

Pediatric WAGR Patient with Aniridia-associated Glaucoma: A Case Report

Patricia Abigail Lim-Tanjutco, MD¹ and Maria Imelda R. Yap-Veloso, MD^{1,2,3}

¹Rizal Medical Center, Pasig City, Philippines

²Asian Eye Institute, Makati City, Philippines

³Sentro Oftalmologico Jose Rizal, Philippine General Hospital, University of the Philippines Manila, Manila, Philippines

ABSTRACT

WAGR syndrome is a rare congenital disorder, occurring in approximately 1 in 500,000 to 1,000,000 individuals, often presenting with ocular malformations such as aniridia. Glaucoma frequently develops when the iris and angle structures are affected, posing a significant risk of vision loss. We report a one-year and seven-month-old patient who presented with corneal opacity of the left eye. Examination revealed corneal opacity, aniridia, and markedly elevated intraocular pressure of 65 mmHg, while the fellow eye, also with aniridia, was normotensive. The patient underwent immediate combined trabeculectomy-trabeculotomy. Postoperative follow-up and timely management of complications allowed acceptable pressure control over one year, though visual prognosis remained guarded. This case highlights the challenges of managing glaucoma in WAGR syndrome, particularly in resource-limited settings. Medical therapy alone is often insufficient, making surgical intervention essential. Combined trabeculectomy-trabeculotomy proved effective in maintaining pressure control when glaucoma drainage devices were not feasible. Multiple interventions and close monitoring are frequently required due to the risk of scarring and postoperative complications. Our experience emphasizes the need for a multidisciplinary ophthalmology approach to optimize outcomes. Despite pressure control, visual outcomes often remain poor due to structural anomalies and the challenges inherent to pediatric patients with this rare syndrome.

Keywords: WAGR, Wilm tumor, glaucoma, aniridia, trabeculectomy, trabeculotomy

INTRODUCTION

Glaucoma has often been cited as the second leading cause of vision loss for people in all age groups worldwide.¹ As devastating as visual loss is in adults, it is far more tragic when the involved patient is a child. Childhood glaucoma is a rare condition in general ophthalmology. It is defined as any primary or secondary eye disease that leads to optic nerve damage, often with elevated intraocular pressure (IOP).²

The British Infantile and Childhood Glaucoma Eye Study, a pilot national population-based study, identified its incidence in the UK and Ireland. In one year, they found 99 affected children. About half (n=47) had primary glaucoma, while the rest (n=52) had secondary glaucoma. The annual incidence was 5.41 per 100,000 (1/18,500) live births in Great Britain and 3.31 per 100,000 (1/30,200) in Ireland. Individuals of Asian (Pakistani) descent were nine times more susceptible than their Western counterparts.²

A similar US study in a mostly Caucasian and Hispanic population found fewer primary glaucoma cases (19.2%). 4% had primary juvenile glaucoma, 45% had secondary glaucoma, and 31% were glaucoma suspects.¹

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Corresponding author: Patricia Abigail Lim-Tanjutco, MD
Rizal Medical Center
425 Pasig Boulevard, Pasig City, Philippines
Email: patriciaabigaillim@gmail.com

This higher prevalence of childhood glaucoma among Asians was also observed in a seven-year retrospective review from Beijing, China. Among 1,452 childhood glaucoma patients, they accounted for 12.91% of total glaucoma cases. Nearly half (46.07%) had congenital glaucoma.³

Secondary congenital glaucoma is a rare childhood case with structural abnormalities at birth that disrupt normal aqueous flow. These cases are often linked with syndromic diseases, such as Axenfeld-Rieger anomaly, Sturge-Weber Syndrome, and congenital aniridia.⁴ Furthermore, congenital aniridia is linked to several syndromic and genetic abnormalities, the most common of which is WAGR syndrome.⁵

First noted by Miller and colleagues in a chart review published in 1964, WAGR syndrome represents a constellation of congenital anomalies found to occur together, with each letter of the syndrome name denoting one of its associated findings: W for Wilms Tumor, A for aniridia, G for genitourinary malformation, and R for retardation of mental development.⁶

For the ocular manifestation, aniridia is the partial or complete loss of the iris. It increases the risk of ocular hypertension and glaucoma. In trauma cases, this results from mechanical damage to angle structures. In congenital cases, it stems from angle dysgenesis or hypoplasia.⁷ The incidence of glaucoma in patients with aniridia is notably high, with reports indicating rates of up to 75%.⁸ Male and female are equally affected. There is also often a genetic component, such as PAX6 and/or WT1 deletion or mutation.⁹ Management of aniridia-associated glaucoma is often challenging, with prognosis being guarded.

