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Resolution of exudative retinal detachment and optic disc edema in a child with Sturge Weber syndrome and congenital cyanotic heart disease after cardiac surgery

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ABSTRACT

Purpose: We present a case of unilateral exudative retinal detachment and optic disc edema with choroidal hemangioma in a patient concurrently diagnosed with Transposition of the Great Arteries (TGA) and Sturge-Weber Syndrome (SWS).

Observations: A seven-year-old patient presented with complaints of headache, photophobia, periocular pain, and diminished vision, accompanied by redness and lid swelling following a febrile illness. Ophthalmic examination revealed abnormally branched episcleral vasculature, exudative retinal detachment, optic disc edema and radiologically evident thickening of the choroid of the right eye. Systemic evaluation revealed hypoxia with cyanosis and clubbed fingers. The patient underwent cardiac interventions, including Fontan and Bidirectional Glenn procedures, aimed at improving systemic oxygenation. The patient's condition showed remarkable improvement following cardiac surgery. The surgical interventions not only improved systemic oxygenation but also resulted in the amelioration of ocular symptoms and visual improvement. Ophthalmic improvements included the resolution of abnormally branched episcleral vasculature, regression of exudative retinal detachment, and reduction in optic disc edema.

Conclusion and Importance: This case highlights the intricate relationship between hemodynamic and vascular abnormalities of the eye and the clinical manifestations observed in patients presenting with the unique combination of SWS and TGA.

1. Introduction

The relationship between congenital heart disease (CHD) and ocular symptoms has garnered limited attention, with reported prevalence ranging from 6.3 % to 65 %.[1] Ocular manifestations in CHD patients are often attributed to associated syndromes, and a possible embryological linkage between ocular pathology and cardiac defects has been speculated.[2] However, retinal vascular dilation and tortuosity are the most common ocular findings in cyanotic CHD, primarily attributed to hypoxemia and increased vascular endothelial growth factor (VEGF) release. Additionally, the surge in angiogenesis-related VEGF is known to cause abnormal blood vessel formation.[3,4]

Hence, there could be a unique possibility of ocular manifestations stemming from the interaction between the syndrome and cyanotic heart disease, which may be altered following cardiac intervention. Nonetheless, this aspect is rarely explored and sparsely documented in the existing literature. Hence, the dearth in literature inspires us to document the association between syndrome and CHD through this case report. The authors present a case of SWS with unoperated transposition of the great arteries (TGA) demonstrating unilateral abnormal branched purplish episcleral vasculature, exudative retinal detachment, and optic disc edema. This case highlights the correlation between cardiac intervention and improvement in ocular symptoms, emphasizing the interplay between hemodynamic alterations and clinical manifestations in patients with concurrent Sturge-Weber syndrome and TGA.

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2. Case report

A 7-year-old male presented to the Oculoplastic service at a tertiary eye care centre with complaints of recurring headaches, acute vision loss in the right eye (OD), accompanied by photophobia, periocular pain, redness, and lid swelling for the past four days. The patient had recently experienced a febrile illness lasting one week, which subsided with oral paracetamol, with no history of trauma, diplopia, vomiting, drug allergies, or recurrent respiratory tract infections. His medical history included an unoperated cyanotic CHD, notably TGA with a large ventricular septal defect, severe pulmonary stenosis, and normal biventricular function (Fig. 1). The patient also had a history of poor feeding and shortness of breath over the last four years. The child's parents were unable to seek prior medical attention due to their remote location and financial constraints. There was no significant family history.

