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OPIS PRZYPADKU
CASE REPORT

Familial angle-closure glaucoma in a 48-year-old female patient: A case report

Rodzinna jaskra zamkniętego kąta u 48-letniej pacjentki – opis przypadku

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ABSTRACT

Primary angle-closure glaucoma (PACG) affects nearly 17 million people worldwide. A shallow anterior chamber and a short axial length increase the risk of angle closure and development of glaucomatous neuropathy. The progress of PACG is also associated with genetic predisposition. A 48-year-old female patient presented to the Outpatient Ophthalmology Clinic. Her sister had undergone two central decompressing vitrectomies due to advanced PACG. The patient was referred for additional examinations: tonometry, anterior segment optical coherence tomography (AS-OCT), gonioscopy, and static perimetry. The patient was qualified for prophylactic laser peripheral iridotomy (LPI) in both eyes. AS-OCT and gonioscopy revealed a closed angle in the right eye (*oculus dexter* – OD), a slit-like angle in the left eye (*oculus sinister* – OS), and a shallow anterior chamber in both eyes. The intraocular pressure (IOP) was measured at 20 mmHg in the OD and 15 mmHg in the OS. A peripheral relative visual field defect was found in the OD. Follow-up tonometry performed 14 months after the initial LPI showed an IOP of 27 mmHg in the OD. Timolol 0.5% was prescribed. Despite this, after 3 months, the IOP in the OD measured 22 mmHg. The patient was qualified for a repeat LPI in the OD. At follow-up, the IOP was 21 mmHg in the OD. Visual acuity was 1.0 in both eyes. LPI is the first-line treatment for PACG. Even though this procedure is highly effective, it does not guarantee long-term results. In case of recurrence, qualification for anti-glaucoma surgical intervention should be considered.

KEYWORDS

angle-closure glaucoma, laser peripheral iridotomy, familial glaucoma, glaucomatous optic neuropathy

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STRESZCZENIE

Jaskra pierwotna zamkniętego kąta (*primary angle-closure glaucoma* – PACG) dotyka ponad 17 milionów ludzi na świecie. Płytka komora przednia i krótka długość osiowa gałki ocznej zwiększają ryzyko zamknięcia kąta przesączania, a w rezultacie rozwoju neuropatii jaskrowej. Rozwój PACG jest także związany z predyspozycją genetyczną. Do poradni okulistycznej zgłosiła się 48-letnia pacjentka. Siostra pacjentki została dwukrotnie poddana witrektomii centralnej odbarczającej z powodu zaawansowanej PACG. Pacjentce zlecono badania dodatkowe: tonometrię, optyczną koherentną tomografię przedniego odcinka oka (*anterior segment optical coherence tomography* – AS-OCT), gonioskopię i perymetrię statyczną. Pacjentka została zakwalifikowana do profilaktycznej laserowej irydotomii obwodowej (*laser peripheral iridotomy* – LPI) obu oczu. W badaniach AS-OCT i gonioskopii stwierdzono zamknięty kąt przesączania w oku prawym (*oculus dexter* – OD), szczelinowaty w oku lewym (*oculus sinister* – OS) i spłyconą komorę przednią w obu oczach. Ciśnienie wewnątrzgałkowe (*intraocular pressure* – IOP) wynosiło 20 mmHg w OD i 15 mmHg w OS. Stwierdzono obwodowy ubytek względny pola widzenia w OD. Tonometria kontrolna po 14 miesiącach od wykonania LPI wykazała, że IOP OD wynosi 27 mmHg. Zalecono tymolol 0,5%. Mimo to po 3 miesiącach IOP OD wynosiło 22 mmHg. Pacjentkę zakwalifikowano do powtórnego zabiegu LPI OD. W badaniu kontrolnym IOP OD wynosiło 21 mmHg. Ostrość wzroku wynosiła 1,0 w obu oczach. LPI jest metodą pierwszego rzutu w leczeniu PACG. Mimo wysokiej skuteczności zabieg nie gwarantuje długotrwałego efektu. W razie nawrotu należy rozważyć kwalifikację do zabiegu przeciwjaskrowego.

