SEVERE JUVENILE OPEN ANGLE GLAUKOMA IN A TEENAGER: A CASE REPORT

Nadya Divina Baihaqi [1], Rika Agustanti [2], Niken Indah Noerdiyani [3]
Faculty of Medicine, Airlangga University, Indonesia [1]
Rumah Sakit Mata Masyarakat, Surabaya, Indonesia [2,3]
*Corresponding Author e-mail: nadivbaihaqi@gmail.com

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Abstract: *Juvenile open-angle glaucoma (JOAG) is a rare but severe form of glaucoma affecting individuals between the ages of 3 and 40. While it shares pathophysiological features with primary open-angle glaucoma (POAG), JOAG often progresses more rapidly and is associated with higher intraocular pressure (IOP) at diagnosis. Genetic mutations, such as in the MYOC and OPTN genes, are common in JOAG, though family history may not always be present. Early diagnosis and intervention are crucial to prevent permanent vision loss. A 15-year-old girl presented with blurry vision and frequent headaches, which had progressively worsened over two years. She experienced sudden loss of vision in the left eye a year prior and episodes of tunnel vision. She had no family history of glaucoma. Examination revealed visual acuity of 6/15 in the right eye and no light perception in the left. IOP was 23 mmHg in the right eye and 18.3 mmHg in the left, despite treatment with Timolol and Latanoprost. Optic nerve examination showed advanced glaucomatous optic neuropathy (GON), with a cup-to-disc ratio of 0.9 in the right eye and 1.0 in the left. Humphrey perimetry revealed severe visual field defects, and optical coherence tomography (OCT) showed significant retinal nerve fiber layer (RNFL) thinning in both eyes. Despite medical therapy, her condition continued to deteriorate, indicating a need for surgical intervention to better control IOP and prevent further vision loss. Gonioscopy confirmed open anterior chamber angles, consistent with JOAG. This case underscores the importance of early detection and aggressive management in JOAG. Despite medical therapy, the patient exhibited significant optic nerve damage and visual field loss, necessitating surgical consideration. Given the rapid progression, early surgical intervention may offer better outcomes in controlling IOP and preserving vision. Long-term follow-up and comprehensive management are crucial to preventing further deterioration. Genetic testing may help clarify the etiology, particularly in cases with no family history.*

Kata Kunci:
Juvenile Open-Angle Glaucoma, Tekanan Intraokular, Neuropati Optik.

Abstrack: Juvenile open-angle glaucoma (JOAG) merupakan bentuk glaukoma yang jarang namun parah, yang mempengaruhi individu berusia antara 3 hingga 40 tahun. Meskipun berbagi fitur patofisiologis dengan glaukoma sudut terbuka primer (POAG), JOAG sering berkembang lebih cepat dan dikaitkan dengan tekanan intraokular (IOP) yang lebih tinggi saat diagnosis. Mutasi genetik, seperti pada gen MYOC dan OPTN, umum terjadi pada JOAG, meskipun riwayat keluarga mungkin tidak selalu ada. Diagnosis dan intervensi dini sangat penting untuk mencegah kehilangan penglihatan permanen. Seorang gadis berusia 15 tahun datang dengan keluhan penglihatan kabur dan sering sakit kepala, yang semakin memburuk selama dua tahun terakhir. Dia mengalami kehilangan penglihatan mendadak pada mata kiri setahun sebelumnya dan episode penglihatan terowongan. Dia tidak memiliki riwayat keluarga glaukoma. Pemeriksaan menunjukkan ketajaman visual 6/15 pada mata kanan dan tidak ada persepsi cahaya pada mata kiri. IOP adalah 23 mmHg di mata kanan dan 18,3 mmHg di mata kiri, meskipun telah diobati dengan Timolol dan Latanoprost. Pemeriksaan saraf optik menunjukkan neuropati optik glaukomatosa (GON) lanjut, dengan rasio cup-to-disc 0,9 pada mata kanan dan 1,0 pada mata kiri. Perimetri Humphrey menunjukkan cacat lapang pandang yang parah, dan tomografi koherensi optik (OCT) menunjukkan penipisan lapisan serabut saraf retina (RNFL) yang signifikan pada kedua mata. Meskipun terapi medis telah dilakukan, kondisinya terus memburuk, mengindikasikan perlunya intervensi bedah untuk mengontrol IOP dengan lebih baik dan mencegah hilangnya penglihatan lebih lanjut. Gonioskopi mengonfirmasi adanya sudut bilik anterior terbuka, sesuai dengan JOAG. Kasus ini menekankan pentingnya deteksi dini dan manajemen agresif pada JOAG. Meskipun terapi medis, pasien menunjukkan kerusakan saraf optik dan kehilangan lapang pandang yang signifikan,

