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Case Report

Secondary open-angle glaucoma and systemic manifestations in a patient with unrepaired tetralogy of fallot

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ABSTRACT

Tetralogy of Fallot (TOF) is the most common cyanotic congenital heart disease. We report a rare case of secondary open-angle glaucoma in a 20-year-old male with longstanding, uncorrected TOF. The patient presented with 6 months of progressive bilateral vision loss. Ocular examination revealed prominent conjunctival vessel engorgement, elevated intraocular pressure, advanced glaucomatous optic neuropathy with increased cup-to-disc ratios, and significant retinal nerve fiber layer thinning. Notable systemic features included severe digital clubbing, skeletal deformities, congenital ear anomalies with hearing loss, short stature and hypogonadism. Prompt initiation of anti-glaucoma therapy successfully normalized intraocular pressure. This case underscores the importance of routine ophthalmic screening in patients with cyanotic congenital heart disease to detect and manage vision-threatening complications early.

Keywords: Tetralogy of fallot, Secondary open-angle glaucoma, Cyanotic congenital heart disease, Ocular complications

INTRODUCTION

The most common form of cyanotic congenital heart disease is Tetralogy of Fallot (TOF) which is characterized by four particular features that include pulmonary stenosis or atresia, ventricular septal defect, hypertrophy of the right ventricle, and overriding aortic root.[1] The incidence of TOF is three to every 10,000 live births and it represents about 5-7 percent of all the congenital heart diseases.[2] Compensatory polycythemia is provoked by chronic hypoxemia caused by right-to-left shunting, which leads to elevated blood viscosity and extensive vascular alterations, including retinal and conjunctival vessels tortuosity, digital clubbing, and other systemic manifestations that are more typical of uncorrected cases.[3,4] There is also evidence that congenital heart disease can be associated with several ocular pathologies such as retinal vascular tortuosity, optic disc hypoplasia, and conjunctival congestion that may be used as clinical signs of hyperviscosity associated with polycythemia.[4,5] TOF has also been reported in rare case reports related to central retinal artery occlusion or bilateral persistent pupillary membranes, which are evidence of rooted in hypoxia retinal vascular pathophysiology.[6,7] Glaucoma is a chronic progressive optic neuropathy with classical morphological changes at the optic nerve head, loss of retinal nerve fibre layer and visual field.[8] The secondary glaucoma develops as a resultant complication of another underlying disease and the causative factors must be identified in order to maintain vision. Secondary glaucoma is highly uncommon in TOF but its clinical manifestation is grave because it might cause permanent loss of vision due to a mechanism that might involve polycythemic hyperviscosity, poor ocular perfusion and increase in episcleral venous pressure.[9] Although familial relationships between TOF and glaucoma have been indicated, practically no reported instances of adult presentation of uncorrected TOF by secondary glaucoma

and multisystem defects have been reported especially in low-resource countries like Bangladesh. The case highlights the importance of full ophthalmic assessment in chronic cyanotic congenital heart disease to diagnose and treat such eye damaging conditions at early stages.

CASE REPORT

A 20-year-old male from rural Bangladesh presented to a tertiary eye hospital in the northern region of the country with a six-month history of progressive bilateral vision loss. Parents reported a known diagnosis of uncorrected TOF since infancy, confirmed by a cardiologist, though surgical intervention had never been pursued due to socioeconomic constraints. Family history was negative for glaucoma, congenital heart disease and consanguineous marriage.

Ophthalmic examination revealed uncorrected visual acuity of 6/60 in the right eye and 6/12 in the left eye, with no improvement on pinhole testing. Goldmann Applanation Tonometry (GAT) measured intraocular pressure (IOP) at 35 mmHg in the right eye and 30 mmHg in the left eye. Slit-lamp biomicroscopy demonstrated normal eyelids and eyebrows bilaterally. Cornea was clear in both eyes. Both eyes exhibited striking bilateral bulbar conjunctival congestion characterized by markedly engorged, tortuous vessels extending toward the limbus- a classic sign of chronic polycythemia secondary to longstanding cyanosis (Figure 1). Anterior chamber depth was normal without cells or flare. Pupil was round, regular, and reactive to light with no relative afferent pupillary defect in both eyes. Lenses appeared clear in the visible pupillary aperture. Gonioscopy revealed open angles bilaterally with normal landmarks and no neovascularization or synechiae.