Ophthalmic examination revealed that the patient's OD had perception of light with a relative afferent pupillary defect while the left eye (OS) had a visual acuity of 20/40. Intraocular pressure (IOP) was 30 mmHg OD and 16 mmHg OS. He had a congenital right sided facial port-wine birthmark involving the forehead, eyelids, and right cheek. OD exhibited abnormal branched purplish episcleral injection, chemosis, and mild lid edema (Fig. 2A). Fundus examination revealed optic disc edema, tortuous retinal vessels, and bullous exudative retinal detachment OD. Retinal vascular tortuosity and dilatation were also observed OS (Fig. 3). Subfoveal choroidal thickness, measured by swept-source optical coherence tomography, was 390 μm OD and 304 μm OS. On general physical examination, the patient appeared lethargic and partially oriented to time, place, and person. His height was 108 cm, he weighed 15 kg, and he had cyanosed lips with clubbed fingers (Fig. 2B). A presumptive diagnosis of vascular hemangioma involving choroid, episclera of OD with presumed SWS was made, and a contrast-enhanced Computerised tomography (CT) scan was advised along with an urgent paediatric cardiology opinion. He was started on oral paracetamol along with OD topical dorzolamide-timolol and moxifloxacin eye drops.

Laboratory investigations revealed normal renal and liver function tests, with elevated hemoglobin levels of 20.1 g/dl, decreased platelet count of 10,000 cells/mm^3 of blood, and a normal total lymphocyte count. Arterial gas analysis showed extremely low pO2 levels at 58 %. Dual-energy CT scan revealed cortical gyri-form calcifications in the right parieto-occipital lobe, extensive bilateral leptomeningeal angiomatosis and right lateral ventricle choroid plexus enlargement, with focal asymmetric enlargement of OD posterolateral quadrant (Fig. 4A–B), confirming the diagnosis of SWS. Magnetic Resonance Imaging (MRI) of the orbits showed diffuse thickening and corresponding enhancement of the choroid OD (Fig. 4C–D), suggestive of a diffuse choroidal hemangioma with normal vascular calibre and flow rate on Magnetic Resonance head angiography. Abdominal ultrasonography was unremarkable.

He subsequently underwent primary Fontan and bidirectional Glenn shunt procedures, involving the anastomosis of both the superior and inferior vena cava to the right pulmonary artery to address systemic hypoxia and inadequate oxygenation. The post-operative period was uneventful with arterial oxygen saturation improving to 85 %, haemoglobin levels falling to 14 g/dl, and platelet count increasing to 150,000 cells/mm3 on post-operative day three. Clinically, cyanosis and dyspnoea also improved and resolved over the next 2 months.

Topical antibiotics and a combination of two anti-glaucoma medications were continued for six weeks in OD. The IOP at six weeks was noted to be 8 mmHg in OD and 16 mmHg in OS. This was followed by the use of a single anti-glaucoma medication, timolol, for another six weeks in OD. On ocular examination at 3 months post-cardiac intervention, the best-corrected visual acuity improved to 20/120 in OD and 20/20 in OS, while IOP was 13 mmHg in OD and 16 mmHg in OS. There was complete resolution of episcleral vascular engorgement (Fig. 2C), lid edema, and exudative retinal detachment in OD, with normalization of retinal vasculature in OS. The facial port-wine birthmark became considerably less pronounced but did not disappear completely. At 9 months follow-up, the child was in good health, with no signs of ocular (Fig. 2D) or systemic recurrence. At 9 months, IOP was noted to be 16 mmHg (off medications since 6 months) in OD and 14 mmHg in OS. This case report conforms to the principles of the Helsinki Declaration and has been conducted with the ethical approval of the Institute Ethics Committee-Postgraduate Institute of Medical Education and Research, Chandigarh,

