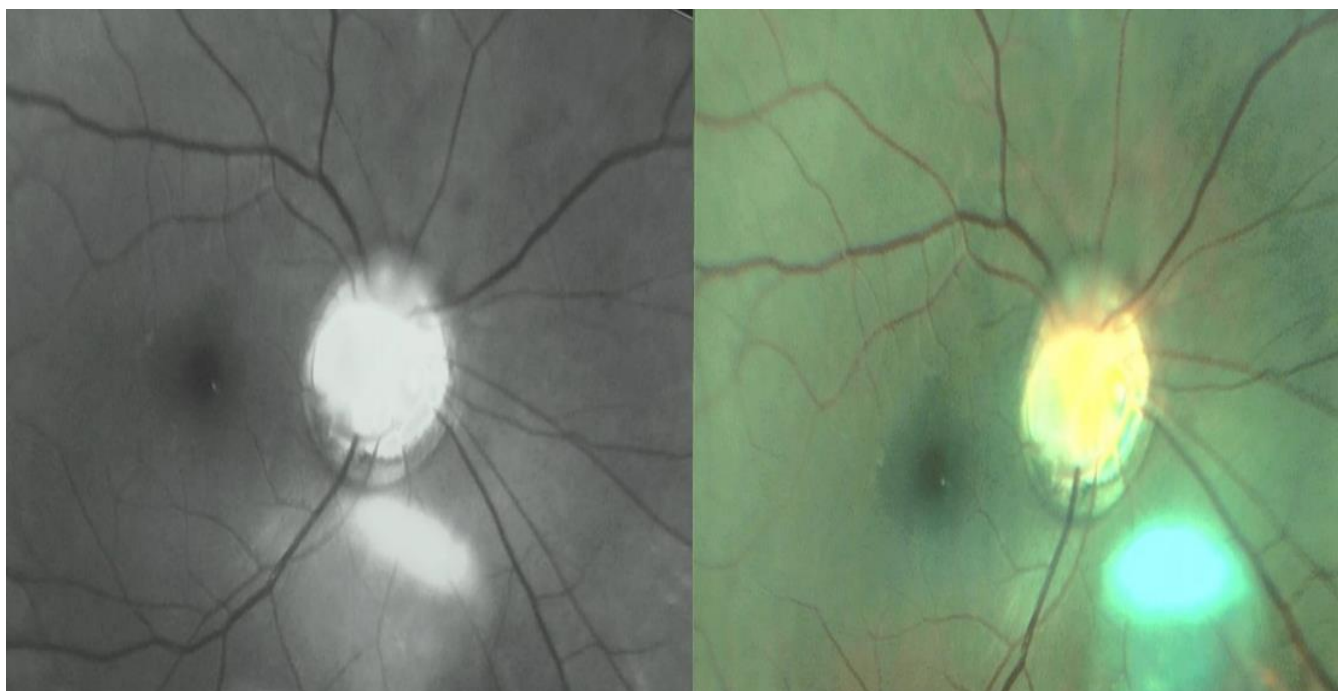


Figure [1]: Color fundus photo and Red free image of right MGA

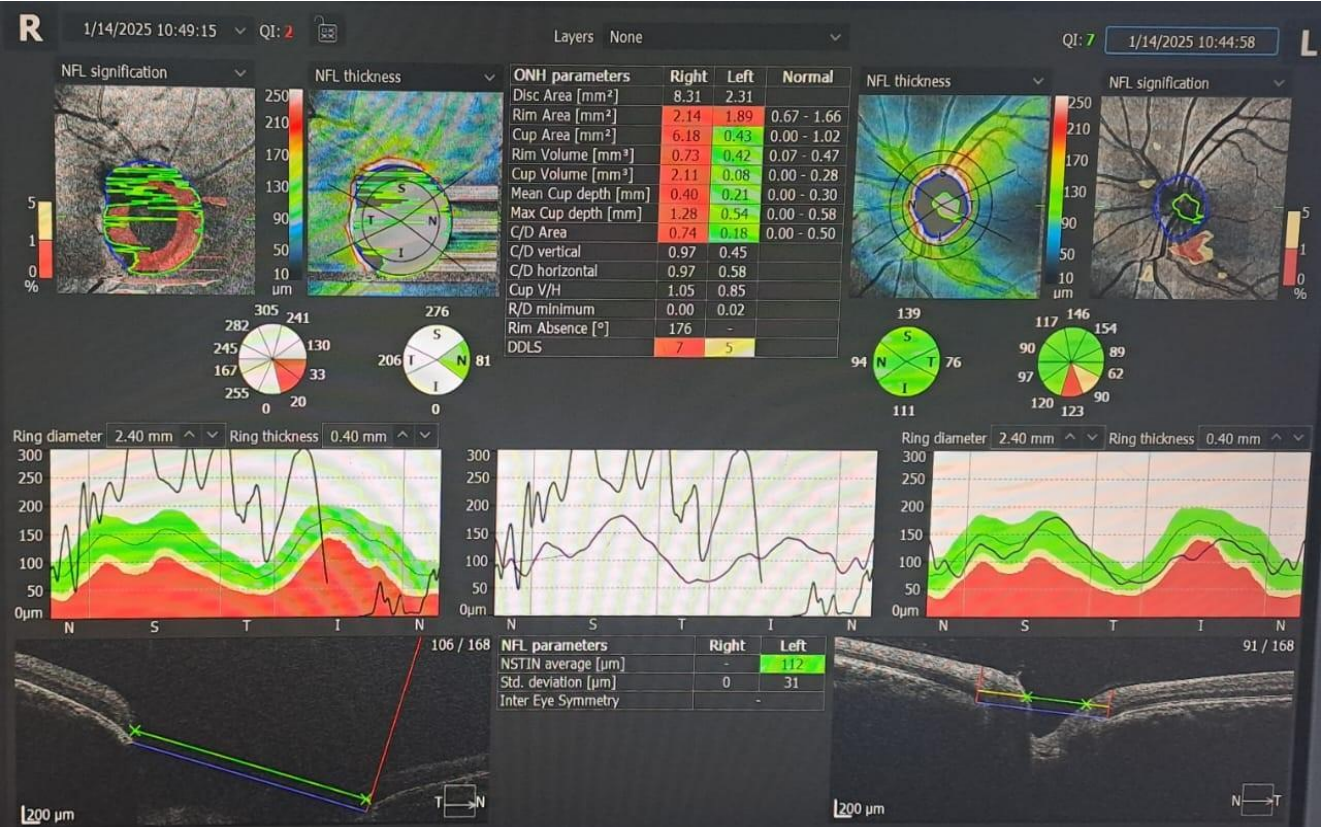


Figure [2]: OCT image of both optic nerve heads

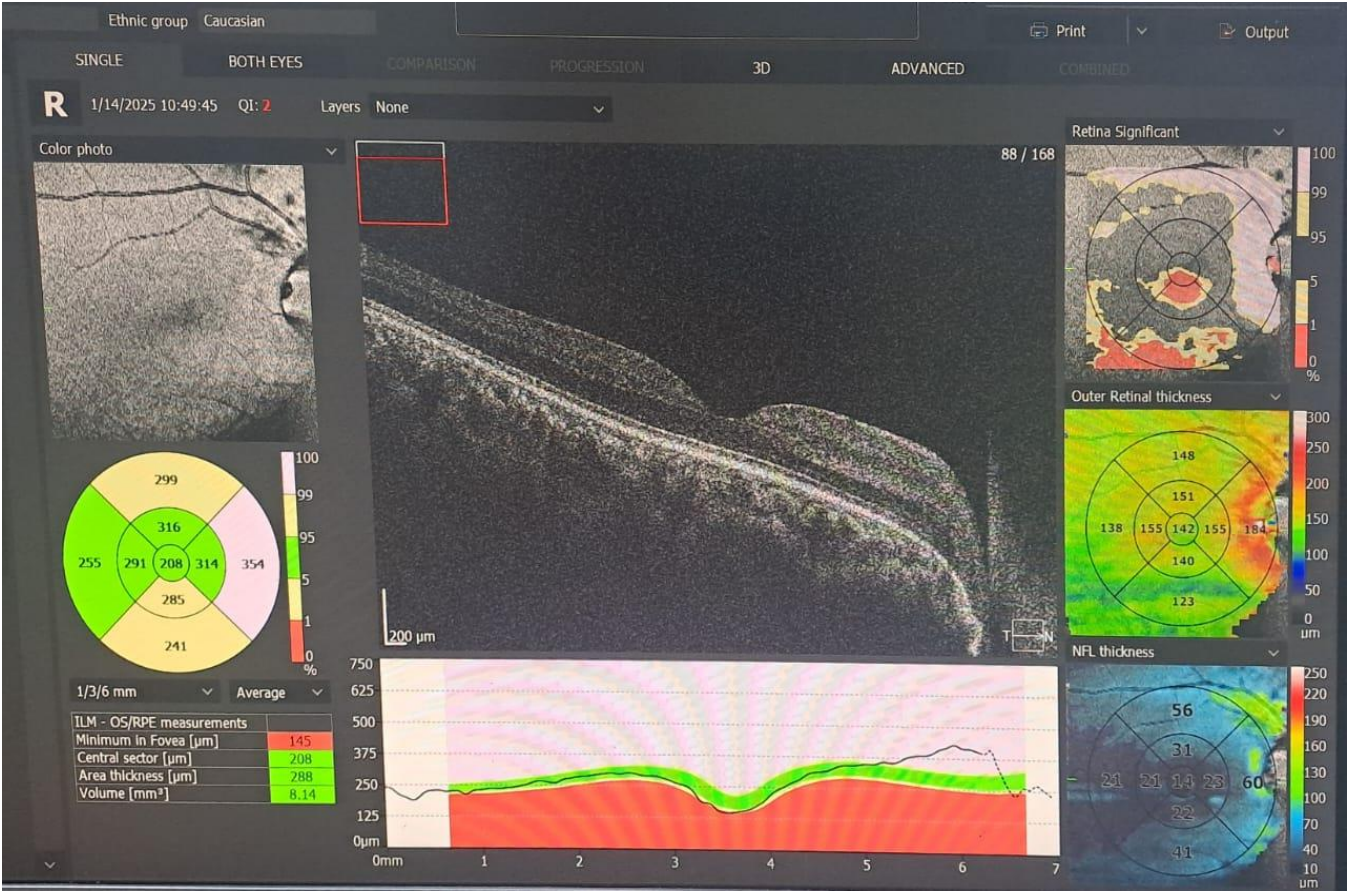


Figure [3]: OCT image of the right macula

DISCUSSION

An otherwise normal ocular examination was conducted on a 15-year-old Saudi female patient who presented with a case of occasional morning glory abnormality. The late presentation of morning glory anomaly is unusual, and there are not enough studies reporting it.

Reis made the initial description, which was subsequently assumed by Handmann, resulting in the term "Handmann anomaly" as it was known until 1970 [3]. Peter Kindler was the first to call it after the English term for Morning Glory, due to its similarity to the morning glory flower. It is an uncommon sporadic condition. He recorded ten cases of congenital disc abnormality, which are distinguished by a funnel-shaped optic nerve head. Chorio-retinal pigmentary alteration ringed a ring-shaped area that contained a nucleus of white fibrous