Medical therapy, including topical and oral anti-glaucoma medications, is used in pediatric patients. However, its use is not as frequent as in adults due to the lack of extensive studies on the efficacy and safety of these drugs in children. There is caution on the part of the ophthalmologist when using these medications on children due to fears of possible systemic side effects.¹⁰ Furthermore, early efforts at medical treatment in these patients were unsuccessful in long-term control in most cases.¹¹ Most pediatric glaucoma cases are best managed surgically. Medical treatment is used initially before surgery. Common procedures include trabeculectomy, goniotomy, valve implantation, and, more recently, microinvasive glaucoma surgery (MIGS). Surgical management has shown acceptable long-term outcomes.^{1,2,12,13} Aniridia and WAGR syndrome are rare conditions. Due to their rarity, research is limited to case reports. These reports come from advanced countries like Canada, Switzerland, and Japan.¹⁴⁻¹⁶ All have favored early surgical management for IOP control. In high-income countries, WAGR syndrome patients benefit from advanced glaucoma interventions such as glaucoma drainage devices and microinvasive glaucoma surgery. However, these options are often unavailable or unaffordable in resource-limited settings. In such contexts, ophthalmologists must rely on conventional procedures like trabeculectomy or combined trabeculectomy-trabeculotomy

to achieve pressure control. This difference underscores the importance of reporting outcomes from developing countries, where management strategies must adapt to available resources.

To our knowledge, there is, as of yet, no case report detailing the management of WAGR syndrome patients in resource-poor countries; thus, we report on the outcome of a case of WAGR syndrome encountered in a specialty tertiary center in the Philippines, a third-world country, detailing our management and outcome.

CASE PRESENTATION

A one year and nine months old male was presented with a concern of left corneal opacity at the outpatient ophthalmology department of the Rizal Medical Center, a specialty government-run tertiary hospital based in Pasig City, Philippines.

The patient was born full term after an uncomplicated pregnancy in a provincial hospital. At birth, a whitish left corneal opacity and bilateral nystagmus were noted, along with ambiguous genitalia. Initial systemic work-up showed the absence of gonadal structures but no renal masses. The patient was referred for ophthalmologic evaluation.

At two months, a pediatric ophthalmologist confirmed bilateral aniridia with a left eye cataract and suspected WAGR syndrome. Financial constraints limited further evaluation. Intermittent follow-up and pandemic restrictions delayed regular monitoring, during which the left corneal opacity progressed.

At the age of one year and seven months old, the patient developed hematuria, abdominal enlargement, and a palpable right mass in the flank. The patient was then brought to the pediatrics department of our institution, wherein an abdominal CT scan revealed a right abdominal renal mass, suspected to be a Wilms tumor (Figure 1). Physical examination also noted ambiguous genitalia (Figure 2). Hematuria was ruled as secondary to a urinary tract infection. The patient was also evaluated and diagnosed with global developmental delay by a developmental pediatrician. Genetic testing was done by sending a blood sample to a biotechnology facility (Invitae Corporation, San Francisco, California). Genomic DNA extracted from the submitted blood sample underwent targeted region enrichment using a hybridization-based approach and was sequenced with Illumina technology. This revealed the deletion (Entire coding sequence) of the WT1 gene. The deletion of the entire coding sequence of the WT1 gene is key in WAGR syndrome, which leads to aniridia, ambiguous genitalia, developmental delays, and an increased risk of Wilms tumor. WT1 is essential for the development of the kidneys, gonads, and eyes. Its deletion disrupts these processes, contributing to the patient's symptoms and highlighting the need for comprehensive care.

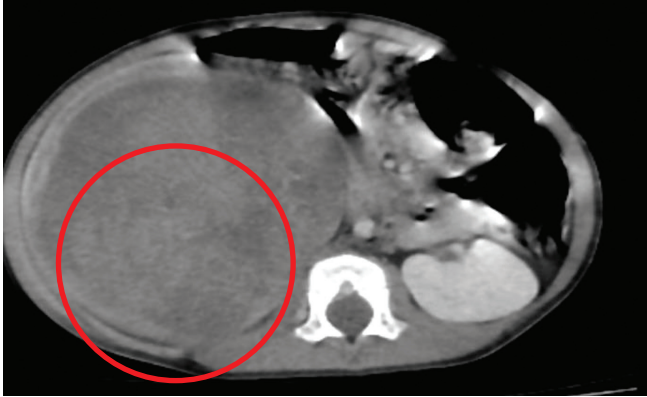


Figure 1. *W* in WAGR stands for Wilms Tumor. The CT scan shows an 11.5 x 10.2 x 10.9 cm well-defined heterogeneously enhancing mass with cystic/necrotic areas of attenuation and no calcifications, arising from the upper half of the right kidney (red circle). CT scan impression of: Right renal mass; consider Wilms Tumor on CT scan was made.