SŁOWA KLUCZOWE

jaskra zamkniętego kąta, laserowa irydotomia obwodowa, jaskra rodzinna, neuropatia jaskrowa

INTRODUCTION

It is estimated that primary angle-closure glaucoma (PACG) affects nearly 17 million people over 40 years old worldwide [1]. In Europe, even 50% of glaucoma cases remain undiagnosed [2]. Anatomical factors such as a shallow anterior chamber and a short axial length of the eyeball increase the risk of angle closure, which leads to glaucomatous neuropathy [1,3,4]. This is one of the leading causes of irreversible vision loss worldwide, affecting up to 1 in 9 people with PACG, while 1.8% of patients lose their vision completely [5]. The development of PACG is also related to age, hyperopia, female sex, and genetic predispositions.

As many as 70% of PACG cases occur in the Asian population [1], and the risk is 3.7 times higher in siblings of affected individuals compared to the general population [4].

The development of PACG is the effect of a polygenic predisposition that affects the morphologic and biochemical parameters of the eyeball [4]. The gene that definitely leads to PACG is *NNO1*, located on chromosome 11 [4,6]. Research shows that there are also eight other loci of genetic liability; however, their role – similarly to that of other genes suspected of association with PACG – needs to be further explored [4,6].

In patients with angle-closure due to a pupillary block mechanism, the first-line strategy for forestalling the development of PACG is preventative laser peripheral iridotomy (LPI) using a Nd:YAG laser [2,7].

CASE REPORT

A 48-year-old female patient presented to the Outpatient Ophthalmology Clinic at the Prof. K. Gibiński University Clinical Center of the Medical University of Silesia in Katowice. Her sister had previously undergone two central decompressing vitrectomy procedures due to advanced PACG. The patient's gonioscopy revealed an appositionally closed anterior chamber angle in the right eye (*oculus dexter* – OD) and a slit-like angle with partially visible trabecular meshwork in the left eye (*oculus sinister* – OS). Anterior segment optical coherence tomography (AS-OCT) using a CASIA2 device showed shallow anterior chambers in both eyes, measuring 1.73 mm in the OD and 2.62 mm in the OS (Figure 1). The patient's best corrected visual acuity (BCVA) was measured at 1.0. In the autorefractometry, the sphere measurement was -0.5 diopters in the OD and +2 diopters in the OS, with a cylindrical component of +0.5 for both eyes and an axis measurement of 128 degrees for the OD and 20 degrees for the OS. Eye fundus examination showed no abnormalities of the optic disc. However, automated

static perimetry on an Octopus perimeter revealed a peripheral visual field defect of -1.8 dB in the OD, which is located in the lower percentile of the Bebie curve (Figure 2). Intraocular pressure (IOP) was 20 mmHg in the OD and 15 mmHg in the OS.

Following the assessment of the clinical presentation and additional tests, the patient was qualified for prophylactic LPI in both eyes. The procedure in the OD was completed without complications. A postoperative IOP of 12 mmHg was successfully achieved, as confirmed by tonometry.

