

# Bilateral Glaucoma in An Infant with Phacomatosis Pigmentovascularis and Sturge-Weber Syndrome: A Rare Case Report



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## ABSTRACT

Prevalence of concurrent Phacomatosis Pigmentovascularis (PPV) and Sturge-Weber syndrome (SWS) is unknown due to its rarity and sporadic occurrence. In this case report, we present a rare case of bilateral glaucoma in an infant with PPV and SWS. A three-month old male infant presented with hazy cornea and visible black spots in the upper sclera and presented with intraocular pressure (IOP) of 40 and 52 mmHg in the left and right eye, respectively. Trabeculectomy-Trabeculotomy for both eyes was done which initially decreased IOP. However, high IOP recurred, and the patient was treated with topical medication. At one year follow-up, the patient had normal IOP and was responsive to light and object. Our case is a rare presentation of concurrent SWS, PPV, and bilateral glaucoma. The potential systemic comorbidities of this condition highlights that it is imperative that these patients are diagnosed early, treated promptly, and are monitored periodically.

**Key Words:** Infantile glaucoma, Phacomatosis Pigmentovascularis; Sturge-Weber syndrome.

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## INTRODUCTION

Phacomatosis Pigmentovascularis (PPV) is a rare group of genetic disorders characterized by the presence of pigmentary and vascular nevus. PPV is classified depending on its cutaneous involvement.<sup>1</sup> Over 50% of PPV have extracutaneous involvement, with blue sclera and glaucoma as the most common ocular findings, and the most common systemic finding being Sturge-Weber syndrome (SWS). SWS is a rare congenital neurocutaneous disorder that involves capillary malformations characterized by the presence of a facial Nevus Flammeus or port wine

stain along with ocular and leptomeninges abnormalities. It is considerably rare and occurs only in approximately 1 in every 50,000 infants.<sup>2</sup> Exact global prevalence of concurrent PPV and SWS is unknown due to its sporadic occurrence, although roughly 200 cases of PPV itself have been reported since it was first diagnosed in 1947 until the early 2000s.<sup>3</sup>

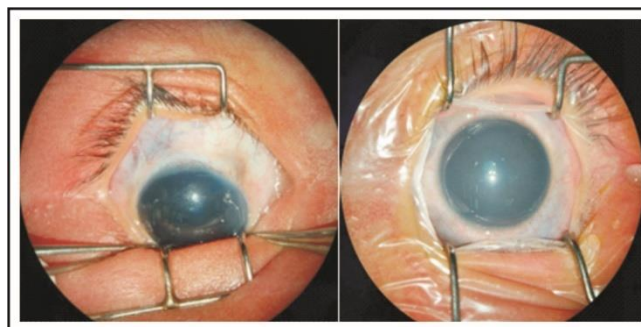
Ocular abnormalities are present in 50% of patients with SWS and glaucoma being the most common manifestation. Typically, glaucoma affects the eye unilaterally and ipsilateral to the port-wine stain. It eventually affects the other eye if left untreated. Early detection and treatment are essential to prevent irreversible vision loss.<sup>2</sup>

In this case report, we present a rare case of bilateral glaucoma in an infant with PPV and Sturge-Weber syndrome. Glaucoma resolved with trabeculectomy, trabeculotomy and topical anti-glaucoma medication with favourable outcome.

## Case Description

A three-month old male infant was referred to our hospital from a local ophthalmologist with chief complaint of hazy grayish cornea and visible black spots in the upper sclera of both eyes for one month. The family also noted that the patient was not responsive to toys or objects and seemingly had photophobia. Moreover, the patient had widespread skin discoloration since birth. At initial examination, the patient had elevated intraocular pressure (IOP) of 40 and 52 mmHg and positive blink reflex in the left and right eye, respectively. Ophthalmologic examination revealed scleral melanocytosis in both eyes (Figure 1). Systemic examination revealed widespread bilateral port-wine stain across the face and dermal melanocytosis across the torso and extremities (Figure 2). The patient also had suspected developmental delay. Echocardiogram revealed mild tricuspid regurgitation, and brain MRI with contrast showed leptomeningeal enhancement of the left fronto-parieto-temporo-occipital region. There was cerebral atrophy predominantly of the left hemisphere, with dilatation of the left posterior horn choroid plexus. The patient was diagnosed with PPV and SWS by the pediatric department at our center. The patient was born full-term with no complications, and the mother was healthy. Patient had no history of seizures and no remarkable medical history prior to this. There was no family history of consanguinity, glaucoma or other neurocutaneous diseases.

Pharmacologic management with topical antiglaucoma medication (Timolol maleate 0.25% twice a day, Latanoprost 0.05 mg/mL once a day and Brinzolamide 10 mg/mL three times a day for both eyes) was initiated prior to surgery. On the day of surgery, pre-operative examination under anesthesia (EUA) was performed.