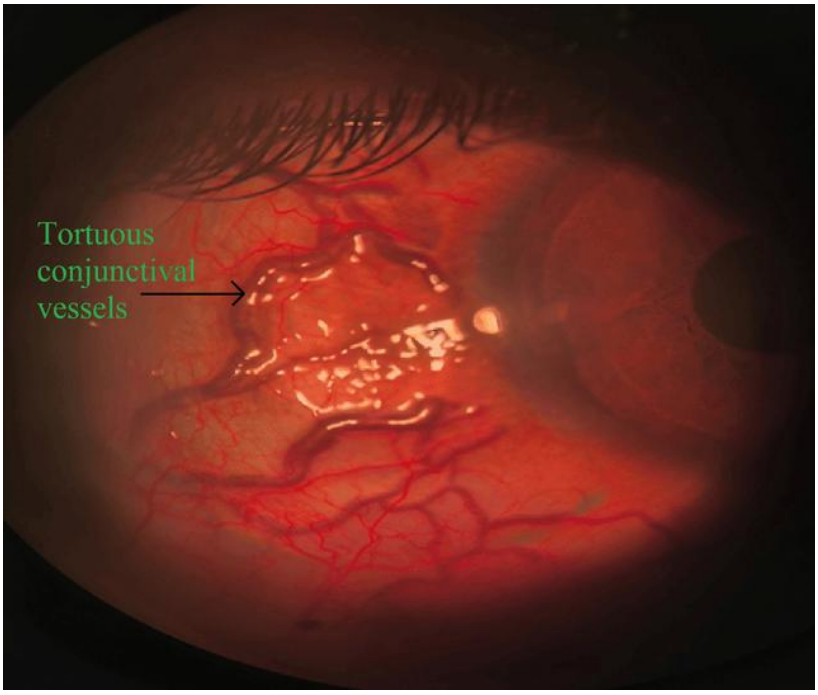

Figure 1: Slit-lamp photograph showing engorged, tortuous conjunctival vessels.

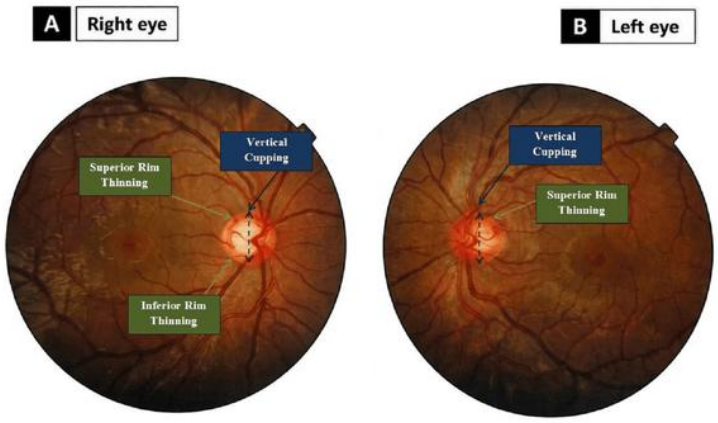

Figure 2 (A and B): Color fundus photography showing glaucomatous optic neuropathy.