Diagnostic Challenges

The diagnosis of WAGR syndrome in this patient was complicated by several resource-related limitations. Access to genetic testing and specialized imaging was delayed due to financial constraints and limited local availability. The COVID-19 lockdown further hindered continuity of follow-up and routine surveillance, resulting in late tumor detection and progression of ocular findings. These challenges highlight the realities of diagnosing complex genetic

syndromes in resource-poor settings, where comprehensive testing and multidisciplinary coordination are often difficult to obtain.

Once stabilized, the patient was referred to our department for co-management due to eye findings.

On initial presentation at our department, the patient was noted to have photophobia. Gross visual acuity showed a right eye that was able to fixate and follow, and a left eye with a positive dazzle response to light. Extraocular movements were noted to be full and equal, but the patient was noted to have a constant horizontal right-beating nystagmus when at rest.

On examination under anesthesia, IOP taken via iCare handheld tonometer (Icare Finland Oy, Vantaa, Finland) was normal for the right eye at 12mmHg, but was elevated for the left eye at 64mmHg. Gross ocular examination showed non-hyperemic conjunctivae in both eyes. Both eyes had a horizontal and vertical diameter of 10.5 mm. The right eye was noted with near total aniridia, a deep chamber, and a clear lens (Figure 3). The left eye was noted with a 6 x 7 mm central corneal opacity, which did not uptake fluorescein dye. There was noted iridocorneal contact inferiorly of the left eye when examined using a handheld slit lamp (Figure 4). Indirect ophthalmoscopy of the right eye revealed the absence of the foveal contour and foveal pigments. The optic disc, the retinal vessels, and the rest of the retina appeared normal. There was no view of the retina available for the left eye due to the large central corneal opacity.

Intraocular pressure-lowering topical medications (Timolol and Dorzolamide eye drops) were started for the left eye after the patient was cleared by pediatrics of asthma,

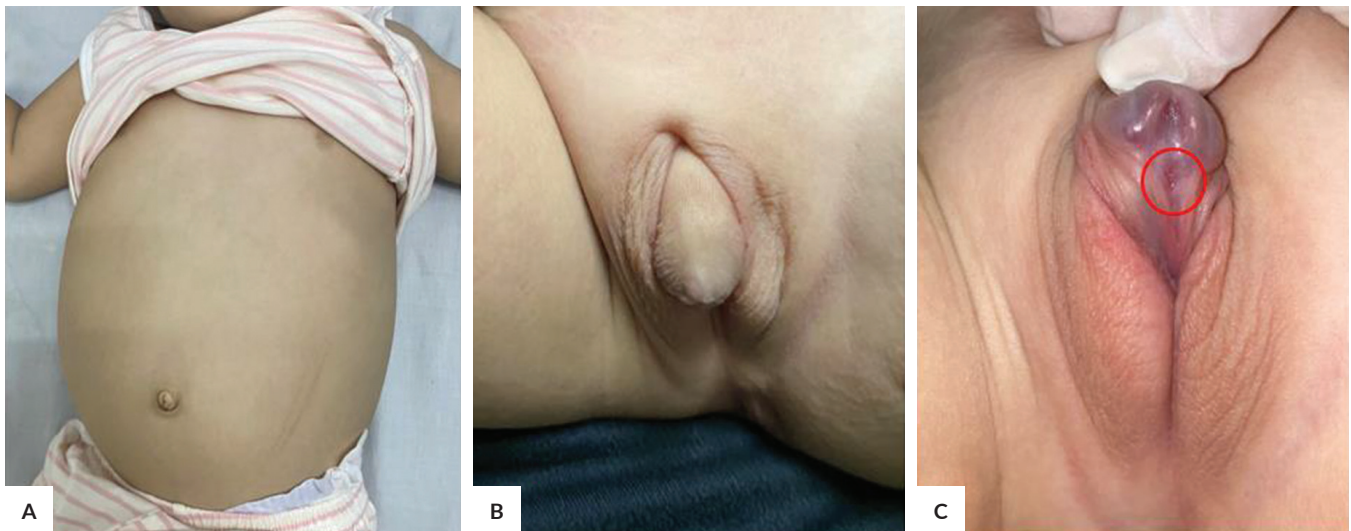


Figure 2. *G* in WAGR stands for Genitourinary malformation. External photographs of the patient taken on initial presentation at our institution at one year and seven months of age. The patient presented with abdominal enlargement, later noted to be secondary to a suspected Wilms tumor (A). Ambiguous genitalia are also noted (B). On further inspection of the proto-penis (C), the patient was also noted to have a urethral malformation opening at the underside of the organ, termed hypospadias (red circle).