sehingga diperlukan pertimbangan bedah. Mengingat perkembangan yang cepat, intervensi bedah dini mungkin memberikan hasil yang lebih baik dalam mengontrol IOP dan menjaga penglihatan. Tindak lanjut jangka panjang dan manajemen komprehensif sangat penting untuk mencegah kerusakan lebih lanjut. Tes genetik dapat membantu mengklarifikasi etiologi, terutama dalam kasus tanpa riwayat keluarga.

Introduction

Juvenile open-angle glaucoma (JOAG) is a rare yet serious form of glaucoma, typically diagnosed in individuals between the ages of 3 and 40 years1. While primary open-angle glaucoma (POAG) is more common in older adults, the incidence of JOAG is much lower, with estimates ranging from 0.38 to 1 per 50,000 individuals globally. JOAG often presents with more aggressive progression compared to POAG, particularly in young individuals, and is associated with higher intraocular pressure (IOP) levels at the time of diagnosis. Despite its rarity, JOAG can have devastating effects on vision if not detected and treated promptly2.

Clinically, JOAG shares pathophysiological features with POAG, particularly in its mechanism involving trabecular meshwork dysfunction and impaired aqueous humor drainage. However, JOAG typically has a strong genetic component, with mutations in genes such as MYOC, which accounts for approximately 10% of cases, and OPTN. This genetic link is often associated with a family history of glaucoma, underscoring the need for early screening in at-risk populations.

The diagnosis of JOAG is challenging due to its insidious onset and asymptomatic nature in the early stages. Common diagnostic criteria include elevated IOP (often above 30 mmHg), optic nerve head cupping, and characteristic visual field defects. Gonioscopy typically reveals an open anterior chamber angle, which distinguishes JOAG from other forms of childhood glaucoma. Early diagnosis is crucial, as the disease can progress rapidly, leading to optic nerve damage and permanent vision loss if untreated.

Management of JOAG focuses on reducing intraocular pressure to halt disease progression. Initial treatment usually involves topical medications, such as prostaglandin analogs, beta-blockers, or carbonic anhydrase inhibitors, aimed at lowering IOP. However, due to the aggressive nature of the disease, many patients require surgical interventions such as trabeculectomy or glaucoma drainage devices to achieve adequate IOP control. Long-term follow-up is essential, as the disease often requires ongoing adjustments in therapy to prevent further deterioration of vision. In this case report, we discuss a teenager diagnosed with severe JOAG, emphasizing the clinical presentation, diagnostic challenges, and the therapeutic interventions that were implemented. This case highlights the importance of early detection and aggressive management to prevent rapid progression to blindness in young patients.

Research Method

This study is a case report using a clinical descriptive method, where data were collected through physical examination, laboratory tests, and ophthalmological assessments of a patient diagnosed with juvenile open-angle glaucoma (JOAG). The patient involved in this study is a 15-year-old female adolescent who has experienced progressive visual deterioration over the past two years. Anamnesis data were collected through direct interviews with the patient, including family history, initial symptoms, and any prior history of disease or trauma. The patient reported no family history of glaucoma.

Standard physical examination was performed to measure blood pressure, body temperature, pulse rate, and the patient's neurological status. Ophthalmological assessments included measurement of visual acuity, intraocular pressure (IOP) using tonometry, and examination of both the anterior and posterior segments of the eye. Gonioscopy was conducted to assess the anterior chamber angle, while Humphrey perimetry was used to evaluate the patient's visual field defects. Further testing using Optical Coherence Tomography (OCT) was performed to evaluate retinal nerve fiber layer (RNFL) thickness and detect the presence of glaucomatous optic neuropathy (GON). This data was then analyzed descriptively to illustrate the patient's clinical condition and her response to the prescribed therapy, including the use of medications such as Timolol and Latanoprost.

