

Beyond Sight: Preserving Quality of Life in A Child With Advanced Juvenile Glaucoma and Severe Low Vision

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ABSTRACT

Introduction: Juvenile open-angle glaucoma (JOAG) is a rare but aggressive form of glaucoma that typically manifests in childhood or early adolescence. The condition is often resistant to medical therapy and leads to progressive visual impairment. Early diagnosis and appropriate intervention are important to preserve vision and improve long-term outcomes.

Case description: An 8-year-old girl presented with progressive vision loss in both eyes. She had a history of bilateral congenital cataract surgery and persistently elevated intraocular pressure (IOP) despite maximal topical anti-glaucoma therapy. The patient underwent glaucoma drainage implant (GDI) surgery in the right eye, which produced a significant and sustained reduction in IOP and prevented further visual deterioration in that eye. Despite a poor visual prognosis in the left eye, the patient achieved functional adaptation through low vision rehabilitation.

Discussion: JOAG is frequently associated with a strong genetic component and poses significant treatment challenges because of its aggressive progression. This case illustrates the limitations of pharmacological management and the role of timely surgical intervention. In addition, the integration of low vision rehabilitation and psychosocial support was essential in maintaining the patient's independence and quality of life.

Conclusion: This case shows the clinical challenges of managing JOAG in a pediatric patient with a complex ocular history. Surgical intervention, particularly the use of GDI, can be effective in cases refractory to medical management. Comprehensive care, including low vision support and psychosocial counseling, plays an important role in optimizing outcomes and quality of life in children with advanced glaucoma.

Keywords: Juvenile open-angle glaucoma, Pediatric glaucoma, Glaucoma drainage implant, Intraocular pressure, Low vision rehabilitation.

I. INTRODUCTION

Low vision refers to a visual impairment not corrected by standard eyeglasses, contact lenses, medication, or surgery, which interferes with the ability to perform everyday activities. Visual impairment may result from a variety of ocular and systemic disorders and can present as reduced central vision, reduced peripheral vision, blind spots, loss of contrast sensitivity, or a combination of these symptoms.¹

Ninety percent of the 18.94 million visually impaired children worldwide reside in low- and middle-income countries. Studies indicate that the quality of life (QoL) of individuals with visual impairment (VI) is significantly affected.²

Glaucoma is a neurodegenerative disorder characterized by progressive degeneration of the optic disc and visual field loss, for which intraocular pressure (IOP) is the chief modifiable risk factor. Glaucoma is the most common cause of permanent blindness. It affects more than 2 million people in the United States and 67 million people worldwide. The two primary subtypes are open-angle glaucoma and angle-closure glaucoma. Juvenile open-angle glaucoma (JOAG) is a rare form of primary open-angle glaucoma.³

A subtype of primary open-angle glaucoma detected before the age of 40 is known as juvenile-onset open-angle glaucoma (JOAG). The disorder may occur in different proportions among populations, depending on whether it is hereditary or non-hereditary.⁴ JOAG is characterized by three features: absence of secondary symptoms, open anterior chamber angle, and onset before the age of 40. Myocilin was the first glaucoma gene identified. Mutations causing autosomal dominant disease may account for up to 36% of JOAG cases and 4% of POAG cases.⁵

Because of its rarity and distinct clinical characteristics, juvenile-onset open-angle glaucoma has long been a subject of research. The condition is relatively common in people of South Asian, West Asian, and African origin when compared with European Caucasians.⁴

The prevalence of juvenile open-angle glaucoma is 0.38 per 100,000, with ages ranging between 4 and 20 years.³ Prevalence varies among populations. JOAG accounted for 3.4% of all newly diagnosed glaucomas at a tertiary care centre in Nigeria, compared with 3.3% in India, 1.9% in Saudi Arabia, and 0.7% in Caucasians. The prevalence in the United States was estimated to be 1 in 50,000.⁴

In complex cases of glaucoma, when prior trabeculectomy has failed and medical therapy is ineffective, glaucoma drainage implant procedures have become an increasingly common alternative in recent years. GDIs drain aqueous humour from the anterior chamber to a reservoir that forms externally several weeks after surgery and maintains the flow. These devices have proven effective in cases of corneal scarring following prior ocular surgery and in failed trabeculectomy.⁶ Failure of a glaucoma drainage device was defined as any of the following: IOP outside the target range (5–18 mmHg inclusive) or a reduction of less than 20% from baseline on two consecutive postoperative visits after four weeks; the need for additional glaucoma surgeries in the same eye to control IOP; the need to remove the implant; severe vision loss associated with surgery (endophthalmitis, phthisis bulbi, suprachoroidal haemorrhage); or vision worsening to no light perception (NLP) for any reason other than glaucoma.⁷

This case report is presented to show the efficacy of a glaucoma drainage implant in advanced glaucoma in children, and how quality of life can be preserved despite limited vision.