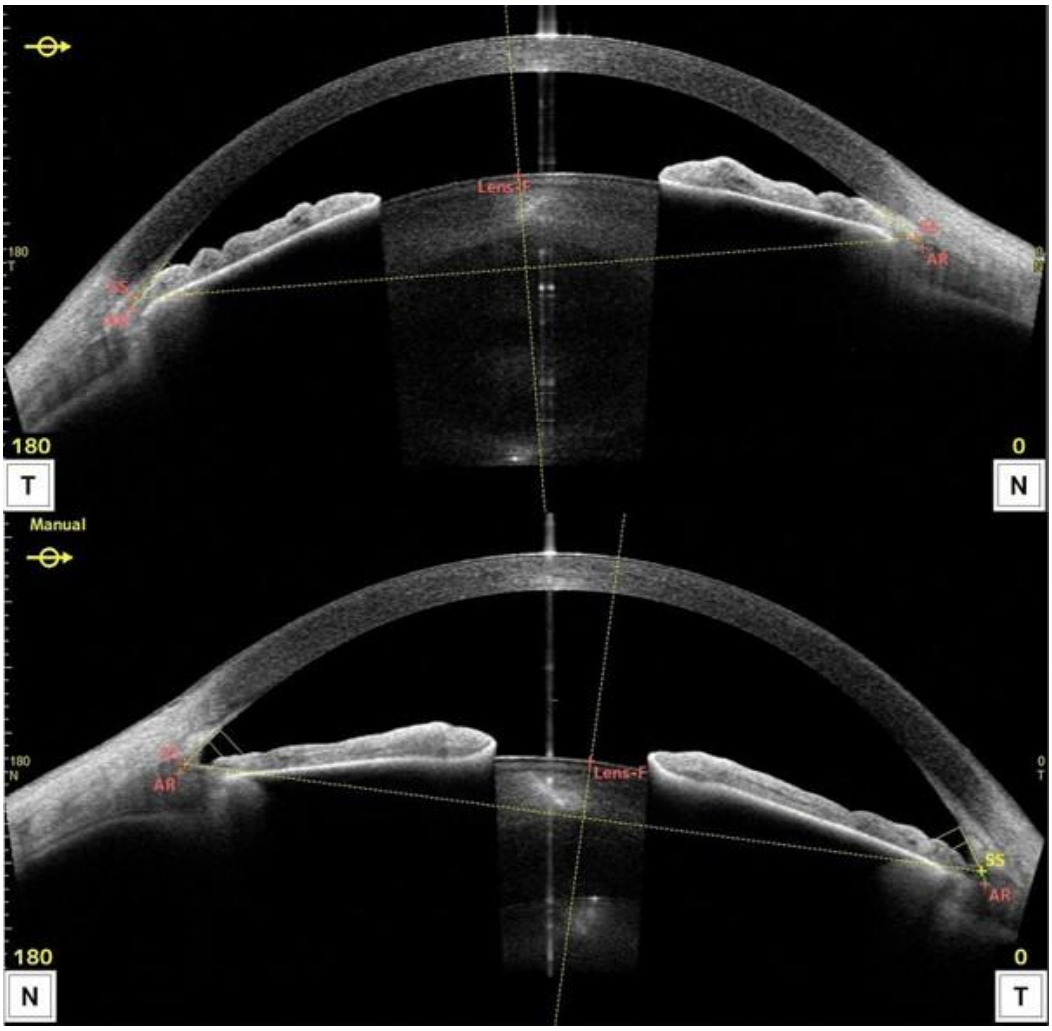

Fig. 1. Anterior segment optical coherence tomography of the right eye and the left eye performed with CASIA2

Three months later an anterior segment eye examination revealed a patent iridotomy opening, a visual acuity of 1.0 with correction, and an IOP of 20 mmHg. LPI was performed in the OS without complications; the postoperative IOP was recorded at 10 mmHg.

During follow-up examinations, an increase in IOP was noted in the OD (Figure 3). The IOP in the OD reached 27 mmHg 1 year after the initial LPI. Administration of timolol 0.5% eye drops twice daily resulted in a reduction of IOP to 16 mmHg at a follow-up visit 2 weeks later. However, within 2 months, the IOP rose again to 22 mmHg.