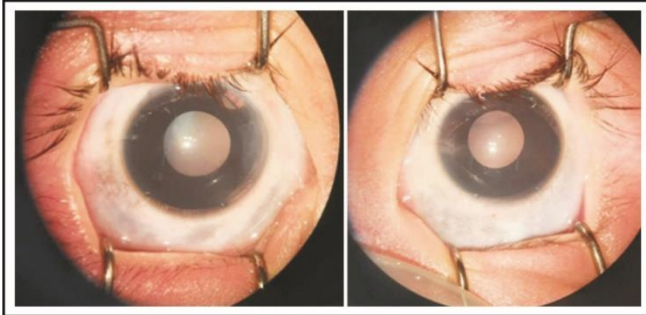


**Figure 1:** Pre-operative examination revealed scleral melanocytosis in the right eye (a) and left eye (b).

The IOP was 33 mmHg in the right eye and 23 mmHg in the left eye and corneal diameter of 13 mm bilaterally. Trabeculectomy and trabeculotomy with mitomycin C (MMC) was done in the right eye under general anesthesia. Post-operative medication included levofloxacin eyedrops (5 mg/mL) once every hour and prednisolone (10 mg/mL) eyedrops once every two hours. At follow-up, IOP of the right eye was 17



**Figure 2:** Widespread nevus flammeus or port-wine stain across the face and dermal melanocytosis across the torso and extremities.



**Figure 3:** Postoperative examination at 1 year follow-up showed less corneal haziness in the right eye (a) and left eye (b).

mmHg at day one, 13 mmHg at one week 20 mmHg at one month post-operatively. The same procedure on the left eye was performed. Pre-operative EUA revealed IOP of left eye as 31 mmHg and corneal diameter of 14 mm. One day after the operation, IOP of the left eye was 15 mmHg. At one-week it was 14 mmHg and at one-month 18 mmHg. Two months after surgery EUA was conducted which showed IOP of 36 mmHg in the right eye and 31 mmHg in the left eye. Corneal diameter was 14 mm in both eyes. The patient was started with topical antiglaucoma medication of Timol maleate 0.5% (5 mg/mL) eyedrops twice a day and Brinzolamide (10 mg/mL) eye drops three times a day. After two weeks IOP lowered to 19.2 mmHg in the right eye and 16.2 mmHg in the left eye.

EUA at one-year follow-up showed IOP of 18.5 mmHg in the right eye and 19.7 mmHg in the left eye, with corneal diameter of 13.5 mm in both eyes. Cornea was less hazy (Figure 3). The patient was responsive to light and object and there was no photophobia. The patient is now routinely monitored for glaucoma and clinical changes and is periodically monitored by the pediatrics and physical rehabilitation team to ensure appropriate growth and development.

## DISCUSSION

PPV is a rare disease that involves vascular malformations and pigmentary nevus. In our case, the patient presented with bilateral Nevus Flammeus and dermal melanocytosis. This was classified as type IIb or cesioflammae PPV according to the new classification by Happle.<sup>5</sup> Cesioflammae PPV serves as the most common type of PPV, accounting for 75-85% of PPV cases.<sup>1</sup> Moreover, the patient also had bilateral ocular melanosis, which was noted as the most common ocular abnormality in a previous study.<sup>4</sup>

The patient was also diagnosed with SWS, which shares similarities to PPV with its manifestation of nevus flammeus particularly in the segment of the ophthalmic branch of the trigeminal nerve. SWS is distinct with its ipsilateral neurological manifestation of leptomeningeal angiomas, which was present in our patient's MRI. Accordingly, our patient can be classified as Type I SWS according to the Roach Scale. Interestingly, SWS classically presents with unilateral intracranial angiomas with unilateral cutaneous involvement. Our patient presented with bilateral cutaneous and neurological involvement along with bilateral glaucoma. There have been sporadic reports of these rare, isolated conditions, but the combination of all these systemic features in concurrence with glaucoma and its bilateral manifestations are few in literature.<sup>5</sup>

Both PPV and SWS share increased likelihood of glaucoma, although the exact underlying mechanism is complex and not well understood. Both PPV and SWS are related to embryological impaired development of the neural crest cells, and are linked to somatic, mosaic mutations of the G protein guanine nucleotide binding protein alpha subunit q (GNAQ). These mutations are predominantly found in the endothelial cells which lead to increased capillary proliferation and growth thus manifesting as its characteristic vascular malformations in the nervous system, skin, and eyes. Pathogenesis of congenital glaucoma is due to anterior chamber angle malformation leading to increased resistance to aqueous humor outflow. Another main mechanism is high episcleral venous pressure due to arteriovenous shunts from episcleral hemangiomas, however this is typically found in late onset glaucoma. Ocular melanocytosis in PPV may also lead to glaucoma due to melanocytic infiltration of the trabecular meshwork, hyperpigmentation of the iris, or pigment dispersion in the iridocorneal angle, owing to secondary childhood glaucoma.<sup>1</sup> Three peaks can be seen in the age of onset of glaucoma in SWS: 40% occur in the first year of life, 20% between five to nine years, whereas 23% occur after 20 years of age.<sup>6</sup> Thus, our patient's presentation of glaucoma at three months can be considered typical for SWS.