II. CASE ILLUSTRATION

An 8-year-old girl was admitted to the ophthalmology department with a chief complaint of difficulty seeing objects. When reading a book, she had to hold it close to her eyes, and she frequently bumped into objects while walking. The condition had persisted for more than two years prior to presentation. Her left eye had been able to perceive only light for the past year, with gradual further deterioration.

She had a history of congenital cataract surgery with intraocular lens implantation in both eyes one year earlier and had been receiving topical timolol in both eyes since before the operation. Her current medications were timolol eye drops every 12 hours, brinzolamide eye drops every 8 hours, and latanoprost eye drops every 24 hours, all applied bilaterally. The patient's mother had been diagnosed with glaucoma two years earlier and was on continuous topical glaucoma medication.

Visual acuity was 0.5 LogMAR at 1 metre in the right eye and no light perception in the left eye. Intraocular pressure measured by non-contact tonometry (NCT) was 33 mmHg in the right eye and 45 mmHg in the left eye. Anterior segment evaluation showed two corneoscleral sutures at 11 and 12 o'clock in the right eye and one corneoscleral suture at 11 o'clock in the left eye. The anterior chamber was Van Herick grade IV bilaterally, with an iradier iris and an iridectomy at 11 o'clock in both eyes. Posterior synechiae were present from 12 to 2 o'clock in the right eye. The pupil was irregular in both eyes, and an intraocular lens prosthesis was present bilaterally with a fibrinous membrane and iris pigment deposits. Funduscopic examination revealed a pale optic disc in both eyes, with a cup-to-disc ratio (CDR) of 0.9 in the right eye and 1.0 in the left eye. Colour matching was preserved.

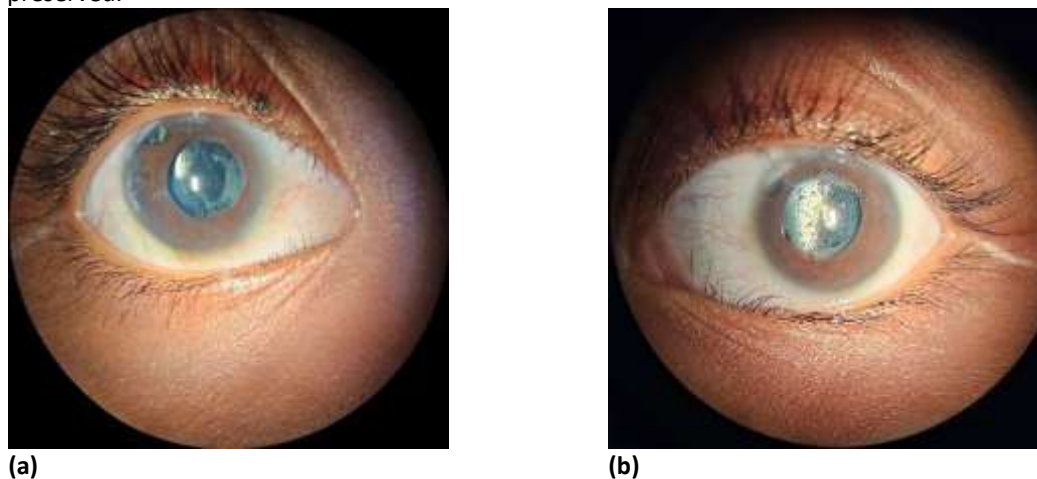


Figure 1. Anterior segment of the right eye (a) and left eye (b) showing a deep anterior chamber, iradier iris, iridectomy at 11 o'clock, and intraocular lens prosthesis with fibrinous membrane and iris pigment. *Pictures taken with the patient's consent. Courtesy: Poli Mata RSUD Dr Soetomo.