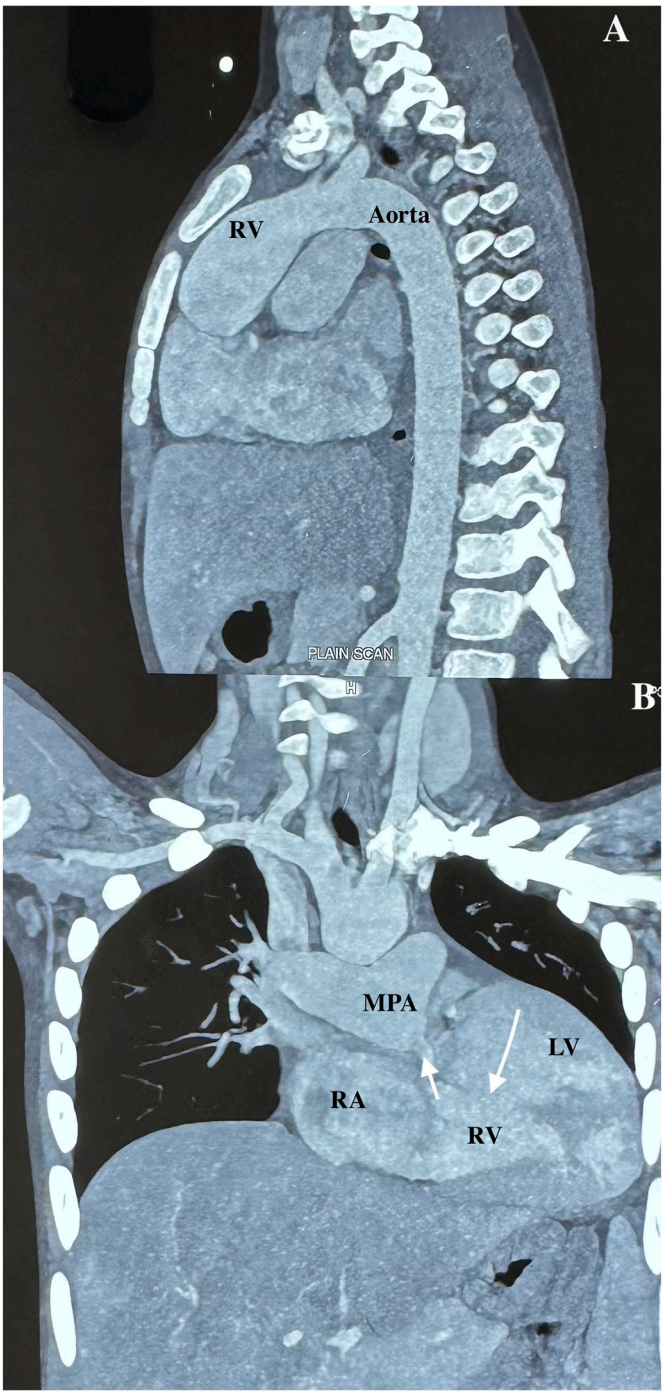

Fig. 1. Computed Tomography angiogram depicting Transposition of the Great Arteries (TGA). A, Aorta originating from the Right Ventricle (RV); B, Coronal section illustrating enlarged chambers of the heart including the Right Atria (RA), right ventricle (RV), left ventricle (LV), and Main Pulmonary Artery (MPA) arising from the left ventricle. The patient had a large ventricular septal defect (indicated by a large arrow) and severe stenosis of the pulmonary artery (small arrow).