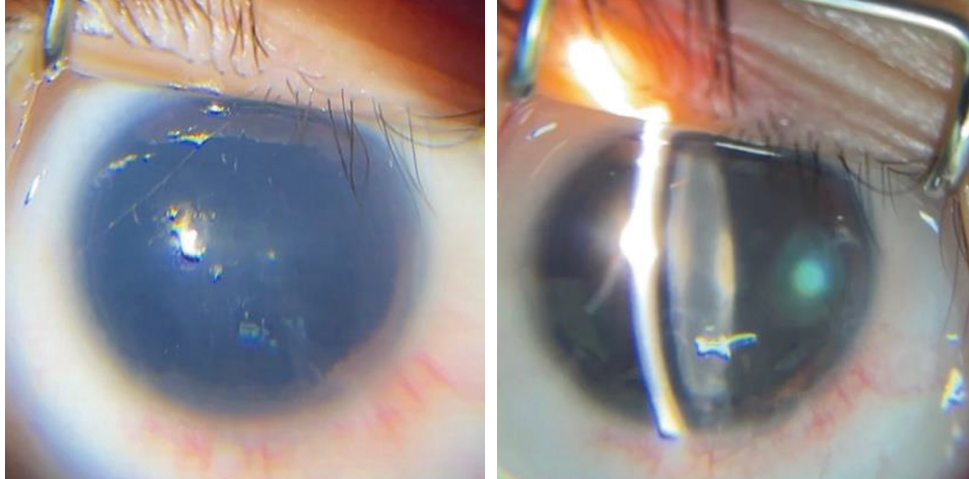


Figure 3. *A in WAGR stands for Aniridia.* Right eye. Pupil was 9 mm, nonreactive to light. The cornea measured 10 x 10 mm. The patient has aniridia. Anterior chamber was deep, and the lens was clear of any opacities.

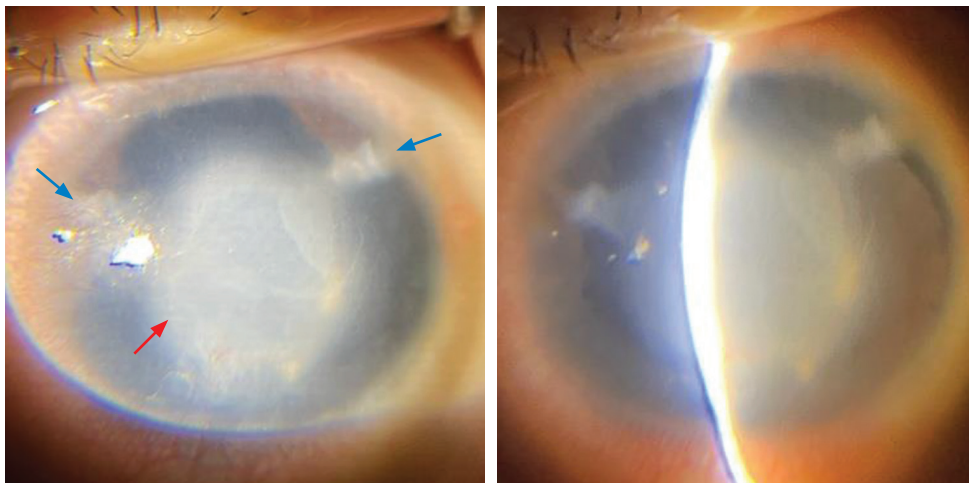


Figure 4. *The left eye external findings during examination under general anesthesia.* There is a large central corneal opacity measuring 6 x 7 mm (red arrow). Two additional corneal opacities are noted superiorly at the 10 o'clock and at the 2 o'clock position, assumed to be from the incisions made during cataract surgery performed earlier in life (blue arrows). No dye uptake was noted on the instillation of fluorescein. Corneal diameter measured 10 x 10 mm. IOP was elevated at 65 mmHg.

heart block, and other contraindications to these topical medications. The patient was also immediately scheduled for examination under anesthesia of both eyes and combined trabeculectomy-trabeculotomy* of the left eye to address the significantly increased IOP.

The trabeculectomy presented difficulties due to the pediatric nature of the case. In children, the scleral tissues

are thinner and more elastic, making the creation of a stable scleral flap technically challenging. A larger, well-delineated scleral flap was fashioned using a crescent blade to allow for better control and avoid excessive leakage postoperatively. Careful attention was given to the placement of sutures, which were adjusted for the pediatric tissue elasticity to prevent over-filtration while ensuring adequate aqueous drainage.

The trabeculotomy also posed significant challenges due to the patient's pronounced corneal opacity, which obscured visualization of the anterior chamber angle structures. To overcome this, the surgeon relied on meticulous tactile feedback and anatomical landmarks. A trabeculotome was used to gently probe the trabecular meshwork and perform a controlled trabeculotomy, ensuring minimal damage to surrounding tissues despite the limited visibility.