At the one-month follow-up after GDI surgery, the patient's right eye IOP had decreased to 13 mmHg by NCT while still on glaucoma medications. Her visual acuity had decreased to 0.7 LogMAR at 30 cm. The GDI tube was observed in the anterior chamber at 10 o'clock. Glaucoma medications were continued for the following month. Two months after GDI surgery, the right eye IOP had decreased to 9 mmHg by NCT, and the patient was advised to discontinue all glaucoma medications in the right eye.

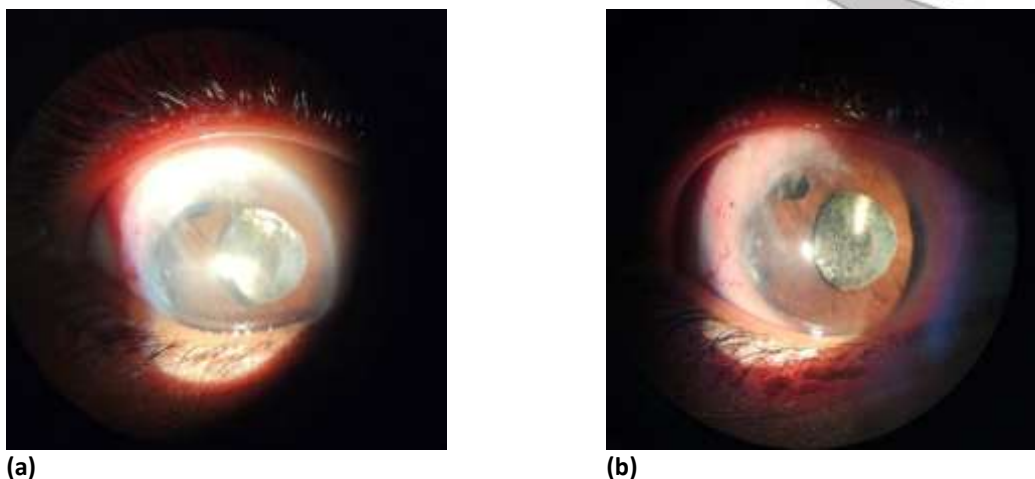


Figure 2. Anterior segment of the right eye at one-month (a) and two-month (b) follow-up after GDI surgery.
*Pictures taken with the patient's consent. Courtesy: Poli Mata RSUD Dr Soetomo.

III. DISCUSSION

Glaucomas affecting children and adolescents, also referred to as pediatric glaucomas, comprise a diverse group of conditions associated with elevated intraocular pressure (IOP). Up to approximately four years of age, these conditions can damage the optic nerve, visual field, cornea, and other structures. These rare glaucomas vary in presentation and etiology.⁵

Pediatric glaucoma is typically classified as primary or secondary. In primary congenital glaucoma (PCG), angle dysgenesis leads to outflow resistance and elevated IOP, which produces the classic features of PCG: enlarged or cloudy corneas, Haab striae, and an enlarged globe (buphthalmos). In juvenile open-angle glaucoma (JOAG), another high-pressure primary glaucoma, an isolated angle abnormality may be present; this form develops later in childhood (generally after age four) or in early adulthood.⁵ Our patient had elevated intraocular pressure diagnosed at seven years of age, before her congenital cataract surgery.

Juvenile-onset open-angle glaucoma (JOAG) has long been a subject of interest because of its rarity and distinct clinical features. It is relatively common in people of South Asian, West Asian, and African descent compared with European Caucasians.⁴ JOAG generally affects individuals during childhood or early adulthood but differs from congenital glaucoma, which presents with buphthalmos, megalocornea, Haab's striae, and ocular or systemic developmental anomalies.⁸ In our patient, there was no ocular enlargement, the angle was open, and the globe appeared normal; however, she had a history of congenital cataract that had been removed with implantation of an intraocular lens prosthesis.

The pathophysiology of glaucoma remains incompletely understood. In patients with juvenile-onset primary open-angle glaucoma, the elevated IOP appears to result from goniodysgenesis, a failure of anterior chamber development that produces thickened trabecular beams and an immature trabecular meshwork.³

In JOAG, symptoms related to visual acuity, including blurred vision and decreased visual acuity, were the major manifestations. However, one-third of patients attended clinics without definite symptoms and were diagnosed with JOAG incidentally.⁸ Our patient presented with limited vision, bumping into objects and difficulty reading. Visual acuity was 0.5 LogMAR at 1 metre in the right eye and no light perception in the left eye. Colour perception was normal.