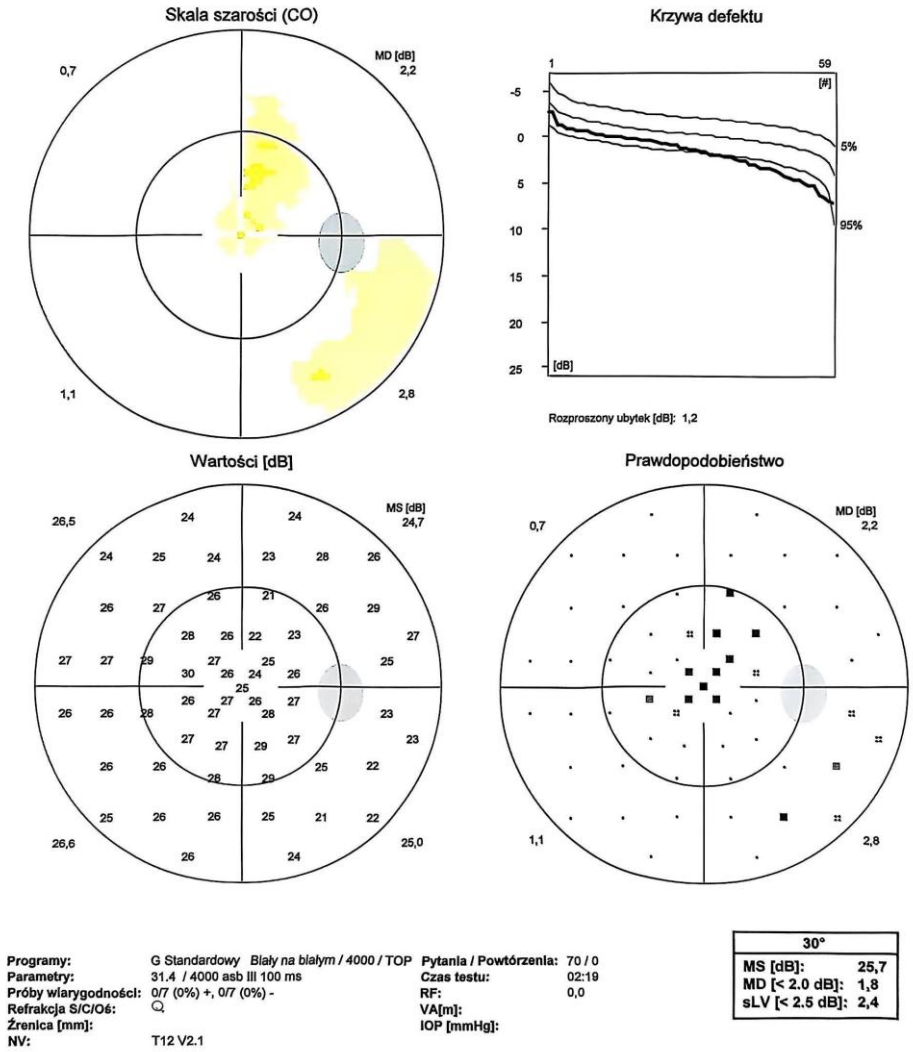

Fig. 2. Automated static perimetry of the right eye performed with an Octopus perimeter

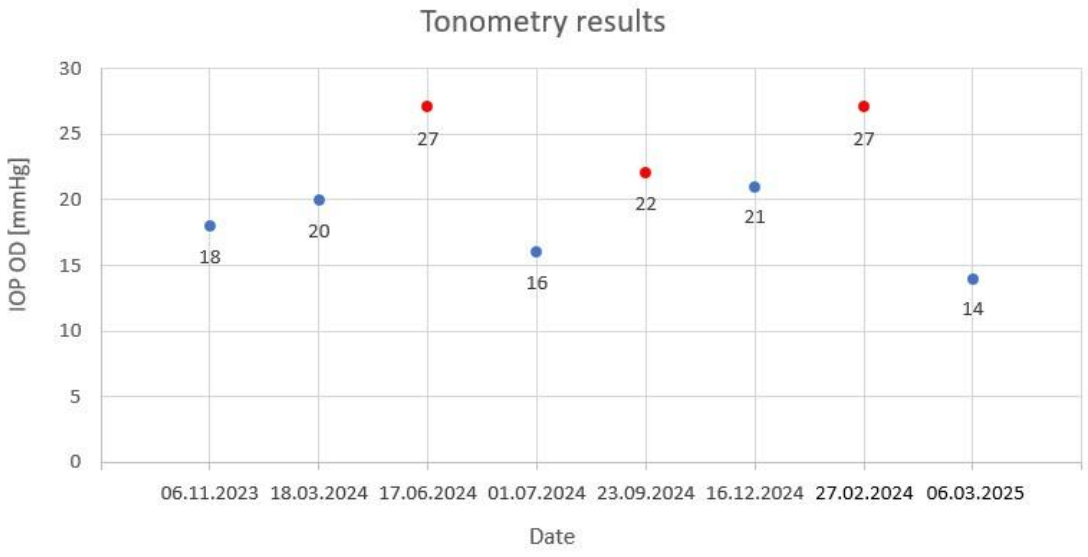

Fig. 3. Graph illustrating the dynamics of intraocular pressure (IOP) changes in the right eye (OD) during follow-up examinations after the initial laser peripheral iridotomy in the OD