The patient's response to medical therapy was closely monitored, and the decision for surgical intervention was considered based on the patient's clinical progression, particularly regarding IOP control and the progression of optic nerve damage. This study also emphasizes the importance of long-term follow-up to monitor disease progression and treatment efficacy, as well as considering genetic testing to further understand the etiology of glaucoma in this patient.

Results and Discussion

A 15 year old teenage girl came with the main complaint of blurry eyes. Blurred eyes on the right and left side accompanied by throbbing that has been felt for approximately 2 years. Since one year ago, to be precise in May 2023, the left eye, suddenly darkened, the head often hurts and has been unconscious. myself due to headaches, followed by a peeping sensation in my right eye (narrowed vision) in January 2024. When walking, I sometimes hit surrounding objects.

The patient fell from a bicycle one year ago and hit his head but did not hit his eye and had no history of nosebleeds. The patient has been wearing glasses since the 7th grade of junior high school, I don't remember the size at that time. This kind of disease in the family is denied. Birth history showed that he was born normal with a weight of 2800 grams, body length 46 cm. Febrile seizures were denied and epilepsy was denied. History of normal growth and development according to the growth period. History of allergies was denied.

On physical examination, generalist status was found as follows: GCS = 456, blood pressure 100/60 mmHg, pulse 80 x/minute, respiration rate (RR) of 20 x/minute and temperature 36.50C. Head and neck: conjunctiva is not anemic, sclera is not icteric. On chest examination, chest movement was symmetrical, there was no retraction. The lungs had vesicular breath sounds, no wheezing or rhonchi. On cardiac examination, a single S1S2 was found, there were no murmurs or gallops. Abdomen looks supple, bowel sounds are normal. The acral extremities feel warm, dry, red.

The patient's local status was found to be VOD: 6/15 with C-4.00 A 1800 correction to 6/10, VOS was found to be LP negative. Right eye intraocular pressure 23 mmHg, left eye intraocular pressure 18.3 mmHg with administration of Timolol maleate 0.5% eye drops twice a day, Acetazolamide 4 times a day 250 mg, Latanoprost eye drops once a day. The eyelids, conjunctiva, cornea and lens appeared normal in both eyes, the anterior chamber was clear and the angles were open with Von Herick IV assessment of the right and left eyes. On examination of the posterior segment, NII papillae were clearly defined and pale in color in both eyes, the C/D ratio in the right eye was 0.9 while the left eye was 1.0; There was nasalization of blood

vessels in the optic nerve papillae accompanied by bearing and bayonating in both eyes. There were no peri-papillary hemorrhages or peripapillary atrophy. This shows the presence of signs of GON in both eyes which can be observed in figure. The presence of GON is a typical sign of glaucoma in both eyes of the patient and the overall clinical picture is presented in table 1 below. Which includes the clinical picture of the anterior segment and posterior segment of both eyes. Anterior segment photos can be seen in figure.

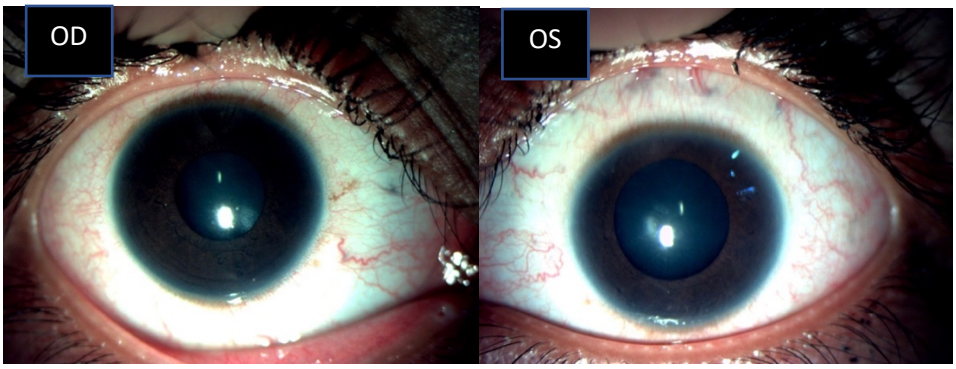

Figure 1. The anterior segment shows an anisocoric pupil with an OD of 3 mm and an OS of 5 mm