*Trabeculectomy is a pressure-lowering procedure that creates a controlled drainage pathway by forming a small scleral flap that allows aqueous humor to bypass the trabecular meshwork and drain into a subconjunctival space. Trabeculotomy, on the other hand, is a procedure that involves incising the trabecular meshwork to open the eye's natural drainage pathway, improving aqueous humor outflow, and reducing intraocular pressure.

Management followed general pediatric glaucoma principles, wherein surgery is indicated when medical therapy fails or when IOP is markedly elevated. Although glaucoma drainage devices are often recommended as the preferred surgical option, these were not available in our setting. A combined trabeculectomy-trabeculotomy was therefore performed as a feasible alternative to achieve adequate pressure control.

During the six-month follow-up period, the patient's mother reported a noticeable reduction in ocular irritation, which she interpreted as an indication of the absence of eye pain compared to the preoperative state. Clinical evaluation revealed no progression of buphthalmos, suggesting adequate IOP control. Postoperatively, IOP measurements using the iCare handheld tonometer (Icare Finland Oy, Vantaa, Finland) ranged between 10 and 15 mmHg. The patient remained asymptomatic, with no signs of ocular discomfort or further globe enlargement. Patient was maintained on Timolol eye drops twice daily, and Dorzolamide eye drops thrice daily to the left eye. A repeat examination under anesthesia was deemed necessary to fully assess IOP and glaucoma status; however, this was deferred as management of the patient's oncologic condition was prioritized during this period. Given the poor overall visual prognosis, financial constraints, and challenges in long-term follow-up, it was determined that a corneal transplant could be deferred at this time. No adverse or unanticipated events were reported during the postoperative and follow-up periods.

The patient's mother shared that the process was difficult due to limited resources and her child's concurrent cancer treatment. She expressed gratitude for the coordinated care and remained hopeful for her child's comfort and vision.

DISCUSSION

The treatment of glaucoma associated with aniridia is challenging. This is because there is often extensive malformation of the various structures needed for aqueous outflow. Histopathologic studies of aniridia patients have found multiple irregularities aside from ciliary body hypoplasia. Specifically, aniridia patients may also have any combination of various anterior chamber angle deformities and attenuation of Bowman's membrane.^{7**} In aniridia, attenuation or absence of Bowman's membrane can lead to progressive corneal clouding and scarring. This further complicates glaucoma management by impairing visualization of the anterior chamber angle and making surgical interventions more technically difficult.

Furthermore, even in aniridia patients who do not present with congenital glaucoma, glaucoma may still develop

later in life from either direct scarring of the overly exposed angle structures or via blockage from adhesions of the malformed iris remnants.¹¹ Investigations of PAX6 knockout mice revealed that the gene deletion, which is associated with aniridia, also leads to predisposition towards trabecular meshwork and Schlemm's canal malformations.¹⁷ PAX6 is a critical gene for eye development, and its knockout results in aniridia, a condition that disrupts the normal formation of various ocular structures, including those involved in aqueous humor drainage. As a result, malformations in the trabecular meshwork and Schlemm's canal can occur, further impairing aqueous outflow and contributing to glaucoma. The incidence of glaucoma in aniridia patients is reported to be as high as 75%.⁸

Early attempts at medical treatment in these patients showed failure to manage glaucoma in the long term in more than half.¹¹ Attempts at management with argon laser trabeculoplasty and via diode laser have, likewise, been unsuccessful.^{18,19} Surgical management is then advocated as first-line treatment in aniridia patients with glaucoma.^{2,20,21}

Multiple surgical interventions have been employed in managing aniridia-associated glaucoma, including goniotomy, trabeculotomy, trabeculectomy, and glaucoma drainage device (GDD) implantation. In our patient, a combined trabeculectomy-trabeculotomy was performed.

Though goniotomy^{***} is a less invasive procedure, when performed as a stand-alone surgery, it is only useful when performed as prophylactic treatment in aniridia patients without pressure elevation. However, in aniridia patients with already elevated IOP, goniotomy alone has a reported dismal long-term success rate of 0-20%.²¹ This is likely due to the structural abnormalities of the trabecular meshwork and Schlemm's canal caused by the underlying genetic defects in aniridia, making the procedure less effective. Additionally, goniotomy requires a clear cornea to allow adequate visualization of the anterior chamber angle, which was not feasible in our patient due to significant corneal opacity. Trabeculectomy, on the other hand, has been reported to have a success rate of 83% in a 10-year follow-up study of aniridia patients, leading the authors to advocate for it as the initial management of aniridia-associated glaucoma.²² Successful short pressure control in our patient following a combined trabeculectomy-trabeculotomy supports this recommendation.