Additional examinations revealed a closed anterior chamber angle in the OD, with a partially visible trabecular meshwork and a narrow (10-degree) angle in the OS upon gonioscopic evaluation. Eventually, the decision was made to perform a repeat LPI in the OD at a different iris location. The procedure was completed without any complications. At the post-operative follow-up 5 days later, BCVA measured 1.0 in both eyes and the IOP was 21 mmHg in the OD and 15 mmHg in the OS. The patient was prescribed a fixed combination of dorzolamide and timolol and advised to return for further follow-up.

At the 3-month follow-up visit, the patient demonstrated a recurrent elevation of IOP in the OD of 27 mmHg. Treatment using brimonidine was initiated, which resulted in a reduction of IOP to 14 mmHg at the 7-day follow-up. Automated static perimetry with a Humphrey Field Analyzer revealed a normal visual field with subtle peripheral defects in the pattern deviation in the OD (Figure 4).

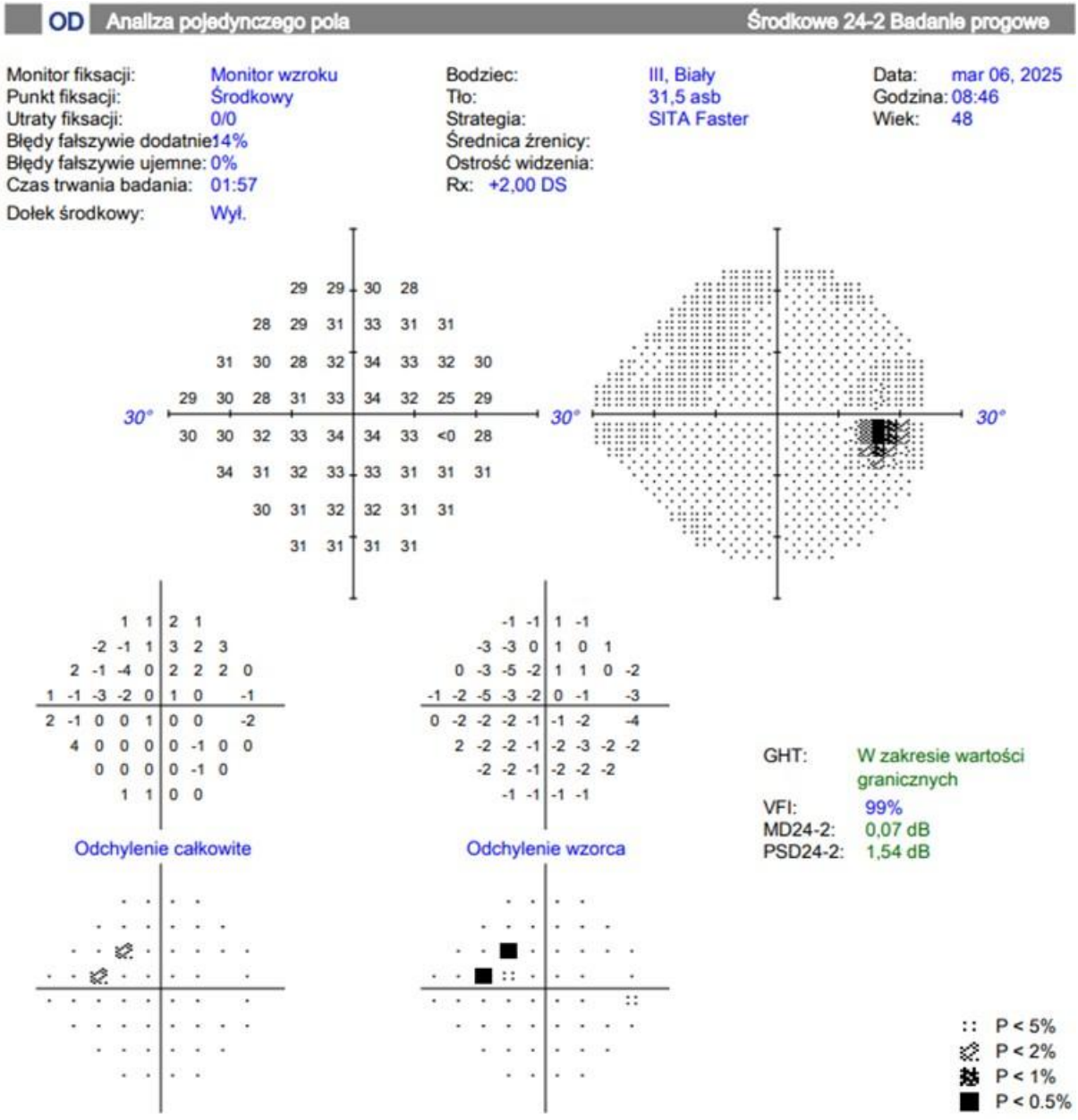

Fig. 4. Automated static perimetry of the right eye (OD) with a Humphrey Field Analyzer

DISCUSSION

As part of the diagnostic protocol, the patient underwent the following examinations: gonioscopy, AS-OCT using a CASIA2 device, anterior segment biomicroscopy, visual acuity assessment, fundus examination, tonometry, and automated static perimetry using an Octopus perimeter and a Humphrey Field Analyzer. Further diagnostic options include dynamic gonioscopy, which can differentiate between appositional and synechial iridocorneal adhesions by

applying pressure to the cornea [7]. When pupillary block is the predominant mechanism of angle closure, the iris becomes peripherally concave. In the case of iris plateau, a double hump sign may be observed. In the lens-related mechanism, indentation does not alter the convex shape of the iris [2].

The presence of peripheral anterior synechiae (PAS) may affect the efficacy of LPI. A prospective study by Lin et al. [8] examined 66 eyes of 42 patients (31 eyes with PAS) and found an inverse correlation between the change in angle width after LPI and the PAS extent at baseline.