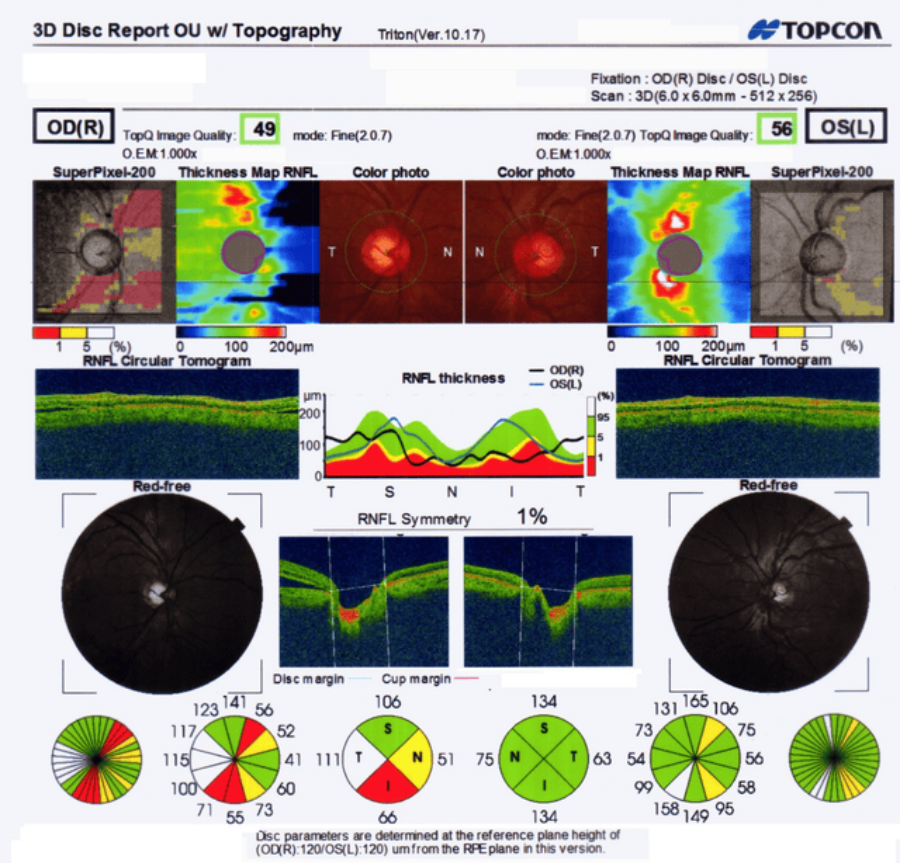

Figure 3: OCT RNFL analysis showing significant thinning and glaucomatous damage.

*Abbreviations: RNFL = retinal nerve fiber layer; RPE = retinal pigment epithelium; OCT = optical coherence tomography; OD = oculus dexter (right eye); OS = oculus sinister (left eye); TopQ = topography quality; O.E.M. = optical eye model (magnification correction); RF = red-free; µm = micrometers; mm = millimeters; OU = oculi unitas (both eyes); 3D = three-dimensional

Post-mydriasis (1% tropicamide) dilated fundus examination disclosed glaucomatous optic neuropathy in both eyes. The optic disc of both eyes showed vertical optic disc cupping, neuroretinal rim thinning and deep optic nerve excavation. Colour fundus photography documented these findings (Figure 2).

Optical Coherence Tomography (OCT) of the right eye showed an average Retinal Nerve Fiber Layer (RNFL) of 84 µm (superior 106 µm, inferior 66 µm), a disc area of 2.86 mm², a vertical C/D ratio of 0.88, and a markedly reduced rim volume of 0.04 mm³. The left eye showed an average RNFL of 102 µm (superior 134 µm, inferior 134 µm), a disc area of 2.74 mm², a vertical C/D ratio of 0.85, and a rim volume of 0.05 mm³ (Table 1). Both eyes demonstrated significant vertical cupping and reduced rim tissue, with the right eye showing more advanced inferior quadrant thinning (Figure 3). Humphrey visual field analysis could not be performed due to decreased visual acuity in right eye and non-cooperation of patient. Pachymetry also revealed normal central corneal thickness.

Systemic examination revealed short stature, mild intellectual disability, and profound sensorineural hearing loss secondary to bilateral congenital external ear deformities (Figure 4). Severe digital clubbing affected all fingers and toes (Figure 5 and 6), with hypertrophic osteoarthropathy evident as skeletal deformities in extremities. The patient reported progressive hypogonadism manifested by decreased libido and bilateral testicular atrophy.