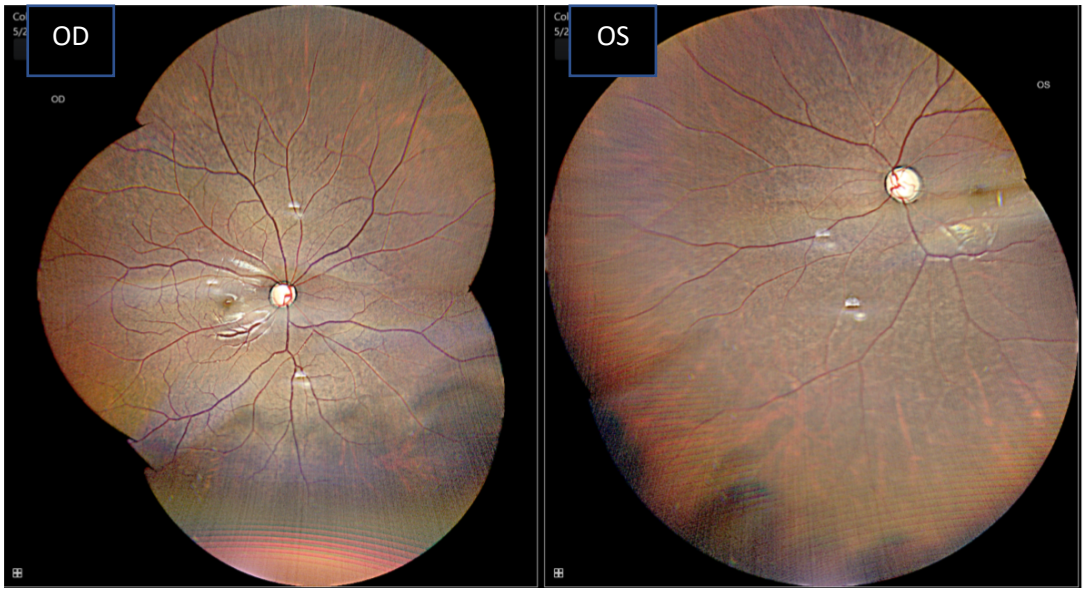

Figure 2. Signs of glaucomatous optic neuropathy (GON) were found in both eyes

Table 1. Clinical features of the anterior segment and posterior segment of both eyes

OD	Examination	OS
6/15 Correction C -4.00 A 180-- -- 6/10	**Visus**	LP -
21,3 mmHg	**Intraocular pressure**	18,3 mmHg
Anterior Segment		
Oedema (-) spasme (-)	**Palpebra**	Oedema (-) spasme (-)
Conjungtival injection (-), Cilliary injection (-), hyperemic (-), follicles (-)	**Conjungtiva**	Conjungtival injection (-), Cilliary injection (-), hyperemic (-), follicles (-)
Clear ∅ 11-12	**Cornea**	Clear ∅ 11-12
Dalam VH IV	**Anterior chamber**	Dalam VH IV
Radier	**Iris**	Radier
Round, diameter 3mm RC (+)	**Pupil**	Round, diameter 3mm RC(+)
Clear	**Lens**	Clear
OD	**Examination**	OS
Posterior Segment		
Positive	**Fundus reflex**	Positive
Firm boundaries, pale color, CD 0,9 , Nasalisasi +, bayoneting +	**Papil N. II**	Firm boundaries, pale color, CD ratio 1,0 nasalisasi +,bayoneting +
Bleeding (-), exudate (-)	**Retina**	Bleeding (-), exudate (-)
positive	**Macula reflex**	positive

On gonioscopy examination it appears that both corners of the anterior chamber are open. Visual field examination with Humphrey perimeter showed that there was a visual field defect in the right eye in the form of a superior and inferior arcuate (biarcuate) which left a visual field of 10-15o central with the image of central tunnel vision. Also a visual field defect was found in the left eye which gave an incomplete superior arcuate image. and inferior leaving a central visual field of 15-20^{o}. The results of visual field examination with the Humphrey perimeter can be seen in pictures 3 and 4 below.