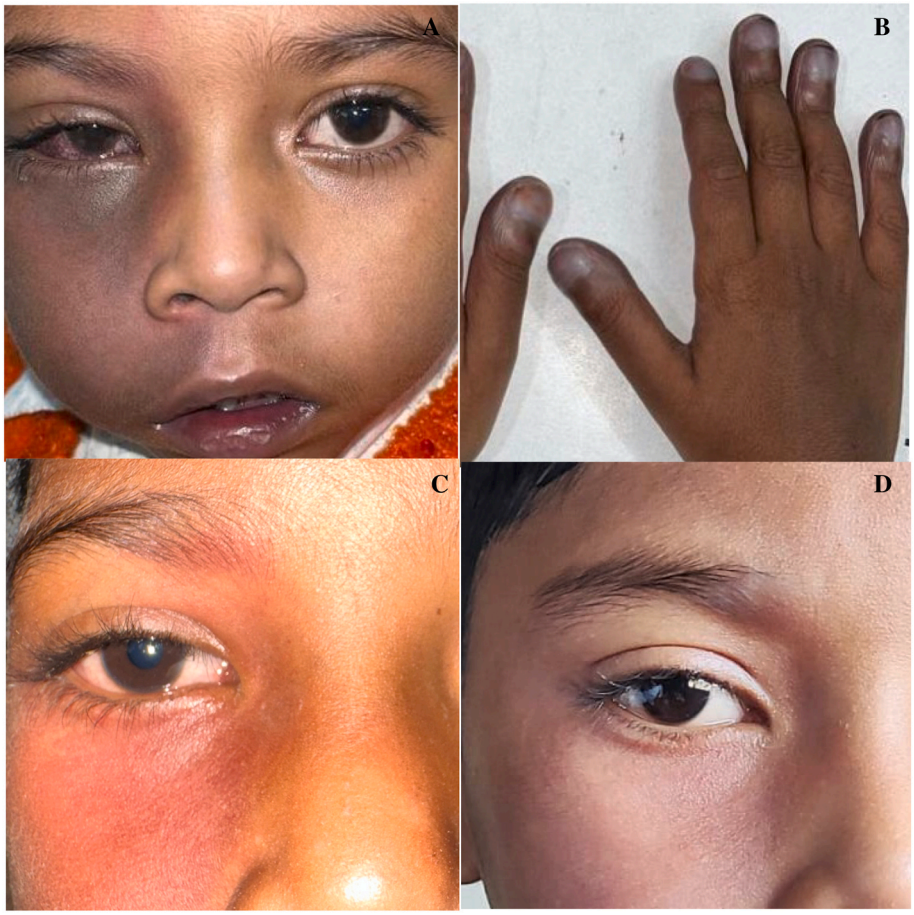

Fig. 2. A, Clinical photograph of the patient at presentation, showing a prominent port wine stain extending across the right side of the forehead, eyelids, cheek and cyanosed lips. The right eye reveals abnormally branched purplish episcleral vessels with minimal upper lid edema and associated mechanical ptosis. B, Bilateral clubbing of fingers with cyanosed nails. C, Marked resolution in episcleral vessel injection, ptosis, and cyanosis at 3 months following cardiac surgery. D, Resolved episcleral vessel injection at 9 months post cardiac surgery.

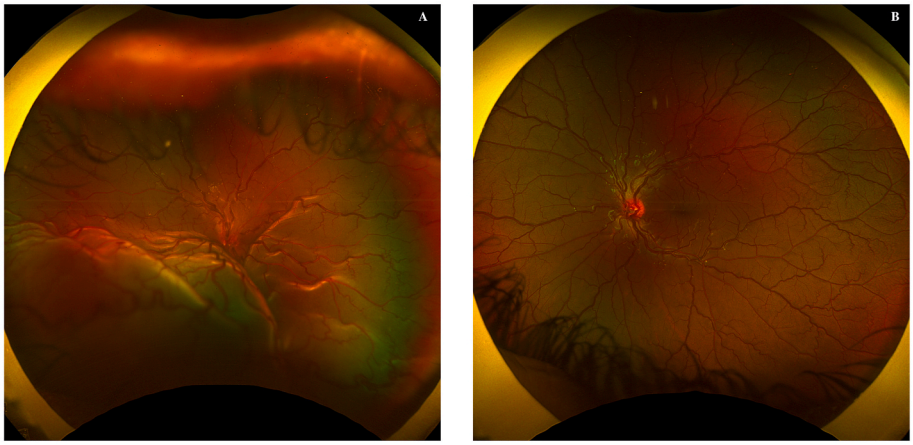

Fig. 3. A, Fundus photograph of the right eye at presentation shows optic disc edema, with inferior sub-total exudative retinal detachment and tortuous retinal vascular dilatation observed in both right and B, left eyes.

India.