Recent evidence suggests that JOAG is genetically heterogeneous. Although it was traditionally considered an autosomal dominant disease with variable penetrance and expressivity, autosomal recessive and sporadic occurrences are also common in certain ethnicities.⁴ A family history of glaucoma was found to carry a relative risk of 2.1 times for association with at least possible OAG. However, the relative importance of family history may vary with the closeness of relationship of a patient to an affected family member (first, second, or even third degree).⁹ Our patient had a family history of glaucoma in her mother.

Myocilin was the first identified glaucoma-causing gene, and mutations are known to cause JOAG. The lifetime risk of developing open-angle glaucoma in MYOC mutation carriers is estimated to be 60–100%. The gene was initially cloned as a steroid-response protein from cultured TM cells, named 'trabecular meshwork-induced glucocorticoid response (TIGR) protein'.⁴

One important risk factor for elevated intraocular pressure (IOP) is the use of systemic steroids. With the use of topical or systemic glucocorticoids, the incidence of ocular hypertension may increase to 30–40% of the general population. However, the frequency of ocular hypertension in endogenous hypercortisolemia and the ocular

changes that occur when surgical resection induces endocrine remission remain uncertain.¹⁰ Our patient was diagnosed with Cushing's syndrome, presumably due to steroids taken to treat her allergic reaction.

Other causes of elevated IOP in the juvenile age group need to be excluded before labelling a patient as having JOAG. The differential diagnosis of JOAG includes:

- a. Pigmentary glaucoma. Pigment dispersion syndrome and pigmentary glaucoma can be distinguished by the presence of a Krukenberg spindle, Sampolesi line, excessive angle pigmentation, a very deep anterior chamber, a Scheie stripe or Zentmayer ring on the lens capsule, and sometimes a concave iris configuration confirmed on UBM.
- b. Steroid-induced glaucoma. Patients may present with post-exercise coloured halos or blurred vision due to transient IOP spikes. It may be difficult to distinguish between systemic or topical steroid use and steroid-induced glaucoma unless there is a clear history of steroid use. Suspicion may be raised by a history of over-the-counter medication use, seasonal or chronic red eye, itching, skin disorders, and systemic conditions such as asthma and nephrotic syndrome. Unlike untreated JOAG, which progressively worsens, steroid-induced glaucoma is associated with very high IOP, and in many but not all cases IOP decreases after the drugs are discontinued.
- c. Congenital glaucoma. After the age of three, congenital glaucoma may be mistakenly identified as JOAG if corneal diameters are not carefully examined. Angle dysgenesis and myopia may be present in both diseases.
- d. Uveitic glaucoma. If uveitis is not treated appropriately, a case with few symptoms may be mislabelled as JOAG. Retrolental cells and residual keratic precipitates can provide a clue. In this setting, long-term steroid use may also result in a white eye with high IOP.

Although several risk factors for the development and progression of glaucoma have been identified (IOP, age, genetic predisposition, and vascular characteristics), decreasing IOP remains the only scientifically proven approach for slowing disease progression. The Early Manifest Glaucoma Trial found that for every 1 mmHg decrease in IOP from baseline, the probability of glaucoma progression decreased by 10%.¹¹

The neuropathy of JOAG is usually severe and rapidly progressive compared with adult-onset POAG. IOP is often greater than 30 mmHg and does not respond well to medications. Treatment is challenging. IOP-lowering medications are started first, but filtration or aqueous drainage device surgery is often required. Angle surgery is generally not helpful.¹²

Medical therapy is used as first-line treatment, but many cases are refractory to maximal medical treatment and may require incisional glaucoma surgery.⁵ Timolol, a topical beta-blocker, is typically the first-line treatment and is well tolerated in children. Timolol carries a minimal risk of bradycardia and bronchospasm, which can be reduced by performing punctal occlusion after instillation or by switching to betaxolol, a beta-1 selective antagonist with a smaller effect on the airways than non-selective beta-blockers. Topical carbonic anhydrase inhibitors such as brinzolamide and dorzolamide are useful options with few adverse effects. Latanoprost is a commonly used prostaglandin analogue that can significantly lower IOP in children. However, its non-response rate in children is higher than in adults, possibly because of an aberrant uveoscleral pathway in the forms of glaucoma that predominate in children.¹³ Our patient had been using multiple topical anti-glaucoma medications, including latanoprost, timolol, and brinzolamide, but her IOP remained high.