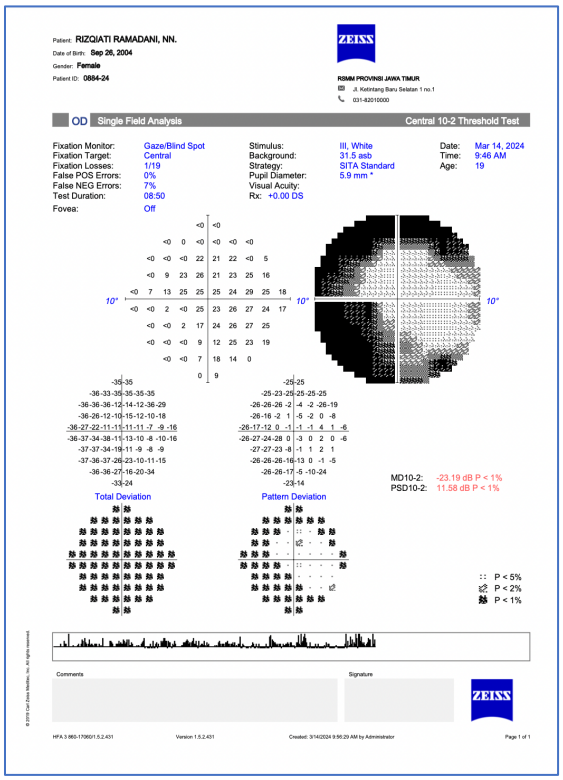

Figure 3. Humphrey's visual field in the right eye shows a biarcuata visual field defect

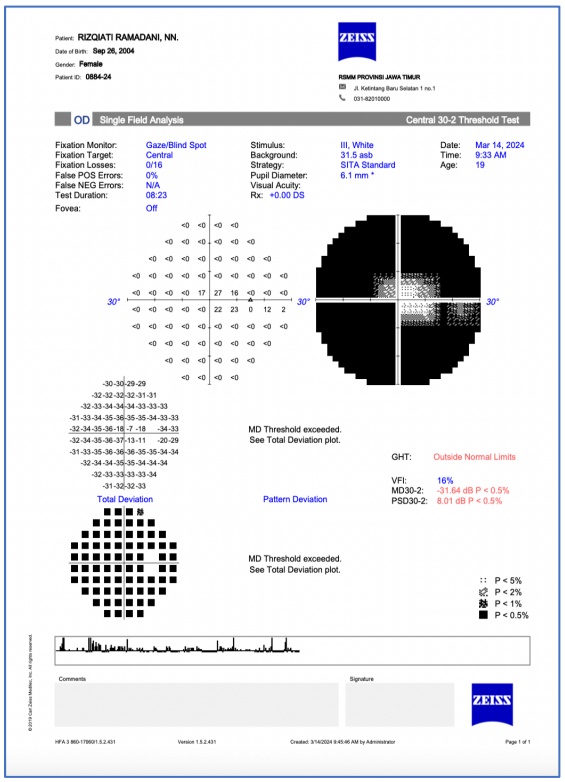

Figure 4. The left eye shows biarcuate scotoma with central tunnel vision

Optical Coherence Tomography (OCT) examination revealed significant thinning of the Retinal Nerve Fiber Layer (RNFL) in both eyes. In the right eye, RNFL thinning was noted in the superior and inferior quadrants, extending into the temporal region. In the left eye, thinning was observed in the superior and inferior quadrants as well. Additionally, OCT of the macula showed thinning in the right eye across the superior, inferior, and temporal quadrants, while in the left eye, thinning was limited to the nasal region.