3. Discussion

SWS belongs to a group of phacomatoses characterized by abnormal cell precursor development within the neural crest during early embryological development. This abnormal development leads to the formation of hamartomas, commonly observed in the central nervous system, skin, and eyes, which are characteristic of SWS.[5] The clinical signs of SWS may include a unilateral facial port-wine birthmark along the first branch of the trigeminal nerve, hemi atrophy, progressive seizures, contralateral hemiparesis, intellectual disability, hemianopia, and ipsilateral glaucoma.[6] The presence of a port-wine birthmark involving the eyelid can induce pathological alterations in ocular blood flow,

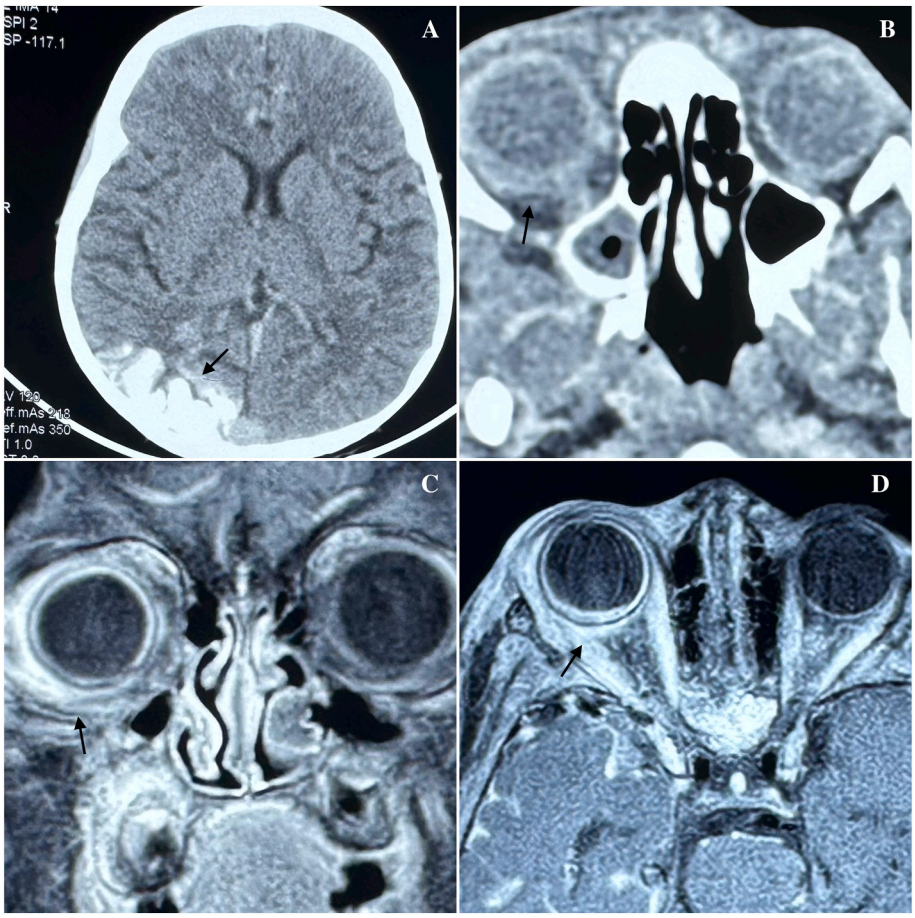

Fig. 4. Computed tomography scan at presentation: A, Axial section of brain revealing cortical gyri-form calcifications in the right parieto-occipital lobe, indicated by the black arrow. B, Axial section of eye showing focal asymmetrical enlargement involving the posterolateral wall of the right eye ball (black arrow) with a thickened optic nerve head. C-D, Coronal and axial views, respectively, on Magnetic Resonance scan demonstrates diffuse thickening of the choroid of the right eye and corresponding enhancement of the choroid as indicated by the black arrow.

resulting in diffuse or localized pinkish discoloration of the limbus, associated with increased bulbar conjunctival vascularization. It's noteworthy that approximately half of SWS patients may develop episcleral vessel dilatation.[7]

The authors present a case of concomitant SWS and TGA in a patient who exhibited unilateral episcleral injection, exudative retinal detachment and diffuse choroid hemangioma. Therefore, it is interesting to acknowledge ocular manifestations in association of CHD. In the literature, conjunctival congestion was observed in 85 % of children aged ≥5 years,[8] while tortuous-dilated retinal blood vessels and papilledema were the predominant ocular presentations seen in CHD.[9] These retinal changes were primarily noted in the posterior pole and were typically observed in paediatric patients,[10] with a more pronounced presence in those with severe right ventricular outflow obstruction.[8] The unique possibility of altered blood vessels with a distinctive arborizing pattern of episcleral vessels in SWS, associated with underlying CHD, prompted our interest in documenting this case.