Early studies of trabeculoplasty in JOAG eyes using argon laser showed a poor response, and worsening occurred in a few eyes with pressure spikes reaching 35–70 mmHg.⁴ Surgical therapy has multiple options, including trabeculectomy, drainage implants, angle procedures (goniotomy, trabeculotomy), and cycloablative procedures. For juvenile open-angle glaucoma, trabeculectomy is the mainstay of surgical therapy and has a good outcome.⁵

Glaucoma drainage implants (GDIs) are an alternative surgical option for JOAG. GDIs are preferred when conjunctival scarring prevents trabeculectomy.³ The basic design of all contemporary GDIs is similar. Through a scleral fistula, a silicone tube is inserted into the anterior chamber (or, in some situations, the vitreous cavity or ciliary sulcus) and directs aqueous humour to an end-plate situated in the equatorial sub-Tenon's space of the globe. This end-plate is usually centred between (and in some cases extends beneath) two adjacent rectus muscles. Because the tissues are easier to access and because there are no oblique recti in the superotemporal quadrant between the superior and lateral recti, GDIs are typically placed there to reduce the risk of postoperative diplopia.¹⁴

GDIs vary in their design with respect to the size, shape, and material of the end-plate. GDIs can be further subdivided into 'valved' or 'non-valved' devices, depending on whether a flow restriction mechanism limits aqueous humour flow. A valved implant is intended to reduce the risk of postoperative hypotony before plate encapsulation while allowing rapid aqueous outflow and lowering intraoperative pressure after surgery. For non-

valved implants, aqueous outflow must be temporarily restricted by internal or external tube ligation until end-plate encapsulation occurs. This manoeuvre lowers the likelihood of postoperative hypotony by allowing resistance to outflow to develop.¹⁴

The Virna Glaucoma Implant (VGI) is an implant made of polymethylmethacrylate (PMMA) with a design intended to facilitate installation in Asian eyes. PMMA is chosen as the plate material because it has been used in various implant operations in humans and is a safe material for intraocular lens implants and eye prostheses.¹⁵ Glaucoma filtration surgery can result in loss of visual acuity through a variety of mechanisms. Recognisable problems such as cataract, hypotony maculopathy, cystoid macular oedema, suprachoroidal or vitreous haemorrhage, retinal detachment, endophthalmitis, and uveitis can cause visual impairment following glaucoma filtration surgery. Risk factors for the wipe-out phenomenon include choroidal effusions (even after resolution), preoperative split fixation on the visual field, the number of split fixation quadrants, undetected excessive intraocular pressure, and severe hypotony following surgery.¹⁶

Our patient has severe low vision with a markedly constricted visual field. Ninety percent of the 18.94 million visually impaired children worldwide reside in low- and middle-income countries. Studies indicate that the quality of life of individuals with visual impairment is significantly affected.¹⁷

Varni et al. created the Pediatric Quality of Life Inventory (PedsQL), a tool used to assess health-related quality of life in children and adolescents with acute and chronic illnesses as well as in healthy populations. PedsQL comprises disease-specific components and a generic core module. This allows the severity of a chronic health condition to be indicated or healthy and sick populations to be differentiated. Consequently, this tool enables comparisons between groups and the evaluation and analysis of a particular case's HRQOL and its position relative to peers.¹⁸

Versions are available for infants (13–24 months), toddlers (ages 2–4), young children (ages 5–7), children (ages 8–12), teenagers (ages 13–18), young adults (18–25), and adults, depending on the age of the sample. The PedsQL measures four dimensions: Physical Functioning, Emotional Functioning, Social Functioning, and School Functioning.¹⁸ For our patient, we used the version for children aged 8–12.

The child report of the PedsQL 4.0 Generic Core Scales consists of 23 items in 4 dimensions. The maximum score on the PedsQL Generic Core Scales is 100, which indicates the highest possible health-related quality of life (HRQOL). Higher scores generally reflect better HRQOL. PedsQL scores are transformed to a 0–100 scale, where 100 represents the best possible HRQOL. Items are reverse-scored and linearly transformed to a 0–100 scale as follows: 0=100, 1=75, 2=50, 3=25, 4=0.¹⁹ Out of the 23 questions, the mean total score for our patient was 67.3.