Another diagnostic tool is ultrasound biomicroscopy, where a high-frequency ultrasound probe (30–75 MHz) provides detailed imaging of the anterior eye segment, enabling a precise determination of the angle closure mechanism. Eye fundus examination and automated static perimetry can also be supplemented with quantitative measurements of the retinal nerve fiber layer using scanning laser polarimetry (GDx) and OCT techniques.

In primary angle-closure suspect (PACS), the first-line prophylactic treatment is LPI. PACG and acute angle closure due to a pupillary block or iris plateau mechanism are also therapeutic indications [2]. Bleeding at the site of iridotomy is the main intraoperative complication. Postoperative complications include transient IOP elevation, visual disturbances (such as glare, blurring, ghost images, halos, and crescents), transient inflammation (in some cases, leading to posterior synechiae), as well as cystoid macular edema and aqueous misdirection [2].

The effectiveness of LPI in PACS cases varies. In a PACG review, Wright et al. [9] reported lower disease progression rates, from 19%–35% to 0%–16% in early-treated patients. The authors also noted a 75% increase in the width of the iridocorneal angle in these patients after LPI.

In a randomized controlled trial, He et al. [10] treated 889 patients at high risk for angle closure in one randomly selected eye. Over a 6-year follow-up, the incidence of PACG was 4.19 per 1,000 eye-years in eyes treated with LPI versus 7.97 per 1,000 eye-years in untreated eyes. However, the primary outcome occurred in only 19 treated eyes and 36 untreated eyes, raising questions about the broad application of this procedure in the PACS population.

A retrospective analysis of the Zhongshan Angle Closure Prevention (ZAP) trial by Bao et al. [11] suggests that the risk of disease progression in PACS eyes after LPI is associated with persistent angle narrowing detected by AS-OCT or cumulative gonioscopy scores. The authors suggest that these diagnostic tools may potentially help identify patients at high risk of disease progression despite undergoing the procedure.