Glaucoma drainage device implantation has also been explored as a treatment option for aniridia-associated glaucoma, demonstrating a success rate of 83.3% at one-month follow-up and 66.6% at one year.²³ Given these favorable outcomes, GDD implantation should be considered as an initial surgical treatment for aniridic glaucoma.

** Bowman's membrane is a thin, transparent layer of the cornea located between the epithelial layer and the underlying stroma. It provides structural support and helps maintain corneal integrity.

*** Goniotomy involves making an incision in the trabecular meshwork to improve aqueous outflow and lower intraocular pressure.

However, clinicians must remain vigilant for concomitant ocular disorders that may complicate management. Despite these promising results, there remains a lack of large-scale studies directly comparing GDD implantation to combined trabeculectomy-trabeculotomy in patients with aniridic glaucoma. Retrospective studies on combined trabeculectomy-trabeculotomy have shown a probability of complete success for IOP ≤ 21 mmHg of 79.9% at one year, supporting its continued viability as a treatment option.²⁴

Notably, prior studies have reported that a mean of 2.8 surgical procedures is required for successful IOP control in aniridic glaucoma patients.²⁵ For our patient, the mother was advised of the potential need for repeat surgical interventions, such as needling with mitomycin C injection. However, they were subsequently lost to follow-up, underscoring the importance of regular monitoring and timely surgical interventions to achieve optimal long-term outcomes.

When dealing specifically with WAGR syndrome patients, however, reports remain limited. It is important to note that these patients often have more extensive aniridia, as well as other possible ophthalmologic malformations, such as corneal opacities, optic disc hypoplasia, and foveal hypoplasia, two of which are present in our patient.^{26,27} These changes lead to a lower mean visual acuity of 20/500 in the better-seeing eye of WAGR syndrome patients compared to 20/125 for patients with non-syndromic aniridia.²⁸

Though our surgical plan of combined trabeculectomy-trabeculotomy has managed to provide satisfactory pressure control in a WAGR syndrome patient with aniridia-associated glaucoma within a six-month period, other surgical techniques have also been reported. A team in Japan have successfully performed bilateral Baerveldt tube transplant in a 15-month old WAGR syndrome patient, with note of pressure stabilization at two to three months postoperatively and a subsequent medication-free pressure control over a two-year period.¹⁶ The successful use of a MIGS device, the XEN45 gel stent (Allergan, Dublin, Ireland), has also been reported by a Canadian team with successful medication-free control of IOP, albeit in an older six-year-old patient who only had persistent mild pressure elevation of 24 mmHg.¹⁴ Despite successful use of device-assisted surgical procedures in WAGR aniridia patients with glaucoma, the successful pressure control in our case proves the use of trabeculectomy and trabeculotomy as a viable option for these patients, which is vital in poorer countries, such as our own.

In both of these cases, however, as with ours, surgical success was defined as adequate pressure control, with good end visual acuity not being part of the necessary goals. This is because, as mentioned earlier, WAGR syndrome patients are prone to a host of other ocular structure abnormalities. The Japanese team, for example, noted macular hypoplasia in both eyes of their patient, while the Canadian team noted corneal scarring in theirs. Our patient, unfortunately, has both. As such, despite successful control of IOP, visual prognosis remains guarded.

Beyond the guarded visual outcome, the overall prognosis in WAGR syndrome depends on the timely management of systemic manifestations. In this case, the patient was co-managed by the oncology department and underwent surgery, chemotherapy, and radiotherapy for a Wilms tumor, with subsequent remission. While oncologic treatment markedly improves survival, developmental delay and genitourinary anomalies may still contribute to long-term morbidity. A holistic, multidisciplinary approach remains essential to optimize overall outcomes.

Furthermore, close follow-up is still deemed necessary due to the increased risk of scarring in this patient, as they are pediatric, have aniridia, and have secondary glaucoma. In pediatric patients, the eye's tissues are more prone to scarring due to their developmental stage, while aniridia contributes to structural malformations, particularly in the trabecular meshwork and cornea, which can impair healing. Additionally, secondary glaucoma increases the likelihood of post-surgical complications, including scarring. Therefore, continuous monitoring is essential. The long-term outcomes for these patients remain undocumented at this point in the literature.

Aside from the possible ocular complications, the possibility of mental retardation may complicate the proper assessment of visual acuity in WAGR syndrome patients.²⁸ All this emphasizes the need for a multidisciplinary and even multi-subspecialty approach in these patients to ensure optimum health.

CONCLUSION

WAGR Syndrome patients may present with several challenges for ophthalmologists, as various ocular structures, such as the cornea, angle, iris, optic nerve, and retina, may be malformed. Angle malformations, whether congenital or acquired, lead to a high incidence of glaucoma in these patients. WAGR syndrome patients with glaucoma require a surgical approach, as medical therapy alone has shown poor long-term efficacy in controlling IOP.