Figure 4: Congenital ear abnormality contributing to hearing impairment.

Table 1: RNFL thickness measurements and optic disc topography parameters.

Parameter	Right eye (OD)	Left eye (OS)
RNFL thickness measurements		
Total thickness (µm)	84	102
Superior thickness (µm)	106	134
Inferior thickness (µm)	66	134
Optic disc topography parameters		
Disc area (mm²)	2.86	2.74
Cup area (mm²)	2.18	2.02
Rim area (mm²)	0.68	0.71
C/D area ratio	0.76	0.74
Linear CDR	0.87	0.86
Vertical CDR	0.88	0.85
Cup volume (mm³)	0.69	0.45
Rim volume (mm³)	0.04	0.05
Horizontal disc diameter (mm)	1.92	1.91
Vertical disc diameter (mm)	1.90	1.83

*Abbreviations: OD = oculus dexter (right eye); OS = oculus sinister (left eye); RNFL = retinal nerve fiber layer; C/D = cup-to-disc; CDR = cup-to-disc ratio; µm = micrometers; mm = millimeters; mm² = square millimeters; mm³ = cubic millimeters

Figure 5: Hand photograph demonstrating advanced clubbing.

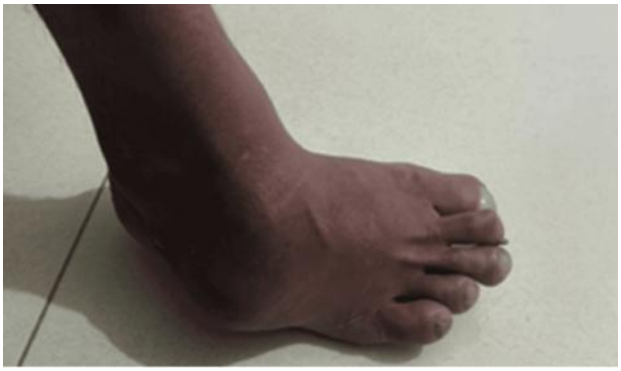

Figure 6: Foot deformity with severe digital clubbing in the patient.

Since glaucoma is irreversible, the treatment was aimed at reduction of IOP to maintain remaining vision. The treatment involved the use of topical brimonidine tartrate 0.2% twice daily, dorzolamide 2% three times daily in both eyes, and oral acetazolamide 250 mg twice daily, with cardiology referral to evaluate TOF. One-month follow-up demonstrated excellent IOP control at 16 mmHg bilaterally (right eye 16 mmHg, left eye 16 mmHg) with GAT. Visual acuity remained stable at 6/60 in right eye and 6/12 in left eye.

DISCUSSION

We report a unique case of secondary open-angle glaucoma in a young adult with untreated TOF, and various systemic effects of chronic hypoxaemia such as severe digital clubbing, skeletal malformation, congenital hearing loss, and hypogonadism. The case is special due to a number of factors: the patient survived to adulthood without being repaired surgically in a low-resource environment, the glaucomatous damage is at an advanced stage when the case was referred, and the patient responded successfully to the traditional IOP-lowering treatment.

Chronic polycythaemia and hyperviscosity are considered the best way to explain the pathophysiology between uncorrected TOF and secondary glaucoma in this patient. Compensatory erythrocytosis results due to longstanding cyanosis caused by right-to-left shunting and regularly raises haematocrit to over 65%.[1] This hyperviscosity elevates episcleral venous pressure that impedes the outflow of the trabecular and consequently elevates IOP.[3,6] Visible clinical evidence of such underlying haemodynamic disruption, which occurs in up to 85% of older children with uncorrected TOF is the striking conjunctival vessel engorgement observed in our patient.4 Also witnessed is the fact that chronic hypoxia can cause endothelial dysfunction, which further deteriorates the dynamics of the aqueous humour.[10] The open angles in the gonioscopy have been effective to rule out the angle closure or neovascular effect and to affirm the secondary nature of the glaucoma of open angles.