Radhakrishnan et al. [12] also conducted a retrospective study to evaluate the effectiveness of LPI in PAC patients. They noted that persistent angle closure after the procedure occurred in 2%–57% of patients, depending on the stage of the disease. Risk factors included a narrower angle and non-pupillary block mechanism. Progression to PACG occurred at rates of 0%–0.3% in PACS and 0%–4% annually in PAC patients, while the need for further treatment in PACS was observed in 0%–8% of eyes.

Considering the difficulties in achieving a therapeutic effect, further treatment strategies should be considered. The AS-OCT findings suggest the involvement of the lens and a shallow anterior chamber in the pathomechanism of angle closure. In patients with phacomorphic glaucoma, phacoemulsification with in-the-bag implantation of an artificial intraocular lens has proven to be an effective method for deepening the anterior chamber and reducing IOP [13,14]. Complications associated with this procedure include endophthalmitis, corneal edema, posterior capsular opacification, uveitis, and uncontrolled elevation of IOP [15].

In a report written by the American Academy of Ophthalmology, Chen et al. [16] analyzed seven prospective and five retrospective studies on the effectiveness of IOP reduction following phacoemulsification. They demonstrated that the procedure resulted in a mean IOP reduction of 30% (6 mmHg) in patients with PACG and 71% (35.4 mmHg) in those with acute angle closure. Moreover, the procedure led to a 58% reduction in the use of antiglaucoma medications in case of PACG.

In a multicenter, randomized controlled trial, Azuara-Blanco et al. [13] compared the effectiveness of phacoemulsification and LPI in patients with PACG and clear lenses. This study demonstrated the effectiveness of phacoemulsification in lowering IOP (mean IOP: 29.5 ± 8.2 mmHg at baseline vs. 16.6 ± 3.5 mmHg after 36-month follow-up). Only 21% of patients who underwent the procedure required ongoing pharmacological treatment and just 1 patient required an additional glaucoma surgery.

Similar conclusions were drawn by Singh et al. [17] in a prospective study assessing the efficacy of phacoemulsification in 40 patients with PACG and a cataract. The mean postoperative reduction in IOP came to 42.2% over a 6-month follow-up period. Effective IOP control was achieved in 95% of patients (with failure being defined as IOP > 21 mmHg or the need for additional glaucoma surgery or an increase in the number of topical antiglaucoma medications). Gonioscopic evaluation of the iridocorneal angle also revealed an improvement of at least one grade in all examined cases.

However, in cases where the therapeutic effect is insufficient or when the procedure is technically challenging due to limited access to posterior chamber structures, an alternative treatment option may involve combining phacoemulsification with central decompressing pars plana vitrectomy [18,19]. Removal of the central part of the vitreous body leads to a deeper anterior chamber and optimal access to the intraocular structures located therein. This procedure allows

phacoemulsification to be performed with a reduced risk of complications [18,20].

Adverse events associated with phacovitrectomy in patients with a shallow anterior chamber include zonular dialysis, Descemet's membrane detachment, corneal epithelial damage, suprachoroidal hemorrhage, iris prolapse, and malignant glaucoma [18,19].

Kamińska et al. [18] described the case of a 70-year-old female patient with cataracts who experienced acute angle-closure in both eyes during pharmacological pupil dilation. LPI was performed, but proved insufficient. During phacoemulsification, IOP rose again. Therefore, due to narrow angles and shallow anterior chambers, the decision was made intra-operatively to perform a partial 23G cortical vitrectomy. This led to a normalized IOP, allowing safe continuation of the procedure. AS-OCT revealed deepening of the anterior chamber and widening of the iridocorneal angles.

Two additional cases described by Dada et al. [21] also support the effectiveness of decompressing phacovitrectomy in the management of phacomorphic glaucoma. In a 60-year-old patient with an IOP of 54 mmHg, a shallow anterior chamber, and closed angles, the procedure resulted in a postoperative IOP reduction to 14 mmHg and a reopening of the angles. In another case involving a 55-year-old female patient with a swollen lens, the surgery led to IOP falling from 46 mmHg to 12 mmHg on the first postoperative day.