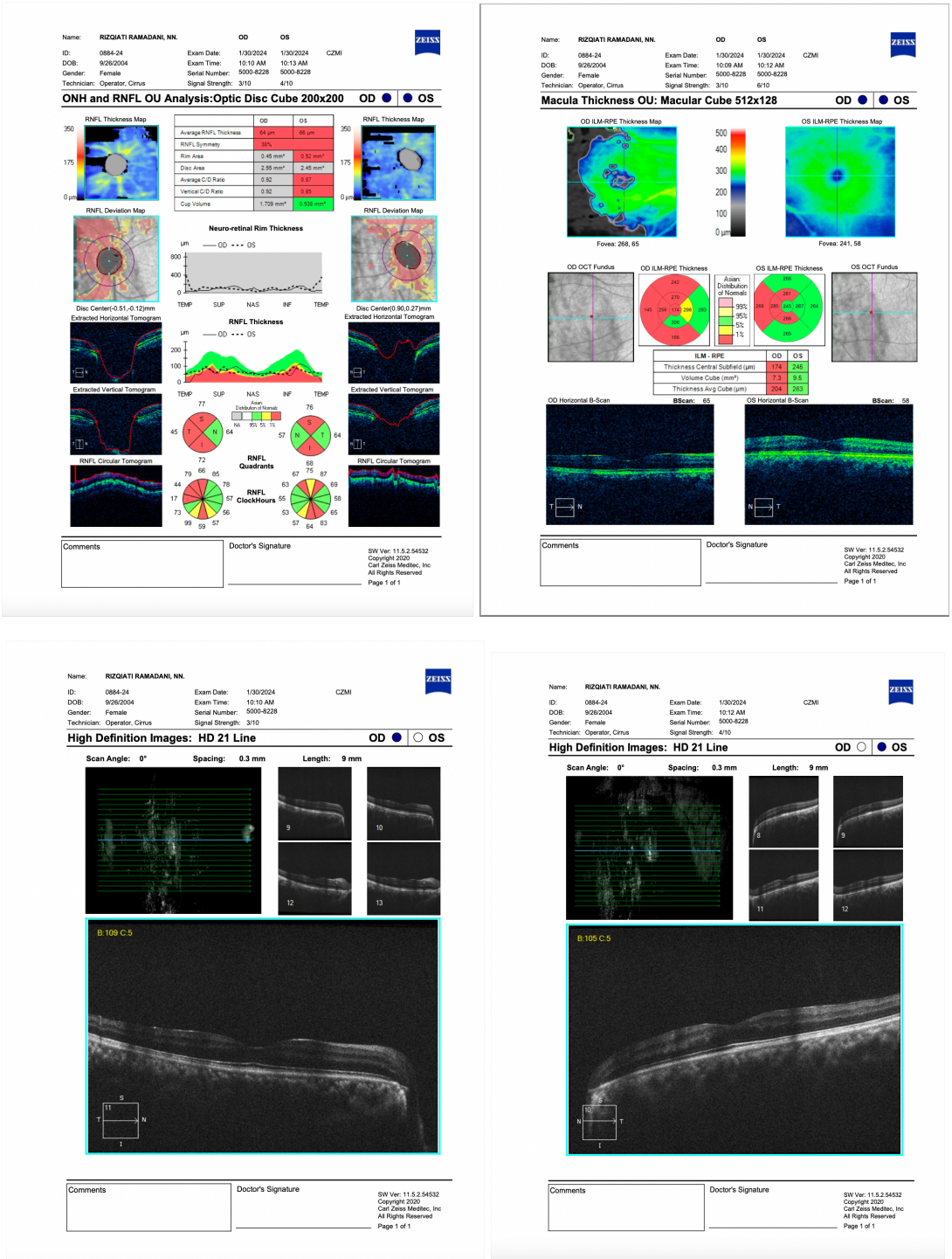

Figure 5. OCT examination

DISCUSSION

Juvenile open-angle glaucoma (JOAG) is a rare but aggressive form of primary open-angle glaucoma (POAG), occurring in individuals typically between the ages of 4 and 35. It is caused by increased intraocular pressure (IOP) due to dysfunction in the trabecular meshwork drainage system, leading to progressive damage to the optic nerve and vision loss. JOAG differs from adult-onset glaucoma in that it progresses more rapidly and often requires early and more aggressive management to prevent significant visual impairment.

The patient in this case, a 15-year-old girl, presents with severe signs of JOAG, including progressive visual field loss, optic nerve cupping, and elevated intraocular pressure (IOP). The pathophysiology of JOAG involves impaired aqueous humor outflow, resulting in chronic elevation of IOP. As in adult POAG, the optic nerve is gradually damaged due to the sustained pressure, leading to the characteristic optic nerve head changes seen in this case. The significant cupping (C/D ratios of 0.9 in the right eye and 1.0 in the left eye) and pale appearance of the optic disc indicate advanced glaucomatous optic neuropathy (GON), a hallmark of this disease.

The mechanism of optic nerve damage in JOAG is multifactorial. Elevated IOP compresses the lamina cribrosa, leading to axonal damage, disrupted axoplasmic flow, and retinal ganglion cell death. Additionally, vascular dysregulation and compromised blood flow to the optic nerve may further exacerbate nerve damage, as evidenced by the pale appearance of the optic disc in this patient. The nasalization of blood vessels and bayoneting seen in the optic nerve head are common in advanced glaucoma, indicating chronic pressure damage over time.