tissue. Near the optic disc's periphery, the retinal arteries first appeared as a network of straight, narrow branches. These branches initially deviated from the typical central branching pattern and were subsequently rectified. Distinguishing arteriolar circulation from venous circulation is typically challenging. There may also be minor peripapillary arteriovenous connections [4]. Disc size is directly proportional to the dug posterior scleral aperture. At times, the disc itself could seem to be at a higher level. The macula may be included in the excavation, a process called "macular capture," depending on the extent of papillary chorio-retinal involvement [5]. MGA is non-hereditary and genetic defects associated with it are not yet established [6]. Altun *et al.* [7] revealed a case of MGA in a case with Down syndrome but no other genetic abnormality was detected apart from the trisomy of chromosome twenty-one which is related to Down syndrome.

The prevalence of MGA is unknown; however, a recent study reports a prevalence of 2.6/100,000 in Stockholm, Sweden. It is usually diagnosed by the age of two. Occasionally, delayed presentations may be observed around the age of five years. Both genders are equally impacted, with symmetrical involvement of both the right and left eyes. It is less frequent among black individuals in the United States. It is predominantly unilateral; although, bilaterality was observed in roughly 16% of instances. In most cases, the visual prognosis is bad, and the range of possible values is from 20/100 to 20/200. The presence of a serous retinal detachment is possible in thirty percent of cases. Although it is not completely understood, it is hypothesized that there is a link among the subarachnoid space in the posterior excavation and the subretinal region. There is a potential for cerebrospinal fluid to migrate subretinally, which could lead to the retina's complete detachment. Evidence supporting this hypothesis includes the resolution of serous detachment following optic nerve sheath fenestration as well as the emergence of a gas bubble in the subarachnoid region following vitrectomy with gas injection and optic nerve sheath fenestration [8].