A retrospective study by Noh and Kim [19], comparing the efficacy and safety of phacoemulsification and transconjunctival limited pars plana phacovitrectomy in patients with acute angle-closure, demonstrated that phacovitrectomy was associated with fewer postoperative complications and smaller corneal epithelial damage. The study also showed a statistically insignificant difference between the two treatment options in terms of reducing IOP or improving visual acuity.

In the context of clinical data, it is also important to consider the possibility of angle closure resulting from the less common plateau iris configuration, characterized by a flat iris profile [3]. In such cases – when LPI proves ineffective – further therapeutic management may include argon laser peripheral iridoplasty [2]. This procedure consists of remodeling of the iris root by inducing contraction of its stromal tissue peripheral to the area of laser application, thereby retracting the iris from the trabecular meshwork and opening the iridocorneal angle [22]. Complications of this process include mild inflammation, peripheral iris atrophy, transient IOP elevation, corneal endothelial burns, posterior synechiae, and permanent mydriasis [2].

A meta-analysis by Bourdon et al. [23], which consisted of one prospective case-control study and seven retrospective cohort studies evaluated the efficacy of laser iridoplasty in patients with angle--closure. This study demonstrated that the procedure effectively lowers IOP in the short term (approximately 20% reduction in IOP at 1-year follow-up). However, its effectiveness diminishes over time, with only a 1% reduction observed after 6 years. In four of the studies in the review, at least 70% success was achieved in terms of angle opening, most commonly defined as a one-grade increase in Shaffer or Spaeth classification in at least two of the four quadrants.

A meta-analysis by Bayliss et al. [22] that evaluated the effectiveness of laser iridoplasty in the treatment of chronically closed angle also suggests that although the procedure may have a beneficial effect on anterior chamber morphology, it is unlikely to offer significant IOP reduction. These findings call into question the validity of performing this procedure in the patient under discussion.

Another alternative is trabeculectomy, during which a fistula is created to allow aqueous humor to drain into a filtering bleb formed in the subconjunctival space, where it is subsequently absorbed [2]. This procedure is associated with multiple complications, including malignant glaucoma, ocular hypotony, endophthalmitis, and – over the long term – an increased risk of cataract development [2,24].

In a randomized controlled trial, Ye et al. [25] reported that trabeculectomy with mitomycin C performed on 27 eyes resulted in a 48.9% reduction in IOP (16.7 ± 11.7 mmHg) in patients with PACG. At the same time, 78.3% of cases ended with complete surgical success (i.e., IOP = 6–21 mm Hg with a reduction of IOP by ≥20% and discontinued pharmacological treatment) and 87% of cases ended with partial surgical success (i.e., regardless of medication use) at the 2-year follow-up.

A similar study by Tham et al. [26] compared the effectiveness of trabeculectomy with mitomycin C to phacoemulsification in patients with PACG, uncontrolled IOP, and no coexisting cataracts. The first procedure, performed on 24 eyes, resulted in a 36% reduction in IOP over a 2-year follow-up period, while the latter, performed on 26 eyes, led to a 34% decrease. Trabeculectomy was associated with fewer required IOP-reducing medications. However, it carried a higher risk of postoperative complications (46% in the trabeculectomy group vs. 4% in the phacoemulsification group) and 33% of eyes treated with trabeculectomy developed a cataract during follow-up.

CONCLUSIONS

LPI represents an effective preventive measure against the development of PACG. However, it may prove insufficient in patients with predisposing anatomical features, such as a shallow anterior chamber. In such cases, phacoemulsification or central decompressing phacovitrectomy offer the most appropriate therapeutic approach, as they deepen the anterior chamber and reduce IOP.

Patients with a family history of glaucoma constitute a high-risk group for treatment-resistant forms of the disease. Therefore, phacoemulsification or central decompressing phacovitrectomy may represent the only viable option for lowering IOP and preserving visual acuity.

Authors' contribution

Study design – W. Żelazna, J. Sokołowski, S. Sirek, D. Wyględowska-Promieńska

Data collection – W. Żelazna, J. Sokołowski

Manuscript preparation – W. Żelazna, J. Sokołowski

Literature research – W. Żelazna, J. Sokołowski

Final approval of the version to be published – S. Sirek, D. Wyględowska-Promieńska

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