At presentation, it was challenging to determine the underlying cause of abnormal purplish episcleral anastomosis and vascular congestion. Following the successful Fontan and Bidirectional Glenn procedures, which establish a direct connection between the superior vena cava and pulmonary artery, rerouting oxygen-rich blood to bypass the right ventricle and flow directly to the lungs, a significant improvement in these ocular symptoms was observed. Post-surgery, the child's systemic oxygenation improved significantly, and systemic manifestations of TGA showed marked amelioration. This highlights the potential positive impact of cardiac surgery on ocular manifestations in patients with TGA and SWS, although the exact underlying mechanism remains speculative. One possible explanation for the ocular symptoms could be chronic hypoxemia due to a right-to-left shunt across the ventricular septal defect, which acts as a stimulus for developing polycythaemia. Consequently, this increases the viscosity of blood, which in turn decrease the venous return and increases the venous pressure because of non-Newtonian properties of blood. Prolonged polycythaemia leads to widespread dilatation and tortuosity of smaller blood vessels, including those in the eyelid, conjunctiva, episclera, choroid, and retina, resulting in exudative retinal detachment with disc edema. Polycythaemia also leads to the manifestation of other symptoms and complications, such as headaches, dizziness, and thromboembolism.[3,11] Moreover, Peterson et al.[12] directly related the severity of fundus changes with arterial oxygen saturation and haematocrit levels.

It was noteworthy that the clinical manifestation was reported following an episode of febrile illness, supported by the observation that the reduction of already low pulmonary blood flow increases the right-to-left shunt fraction, thereby elevating the amount of deoxygenated blood in the systemic circulation. This condition is further exacerbated when body temperature rises after a febrile illness, creating a feedback loop that progressively worsens the patient's hypoxemia.[13] Therefore, hypoxemia induces reactivity in retinal vessels, primarily through the release of tissue metabolites in response to the abnormally low PO2 in arterial blood. Similarly, inadequate oxygenation jeopardizes normal cellular function, leading to a vasodilatory response[14] manifesting as exudative retinal detachment, disc edema, and arborized episcleral vessels.

Its differential presentation, involving only the right side of the face and the right eye, emphasizes the contributing effect of altered hemodynamic and vascular abnormalities associated with SWS. The amelioration of ocular and cyanotic CHD symptoms after surgical correction suggest that the combined effect of polycythaemia and hypoxia was the precipitating factor in this case.

4. Conclusion

The differential presentation limited to the right eye emphasize the role of altered hemodynamic and vascular abnormalities associated with SWS and TGA. This case report highlights the potential positive impact of cardiac surgery on ocular manifestations in patients with both TGA and SWS, emphasizing the need for a multidisciplinary approach to address intricate clinical conditions and improve patient outcomes. Further investigation is imperative to elucidate the exact mechanisms behind the observed ocular alterations and to discern the therapeutic implications of cardiac interventions in similar cases.

Patient consent for publication

Written consent to publish personal information and case details has been obtained from his parents.

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CRediT authorship contribution statement

Manu Saini: Writing – original draft, Visualization, Validation, Supervision, Methodology, Conceptualization. **Mohit Dogra:** Writing – review & editing, Visualization, Validation, Supervision, Investigation, Data curation. **Kulbhushan Saini:** Writing – review & editing, Writing – original draft, Validation, Supervision, Methodology, Conceptualization. **Ravindra Kumar Jaiswal:** Supervision, Methodology, Investigation, Formal analysis, Data curation.

Declaration of competing interest

We declare that there is no conflict of interest.

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