As many as fifty percent of individuals may exhibit concomitant cerebrovascular abnormalities. Alterations in the endocrine, respiratory, and renal systems may also manifest. It may be linked to a mutation of the PAX6 gene, neurofibromatosis type 2, and CHARGE syndrome. The predominant finding reported is trans-sphenoidal basal encephalocele accompanied by mid-facial abnormalities. Individuals exhibiting these characteristics generally possess a broad cranium, a recessed nasal bridge, and a deformity or cleft in the mid-upper lip. If the hypothalamus is implicated in the encephalocele, hypopituitarism may arise, impacting the generation of growth hormone and antidiuretic hormone [9]. Agenesis of the corpus callosum, absent optic chiasma, and an association with renal agenesis may also occur [10]. Visual impairment, optic nerve glioma, nystagmus, corneal leucoma,

strabismus, microphthalmia, cataract, and persistent hyperplastic primary vitreous [PHPV] are some of the ocular abnormalities that may present. Patients with MGA often have low levels of visual acuity [11]. The average best-corrected visual acuity in research using 12 MGA eyes was 20/300, with a range of 20/30 [hand motion included]. Strabismus associated with MGA is of the horizontal deviation [exotropia or esotropia] commonly, but few vertical deviations have been recorded. Eighty percent of individuals may involve strabismus. It is usual to see visual field defects and relative afferent pupillary deficiencies [12].

A large, funnel-shaped excavation of the optic nerve head as well as peripapillary retina is observed during a fundus examination. The surrounding annular pigmentary changes are associated with a central glial tuft, and multiple narrow branches of retinal vessels radiate from the optic disc margin in a straight fashion. "Macular capture" is a phenomenon that may involve the adjacent macula in the broader excavation area. Other fundus findings may include retinal detachment and subretinal fluid [13].

Numerous clinical forms exist. Previous reports have documented contractile forms, which occasionally result in a brief amaurosis due to the papilla's contraction movements [14]. Their movements could be explained by the presence of smooth muscle fibre heterotopic tissue in the choroid, which is where contraction occurs. Furthermore, the improper connectivity between the retinal and sub-arachnoid spaces would allow the flux and reflux of sub-arachnoid fluid, which would explain the pulsatile nature [15]. There may be a connection among the mechanism and the fluctuation of venous pressure during respiration, in accordance with Sugar [16]. In his MGA case reports, Yamana documented instances of macular aplasia [17].

Amblyopia, strabismus, and nystagmus are the normal prerequisites for its detection. When we examined the patient, they did not exhibit this symptom of deep amblyopia. Common forms of refraction anomalies included myopia, hypermetropia, and astigmatism. All of the refractive abnormalities other than hypermetropia were likewise revealed by our observations. Most people have unimprovable visual acuity of 20/200 or worse [17]. In our case, the patient's visual acuity is tracking hand movement. Nevertheless, reports of 20/20 acuities have been made [18]. After functional therapy, a 2 ½-year-old infant with MGA, exotropia, and unilateral severe amblyopia showed a visual acuity recovery of 20/100, in accordance with Sugar HS [16]. Because of this, occlusion therapy is essential for treating optic nerve abnormalities that are asymmetrical or unilateral.

In order to diagnose an optic disc abnormality, OCT is a useful tool. A well-defined epiretinal membrane is centripetally pulling on the retina, based on the research [19]. There have been very few reports of optic disc constriction in MGA cases. OCT is useful for analysing these motions. The swept-source OCT characteristics were delineated by Yoshida *et al.* The area at the base of the peripapillary excavation shrank during contraction, according to their research [20].

The most prominent motion throughout contraction was the forward movement of the peripapillary excavation walls, as stated by Rajendran and Kumar in their description of the spectral domain OCT findings [21]. There were two or three times every minute of the optic disc contracting, and each one lasted four or five seconds. Sawada *et al.* also observed similar results [22].

Features of OCT angiography in MGA cases with contractile optic discs have been documented by Cennamo *et al.* In the peripapillary capillary layer, they discovered a dense microvascular network that was

identical to the deep vascular plexus surrounding the optic nerve and the superficial vascular plexus [23]. OCT can identify small slit-like fissures at the excavation's edge in cases of retinal detachment. Direct connectivity between the vitreous and subarachnoid space can be revealed, and superficial retinal detachments can be detected. Tests for peripapillary vitreous traction are possible [24].

Lytvynchuk LM et al. recorded a case of MGA retinal detachment treated utilizing intraoperative OCT-assisted pars plana vitrectomy. The retina detached because of intense vitreous traction and adhesion that was discovered above the optic disc and macula. The epiretinal and internal limiting membranes were penetrated, and an air tamponade was administered to induce posterior vitreous detachment. Retinal reattachment was subsequently attained [25].