We presented a WAGR syndrome patient with aniridia and glaucoma, managed via combined trabeculectomy-trabeculotomy at one year and eight months of age, with successful pressure control over a six-month period. This highlights the potential efficacy of surgical management for these patients. Glaucoma drainage devices (GDDs) should always be considered a primary or adjunct surgical option, given their documented efficacy in achieving long-term pressure control in pediatric cases. However, when these devices are unavailable, non-device-assisted glaucoma surgical procedures, such as trabeculectomy, remain a viable option, as seen in our case.

Regardless of the surgical intervention, regular follow-up is crucial due to the aggressive scarring often seen in these cases. Other comorbid eye malformations may still lead to unsatisfactory visual outcomes, emphasizing the need

for both multi-subspecialty ophthalmologic care and close monitoring for these individuals.

Ethical Considerations

Written informed consent was obtained from the patient's parents for publication of this case report and accompanying images.

Statement of Authorship

Both authors certified fulfillment of ICMJE authorship criteria.

Author Disclosure

Both authors declared no conflicts of interest.

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REFERENCES

- Fung DS, Roensch MA, Kooner KS, Cavanagh HD, Whitson JT. Epidemiology and characteristics of childhood glaucoma: results from the Dallas Glaucoma Registry. *Clin Ophthalmol*. 2013;7:1739-46. doi: 10.2147/opth.s45480 PMID: 24039394; PMCID: PMC3770714
- Papadopoulos M, Cable N, Rahi J, Khaw PT; BIG Eye Study Investigators. The British Infantile and Childhood Glaucoma (BIG) Eye Study. *Invest Ophthalmol Vis Sci*. 2007 Sep;48(9):4100-6. doi: 10.1167/iovs.06-1350. PMID: 17724193.
- Qiao CY, Wang LH, Tang X, Wang T, Yang DY, Wang NL. Epidemiology of hospitalized pediatric glaucoma patients in Beijing Tongren Hospital. *Chin Med J (Engl)*. 2009 May;122(10):1162-6. PMID: 19493464.
- Karaconji T, Zagora S, Grigg JR. Approach to childhood glaucoma: A review. *Clin Exp Ophthalmol*. 2022;50(2):232-246. doi: 10.1111/ceo.14039 PMID: 35023613.
- Tripathy K, Salini B. Aniridia. In: *StatPearls*. Treasure Island (FL): StatPearls Publishing; August 25, 2023. PMID: 30844160.
- Miller RW, Fraumeni JF Jr, Manning MD. Association of Wilms's tumor with aniridia, hemihypertrophy and other congenital malformations. *N Engl J Med*. 1964 Apr;270:922-7. doi: 10.1056/NEJM196404302701802. PMID: 14114111.
- Margo CE. Congenital aniridia: a histopathologic study of the anterior segment in children. *J Pediatr Ophthalmol Strabismus*. 1983 Sep-Oct; 20(5):192-8. doi: 10.3928/0191-3913-19830901-06. PMID: 6631651.
- Nelson LB, Spaeth GL, Nowinski TS, Margo CE, Jackson L. Aniridia. A review. *Surv Ophthalmol*. 1984 May-Jun;28(6):621-642. doi: 10.1016/0039-6257(84)90184-x PMID: 6330922.
- Moosajee M, Hingorani M, Moore AT. PAX6-Related Aniridia. In: Adam MP, Feldman J, Mirzaa GM, Pagon RA, Wallace SE, Amemiya A, eds. *GeneReviews*®. Seattle (WA): University of Washington, Seattle; May 20, 2003. PMID: 20301534.
- Samant M, Medsinge A, Nischal KK. Pediatric Glaucoma: Pharmacotherapeutic Options. *Paediatr Drugs*. 2016 Jun;18(3): 209-19. doi: 10.1007/s40272-016-0174-4. PMID: 27093864.
- Grant WM, Walton DS. Progressive changes in the angle in congenital aniridia, with development of glaucoma. *Trans Am Ophthalmol Soc*. 1974;72:207-28. PMID: 4462243.
- Ikeda H, Ishigooka H, Muto T, Tanihara H, Nagata M. Long-term outcome of trabeculectomy for the treatment of developmental glaucoma. *Arch Ophthalmol*. 2004 Aug;122(8):1122-8. doi: 10.1001/archophth.122.8.1122. PMID: 15302651.