The patient's visual field defects, as demonstrated on the Humphrey perimeter test, are consistent with glaucomatous damage. The superior and inferior arcuate scotomas (biarcuate defects) in the right eye and the incomplete arcuate defects in the left eye reflect damage to the retinal nerve fibers in the corresponding areas of the optic nerve. These visual field patterns are typically seen in patients with advanced glaucoma and correspond to the thinning of the retinal nerve fiber layer (RNFL) detected by optical coherence tomography (OCT). The OCT findings of RNFL thinning in the superior, inferior, and temporal quadrants further confirm the extent of the optic nerve damage.

The progression of this patient’s symptoms—from gradual blurring of vision to sudden visual loss and tunnel vision—follows the natural course of untreated or poorly controlled JOAG. The patient’s early symptoms of headaches and blurred vision may have been initially dismissed, as JOAG can be asymptomatic until significant damage has occurred. The sudden worsening of symptoms in the left eye, including visual blackout and severe headaches, likely represents a rapid progression of optic nerve damage due to chronically elevated IOP.

JOAG is known to have a stronger genetic component compared to adult POAG. Mutations in genes such as MYOC (myocilin), OPTN (optineurin), and CYP1B1 are often implicated in the pathogenesis of JOAG. Myocilin mutations, in particular, are associated with early-onset glaucoma and contribute to increased resistance in the trabecular meshwork outflow, leading to elevated IOP. Although the patient denies a family history of glaucoma, the early onset of the disease and rapid progression suggest a possible genetic predisposition.

Genetic testing could be beneficial in confirming a hereditary cause and potentially informing the patient's future management and family planning.

JOAG can be challenging to diagnose early, as the disease is often asymptomatic in its early stages. Gonioscopy, performed in this case, confirmed the presence of open anterior chamber angles, ruling out angle-closure glaucoma, which can have a similar presentation. Visual field testing and OCT are crucial diagnostic tools in assessing the extent of optic nerve and visual field damage in JOAG. The Humphrey visual field testing, which showed significant central tunnel vision in the right eye and reduced peripheral vision in the left eye, correlates with the extensive optic nerve damage observed on OCT.

Management of JOAG focuses on lowering IOP to prevent further damage to the optic nerve. The current treatment regimen, which includes timolol maleate, acetazolamide, and latanoprost, is aimed at reducing aqueous humor production and increasing outflow. However, despite these interventions, the right eye IOP remained elevated (23 mmHg), suggesting that the current medical therapy may not be sufficient. This resistance to medical treatment is common in JOAG due to the aggressive nature of the disease.

Theories regarding IOP management in JOAG emphasize the need for early surgical intervention when medical therapy fails to control IOP adequately. Procedures such as trabeculectomy or glaucoma drainage devices (e.g., Ahmed valve or Baerveldt implant) are often required to achieve long-term control of IOP in JOAG patients. Surgical intervention could be a viable option for this patient, given the insufficient response to medications and the risk of further vision loss. Studies have shown that surgical options for JOAG generally result in better long-term outcomes compared to medical therapy alone, particularly in preserving the remaining visual field.

The patient's history of head trauma from a fall may also be of concern, though it is unlikely to be a primary cause of her glaucoma. While trauma can lead to angle-recession glaucoma or traumatic optic neuropathy, the absence of direct ocular trauma and the lack of acute post-traumatic symptoms make it less likely that the head injury played a significant role in the disease progression. However, it is essential to rule out any additional factors that could exacerbate her condition, such as secondary effects of trauma or vascular dysregulation.

Conclusion

In conclusion, this case of severe JOAG in a 15-year-old highlights the importance of early detection, comprehensive diagnostic evaluation, and aggressive management in preventing further vision loss. Given the advanced optic nerve damage, visual field defects, and insufficient response to medical therapy, surgical intervention should be considered. Early surgical treatment has been shown to be more effective in controlling IOP and preventing irreversible vision loss in JOAG. Genetic testing may provide further insight into the etiology of the disease, particularly in cases with no apparent family history. Long-term follow-up is critical in managing JOAG, as the risk of progression remains high despite initial treatment efforts.

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