Treatment of glaucoma in SWS or PPV is frequently challenging. Medical management may have a role in late-onset glaucoma in SWS, although its use in congenital glaucoma in SWS is noted to be ineffective and serves a more supportive role prior to surgery. Moreover, surgical intervention is widely

adopted for treatment of glaucoma in children below two years of age. Although goniotomy is considered first-line of surgical management of congenital glaucoma with anterior chamber malformation, hazy corneas warrant filtering surgery. The patient in our study presented with a relatively hazy cornea, thus goniotomy surgery would not be appropriate. In a previous study on glaucoma in children with SWS by Gjerde et al, 50% of their patients required surgery which included trabeculotomy, goniotomy, and trabeculectomy with mitomycin C, or glaucoma drainage device implant.<sup>7</sup> A study by Mandal et al, on glaucoma in children with PPV for a period of 14 years reported that 36 of 38 patients underwent surgical procedure, 10 of which were repeat surgeries, the most common being combined trabeculotomy-trabeculectomy (39 of 55 eyes).<sup>8</sup> Similarly, Sood et al, also reported 41.7% success rate of combined trabeculotomy-trabeculectomy in early-onset glaucoma.<sup>9</sup> Combined trabeculotomy-trabeculectomy involves the simultaneous creation of two drainage pathways to overcome abnormalities in the anterior chamber and facilitate the outflow of aqueous humor. Trabeculotomy establishes an anatomical communication between the anterior chamber and the Schlemm canal, while trabeculectomy creates a fistula connecting the anterior chamber with the subconjunctival space. These two procedures establish alternative pathways for aqueous humor drainage, bypassing the conventional episcleral venous system. Theoretically, this dual approach offers the potential for long-term and enhanced control of IOP.

In terms of IOP control, Mandal et al. reported that post-operatively, 33% of eyes in their cohort still required medication to control IOP.<sup>8</sup> This is in line with our study in which the patient underwent trabeculotomy-trabeculectomy which resolved glaucoma initially, however later elevation of IOP required continuation of anti-glaucoma medication and this successfully controlled IOP at one year follow-up. Consideration for repeat filtering surgery or glaucoma drainage device (GDD) in cases of refractory glaucoma in SWS or PPV is often made based on clinician's judgement and preference. A prior study by Sood et al. on glaucoma associated with SWS reported that the first additional filtration surgery was conducted at an average of 23.13 months following primary combined trabeculotomy-trabeculectomy surgery. The second surgery was conducted at 55.5 months after the primary surgery. The third and fourth

surgeries were required in four and one eye respectively, conducted more than an average of 120 months after initial surgery. These findings highlight the importance of continued long-term follow-up in patients due to the high rates of glaucoma recurrence.<sup>9</sup>

A prior study on GDD implantation in SWS patients reported that although the average age at diagnosis was 0.73 years, GDD surgery was typically performed much later, at an average age of 4.8 years. The study indicated a 36% overall failure rate, with initially high success rates that declined over time: 91% at 1 year, 75% at 3 years, and 52% at 5 years. Additionally, there was a high rate of complications, including infection, persistent hypotony, and cilioretinal artery occlusion.<sup>10</sup> Another study on Ahmed valve implants in SWS patients with childhood glaucoma reported a success rate of 75% and found the procedure to be relatively safe for treating childhood glaucoma.<sup>11</sup> With these studies, it is implied that GDD may be an effective and safe option in childhood glaucoma, however, its superiority over other surgical options within the context of a rare case such as childhood glaucoma with SWS or PPV cannot be clearly determined.

Post-operative complications include hyphema, choroidal hemorrhage, and shallow anterior chamber. Our patient did not experience these complications, and at last follow-up achieved normal IOP.

This case highlights the complexity of treating childhood bilateral glaucoma in a patient with coexisting SWS and PPV. No clear guidelines exist to treat this complex case; however, first-line treatment remains to be surgery with close follow-up with consideration of additional antiglaucoma medication or potential of repeat surgery in case of recurrence. Considering the high potential for recurrence in glaucoma in PPV and SWS, life-long glaucoma surveillance is essential.

## CONCLUSION

Our case is a rare presentation of concurrent SWS, PPV, and childhood bilateral glaucoma. The potential systemic comorbidities of this condition highlights that it is imperative that these patients are diagnosed early, treated promptly, and are monitored periodically.

**Patient's Consent:** Researchers followed the guidelines set forth in the Declaration of Helsinki.



**Conflict of Interest:** Authors declared no conflict of interest.

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## Authors Designation and Contribution

Dian Estu Yulia; Consultant Ophthalmologist: *Concepts, Design, Literature search Data acquisition, Data analysis, Statistical analysis, Manuscript preparation, Manuscript editing, Manuscript review.*

Diajeng Ayesha Soeharto; General Practitioner: *Design, Literature search Data acquisition, Data analysis, Statistical analysis, Manuscript preparation, Manuscript editing, Manuscript review.*