Cyanotic congenital heart disease is also known to affect the eye, the most common manifestations of which include retinal vascular tortuosity, optic disc hypoplasia, and conjunctival congestion.[3,5] Nevertheless, secondary glaucoma is extremely rare. There is only one other previous report, a family history of TOF in 1990, that has indicated an association between TOF and glaucoma.[9] The additional reported vascular complication is the central retinal artery occlusion caused by hypercoagulability6 and the persistent pupillary membranes.[7] The vertical optic disc cupping ,neuroretinal rim thinning and deep optic nerve excavation are indicative of secondary open-angle glaucoma in addition to other vision-threatening complications of uncorrected TOF in our patient.

It multisystem extracardiac manifestations in the present case, such as extreme digital clubbing, skeletal abnormalities, ear abnormalities and hearing impairment, and hypogonadism, depict the extreme cyanotic systemic impact. The platelet-derived growth factor release in chronic hypoxia leads to clubbing, and skeletal alterations are evidence of the impaired endochondral ossification.[1] Hypogonadism, which is represented as testicular atrophy and decreased libido, is probably due to dysfunction of Leydig cells in the face of sustained hypoxaemia, as seen with other cyanotic adults.[10] Such results can hardly be found in mended TOF owing to early surgical intervention, emphasizing the fact of late or no cardiac care in resource-restricted areas like rural Bangladesh.[2]

Another interesting point of this case was the very good response to therapy. In contrast to certain types of secondary glaucoma, which are often refractory to medical treatment, our patient could be bilaterally normalised in her IOP in just one month of topical brimonidine and topical dorzolamide combined with oral acetazolamide. This strong reaction justifies the application of carbonic anhydrase inhibitors in glaucomas linked to a high episcleral venous pressure during which they decrease aqueous generation and substitute the compromised outflow.[11] Normal central corneal thickness on pachymetry was used to ascertain that the IOP measurements were correct and not effectually high.

There are clinical implications to this case. First, it highlights the importance of regular ophthalmic examination of patients with uncorrected or late developing cyanotic heart disease. Even bedside tests such as conjunctival examination, IOP test, and optic disc could identify subclinical damage before the eyes are permanently damaged. Second, it emphasizes the benefit of interdisciplinary care; although dealing with TOF cardiac repair is the absolute treatment, ophthalmologists are essential in maintaining vision in individuals where surgery has not been done or is not accessible. Third, the fact that several congenital anomalies (ear deformities, hearing loss) are present, makes the scenario of a present genetic syndrome like 22q11.2 microdeletion, likely to exist in as many as 20-25% cases of TOF.[12] Similar presentations should also be considered as genetic counselling, but it was not done in our case because of deficiency of resources.

Several limitations of this report should be acknowledged. Genetic testing was not performed, so a syndromic association cannot be definitively excluded, though the sporadic family history makes this less likely. Haematocrit and blood viscosity were not measured at presentation, limiting direct correlation with IOP elevation. Long-term follow-up beyond one month is not yet available.

CONCLUSION

This case highlights an uncommon ocular complication in a young adult with unrepaired TOF surviving into adulthood in rural Bangladesh. Prolonged cyanosis and compensatory polycythemia likely contributed to elevated episcleral venous pressure, leading to secondary open-angle glaucoma with advanced optic nerve damage at presentation. The presence of multisystem features-digital clubbing, skeletal deformities, congenital ear anomalies with hearing loss, and hypogonadism-reflects the extensive systemic burden of chronic hypoxemia when cardiac correction is delayed due to socioeconomic constraints. Prompt initiation of standard anti-glaucoma therapy successfully controlled IOP, though established visual loss proved irreversible. This case serves as a reminder for clinicians in resource-limited settings to include routine ophthalmic evaluation in the long-term management of patients with cyanotic congenital heart disease. Early detection of raised IOP and glaucomatous changes can prevent permanent visual disability in this vulnerable population.

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