- Beck AD, Freedman S, Kammer J, Jin J. Aqueous shunt devices compared with trabeculectomy with Mitomycin-C for children in the first two years of life. *Am J Ophthalmol*. 2003 Dec;136(6):994-1000. doi: 10.1016/s0002-9394(03)00714-1. PMID: 14644208.
- Rawlyk B, Thatcher MD, Rubab S, Campos-Baniak MG. Transconjunctival XEN45 implantation for secondary open-angle glaucoma management in a pediatric patient with WAGR syndrome. *Am J Ophthalmol Case Rep*. 2023 Jul;32:101888. doi: 10.1016/j.ajoc.2023.101888. PMID: 37533700; PMCID: PMC10393533.
- von Weissenfluh C, Gerber-Hollbach N, Früh B, August P, Goldblum D. A case of WAGR syndrome with Peters' anomaly. *Klin Monbl Augenheilkd*. 2015 Apr;232(4):382-3. doi: 10.1055/s-0034-1396329. PMID: 25902081.
- Akagi T, Yoshikawa M, Nakanishi H, Yoshimura N. A case of WAGR syndrome in association with developmental glaucoma requiring bilateral Baerveldt glaucoma implants and subsequent tube repositioning. *Clin Ophthalmol*. 2015 Jun;9:1081-4. doi: 10.2147/OPTH.S80444. PMID: 26109842; PMCID: PMC4474385.
- Baulmann DC, Ohlmann A, Flügel-Koch C, Goswami S, Cvekl A, Tamm ER. Pax6 heterozygous eyes show defects in chamber angle differentiation that are associated with a wide spectrum of other anterior eye segment abnormalities. *Mech Dev*. 2002 Oct;118(1-2): 3-17. doi: 10.1016/s0925-4773(02)00260-5 PMID: 12351165.
- Wiggins RE Jr, Tomey KF. The results of glaucoma surgery in aniridia. *Arch Ophthalmol*. 1992 Apr;110(4):503-5. doi: 10.1001/archophth.1992.01080160081036. PMID: 1562257.
- Plager DA, Neely DE. Intermediate-term results of endoscopic diode laser cyclophotocoagulation for pediatric glaucoma. *J AAPOS*. 1999 Jun;3(3):131-7. doi: 10.1016/s1091-8531(99)70057-1. PMID: 10428585.
- Karaconji T, Zagora S, Grigg JR. Approach to childhood glaucoma: a review. *Clin Exp Ophthalmol*. 2022 Mar;50(2):232-46. doi: 10.1111/ceo.14039. PMID: 35023613.
- Lee H, Khan R, O'Keefe M. Aniridia: current pathology and management. *Acta Ophthalmol*. 2008 Nov;86(7):708-15. doi: 10.1111/j.1755-3768.2008.01427.x. PMID: 18937825.
- Adachi M, Dickens CJ, Hetherington J Jr, Hoskins HD, Iwach AG, Wong PC, et al. Clinical experience of trabeculectomy for the surgical treatment of aniridic glaucoma. *Ophthalmology*. 1997 Dec;104(12):2121-5. doi: 10.1016/s0161-6420(97)30041-4. PMID: 9400774.
- Demirok GS, Ekşiöglü Ü, Yakın M, Kaderli A, Kaderli ST, Örnek F. Short- and long-term results of glaucoma valve implantation for aniridia-related glaucoma: a case series and literature review. *Turk J Ophthalmol*. 2019 Sep;49(4):183-7. doi: 10.4274/tjo.galenos.2019.07348. PMID: 31486604; PMCID: PMC6761385.
- Outcomes of Primary Combined Trabeculectomy and Trabeculectomy in Early-Onset Glaucoma in Children with Congenital Aniridia [Internet]. May 2021 [cited 2024 Sep 25]. Available from: [https://www.ophtalmologyglaucoma.org/article/S2589-4196\(20\)30257-X/abstract](https://www.ophtalmologyglaucoma.org/article/S2589-4196(20)30257-X/abstract) PMID: 32966898.
- Wiggins RE Jr, Tomey KF. The results of glaucoma surgery in Aniridia. *Arch Ophthalmol*. 1992 Apr;110(4):503-5. doi: 10.1001/archophth.1992.01080160081036. PMID: 1562257.
- Fischbach BV, Trout KL, Lewis J, Luis CA, Sika M. WAGR syndrome: a clinical review of 54 cases. *Pediatrics*. 2005 Oct;116(4):984-8. doi: 10.1542/peds.2004-0467. PMID: 16199712.
- Duffy KA, Trout KL, Gunckle JM, Krantz SM, Morris J, Kalish JM. Results from the WAGR Syndrome Patient Registry: Characterization of WAGR spectrum and recommendations for care management. *Front Pediatr*. 2021 Dec;9:733018. doi: 10.3389/fped.2021.733018. PMID: 34970513; PMCID: PMC8712693.
- Krause MA, Trout KL, Lauderdale JD, Netland PA. Visual acuity in aniridia and WAGR Syndrome. *Clin Ophthalmol*. 2023 May;17: 1255-61. doi: 10.2147/OPTH.S405003. PMID: 37152637; PMCID: PMC10162095.