Many people have different opinions on how MGA develops. Schneider initially thought of it as a subtype of coloboma, characterized by a central colobomatous defect that results from an abnormality in the closure of the embryonic fissure [15]. Neuroectodermal closure defect does not exist, in accordance with pathological research by Mauscho, because the retinal pigment epithelium and the inner border of the papilla do not meet at a point of continuity [26]. MGA, in his view, is not an optic nerve coloboma but rather a congenital disease of the mesodermal layer. To add to that, Pollock proposed that MGA is a main mesenchymal anomaly caused by a combination of factors, including incomplete scleral posterior wall closure and underdeveloped cribriform plate. Mohamed also reported that it may run in families at times [18]. In this example, **Nagy et al.** detailed a mother and daughter's experience with MGA [27].

A differential diagnosis of MGA includes optic disc coloboma. In contrast to optic disc coloboma, which involves excavation within the optic disc, usually in the inferior aspect of the disc, as well as the discernible superior neuro-retinal margin, MGA excavates the entire disc [28].

We also considered posterior staphyloma as a possible diagnosis for our patient with extreme myopia. A normal retinal vasculature and a flat optic disc devoid of a central tuft characterize posterior staphyloma, which is characterized by excavation of the posterior fundus surrounding the optic disc [2]. Additionally, there is an abnormal retinal vasculature and a central tuft of tissue on the disc. Optic disc pit, or ODP, is another possible diagnosis that was suggested. ODP is typically characterized by a single, oval-shaped, greyish-white excavation in the optic disc's inferotemporal quadrant. It is usually unilateral. Pit dimensions vary from 1/8 to 1/4 of the diameter of the optic disc [29]. A serous retinal detachment, macular schisis, or macular oedema can complicate the condition, but in most cases, vision is not severely affected [30].

It is possible for advanced glaucomatous optic neuropathy [AGON] to pose as MGA. An enlarged cup, either all over or in one specific area, with a vertical cup disc ratio of 0.9 is an indicator of AGON. Thinning of the neuro-retinal rim is typically observed at the superior and inferior poles. Vessels become more nasalized and circum-linear vessels become more visible, in addition to atrophy of the parapapillary space and the presence of laminar spots [31].

MGA is typically treated as an isolated condition. Basal encephalocele may be present in conjunction with this condition. In addition, adolescents with MGA are at risk of developing endocrine, respiratory, in addition to neurological disorders. The median changes of the face, such as hypertelorism and anomalies of the base of the palate, nose, as well as anterior section of the base of the cranium, are found in

some of these individuals on occasion. These individuals may even display symptoms of a hypothalamic-pituitary hernia in certain instances. One-third of patients lack optic chiasma. In seventy-five percent of individual instances, the trans-sphenoidal encephalocele is linked to the agenesis of the corpus callosum [32]. Some cases of type 2 neurofibromatosis, CHARGE syndrome, Moya-Moya disease, sella turcica abnormality, trisomy 4q, as well as other genetic disorders have been linked [3]. There is disagreement on the aetiology of linked retinal detachments. Predominantly, these retinal detachments are thought to originate from sources other than retinal breaks, and may rarely be rhegmatogenous in nature. It is crucial to diagnose and treat MGA early. Preventing amblyopia requires correction of strabismus and anisometropia. Glasses are prescribed after a full cycloplegic refraction. Surgical correction of squint is possible. An accompanying issue that necessitates surgical therapy is retinal detachment. Retinal reattachment was documented in every case of pars plana vitrectomy by **Chang et al.** [8] and **Zhang et al.** [33].

There have been instances of spontaneous attachment in patients with retinal detachment. After 7.5 years, four patients showed signs of spontaneous attachment, according to **Haik et al.** Because of this, thinking about this aspect before undergoing surgery is essential [34].

In the case of associated systemic abnormalities, the importance of a multidisciplinary approach in the management of patients with this condition is crucial.

It is mandatory to closely observe the other eye in situations with unilateral MGA. A detachment of the retina or cataract could form in the other eye. Consequently, vitreoretinal surgery or phacoemulsification with lens implantation are performed.

Conclusion: Ocular and non-ocular symptoms can coexist in MGA, a rare congenital aberration of the optic disc that can cause visual loss. On the event that a case of MGA is detected, it is imperative that the patient undergoes comprehensive clinical evaluation, a comprehensive morphological evaluation, regular monitoring, and multidisciplinary treatment.

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