Table 1. The Child Reports of PedsQL 4.0 Generic Core Scales for Children (Varni, 2023).

In the past ONE month, how much of a problem has this been for you...

About My Health and Activities (problems with...)	Never	Almost Never	Sometimes	Often	Almost Always
It is hard for me to walk more than one block	0	1	2	3	4
It is hard for me to run	0	1	2	3	4
It is hard for me to do sports activity or exercise	0	1	2	3	4
It is hard for me to lift something heavy	0	1	2	3	4
It is hard for me to take a bath or shower by myself	0	1	2	3	4
It is hard for me to do chores around the house	0	1	2	3	4
I hurt or ache	0	1	2	3	4
I have low energy	0	1	2	3	4
About My Feelings (problems with...)	Never	Almost Never	Sometimes	Often	Almost Always
I feel afraid or scared	0	1	2	3	4
I feel sad or blue	0	1	2	3	4
I feel angry	0	1	2	3	4
I have trouble sleeping	0	1	2	3	4
I worry about what will happen to me	0	1	2	3	4
How I Get Along with Others (problems with...)	Never	Almost Never	Sometimes	Often	Almost Always
I have trouble getting along with other kids	0	1	2	3	4
Other kids do not want to be my friend	0	1	2	3	4
Other kids tease me	0	1	2	3	4
I cannot do things that other kids my age can do	0	1	2	3	4

It is hard to keep up when I play with other kids	0	1	2	3	4
About School (problems with...)	Never	Almost Never	Sometimes	Often	Almost Always
It is hard to pay attention in class	0	1	2	3	4
I forget things	0	1	2	3	4
I have trouble keeping up with my schoolwork	0	1	2	3	4
I miss school because of not feeling well	0	1	2	3	4
I miss school to go to the doctor or hospital	0	1	2	3	4

Families are considered the main setting in which children learn, develop, and mature. Our patient attends a primary school for children with special needs, learns to read Braille, and participates actively in social activities with her friends. She has a supportive family to help her cope with her limited vision, although caring for a child with low vision is challenging for most parents. In addition to teaching methods that enhance visual functioning in daily life, low vision rehabilitation aims to maximise the use of residual vision following severe vision loss. Further goals include enhancing psychosocial functioning and helping individuals adjust to irreversible vision loss. These skills encourage self-reliance and social engagement. Low vision rehabilitation should ultimately improve the quality of life of individuals with visual impairment.

Low vision rehabilitation can take various forms, including group programmes and psychological interventions to help individuals cope with irreversible vision loss and enhance their well-being, techniques to improve eyesight such as training in the use of magnifying glasses or other devices, and strategies to make better use of residual vision in daily situations. Multidisciplinary rehabilitation programmes may involve psychiatric therapies, the use of magnifying devices, and services delivered in the patient's home. Other, less common forms of rehabilitation services include home safety programmes and balance training.²⁰

Treatment of sensory input impairments should start as early as possible in a supportive emotional environment that improves the child's motivation and interaction with caregivers. Early intervention is required for visually impaired children.²

IV. CONCLUSION

Juvenile-onset open-angle glaucoma (JOAG) is a rare but serious condition characterised by elevated intraocular pressure that can lead to progressive and irreversible vision loss, especially in pediatric populations. The condition presents unique diagnostic and management challenges, particularly when associated with other ocular anomalies such as congenital cataracts and steroid-induced complications, as observed in this case.

In this 8-year-old patient, despite the use of maximal medical therapy, intraocular pressure remained uncontrolled, requiring surgical intervention with a glaucoma drainage implant (GDI). The surgical outcome showed a significant reduction in IOP and allowed discontinuation of glaucoma medications, preventing further optic nerve damage and preserving the remaining vision.

The case highlights the importance of early diagnosis, genetic screening when indicated, and a multidisciplinary approach to managing pediatric glaucoma. Low vision rehabilitation plays an important role in improving the quality of life for children with irreversible visual impairment. Supportive family involvement, specialised education, and adaptive skills training are essential to help such children lead fulfilling, socially engaged lives. Ultimately, prompt intervention, continued follow-up, and comprehensive support systems are crucial to preserving visual function and enhancing the quality of life in children affected by JOAG.

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