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Case Report

Internal Medicine Section

A Rare Association of Atrial Septal Defect with Retinitis Pigmentosa in an Adult: Coincidence or Syndromic Link

SUMEDH KRISHNA[1], VIGNESSH RAVEEKUMARAN[2]

ABSTRACT

Atrial Septal Defect (ASD) and Retinitis Pigmentosa (RP) are individually well-recognised clinical entities, but their co-occurrence in an adult patient without a defined syndromic diagnosis is exceptionally uncommon. A 52-year-old female with Type 2 Diabetes Mellitus (T2DM) who presented with acute gastroenteritis complicated by septic and hypovolemic shock. During evaluation, she was incidentally found to have an ostium secundum ASD measuring 1.07 cm with left-to-right shunt, dilated right-sided cardiac chambers, dilated main pulmonary artery and Pulmonary Arterial Systolic Pressure (PASP) of 43 mmHg. Ophthalmological assessment revealed bilateral advanced RP, with Visual Acuity (VA) limited to light perception in both eyes and fundus findings of peripheral bone-spicule pigmentation, attenuated retinal vessels and optic disc pallor. The family history was strongly suggestive of inherited retinal disease, with multiple siblings affected by progressive visual loss and a background of consanguinity. During admission, the patient developed a transient episode of left upper limb weakness, clinically consistent with Transient Ischaemic Attack (TIA); neuroimaging showed no acute infarct. Usher syndrome was excluded by audiometry (normal bilateral hearing) and that Bardet-Biedl syndrome and Refsum disease were excluded on separate clinical grounds. The present case is reported because the non syndromic co-existence of ASD and RP may represent a coincidental association or an unrecognised shared genetic/developmental pathway. The case emphasises the need for multidisciplinary evaluation, genetic counselling and consideration of whole-exome sequencing in adults with congenital cardiac anomalies and familial retinal dystrophy.

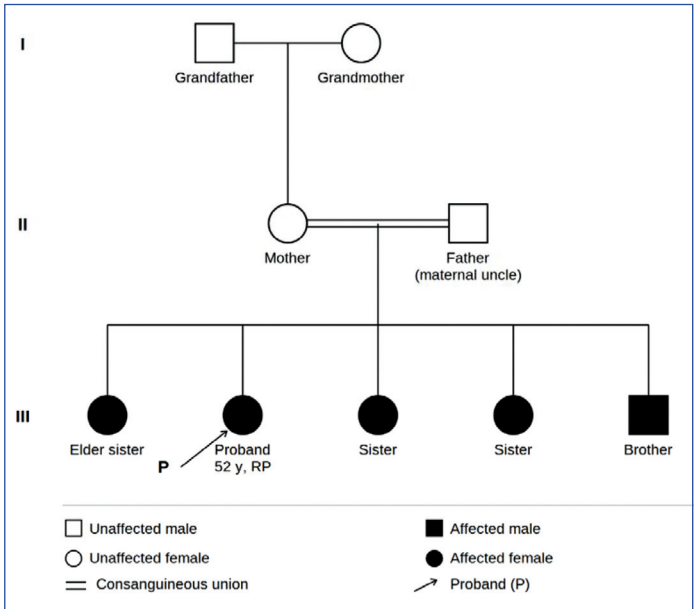
Keywords: Consanguinity, Genetic association studies, Genetic counselling, Heart septal defects

CASE REPORT

A 52-year-old female, known case of T2DM {Glycated Haemoglobin (HbA1c) 6.3%} for a year on Glycomet SR 500 mg once daily, presented to the Emergency Department with a three-day history of vomiting (4-6 episodes per day, aggravated by food intake) and loose stools (4-6 watery, non bloody episodes per day), accompanied by a single episode of low-grade fever. She also reported two days of self-spinning giddiness without specific aggravating or relieving factors, decreased appetite and reduced urine output. There was no history of chest pain, palpitations, shortness of breath, limb weakness, or slurring of speech at presentation.

The patient had been diagnosed with T2DM one year prior to this admission. She also carried a history of uterine fibroids. Of particular relevance, she had experienced progressive loss of vision in both eyes over the preceding year, with VA at presentation limited to light perception bilaterally. No prior cardiac evaluation had been undertaken.

Family history was strongly positive for progressive visual loss. The patient reported that one elder sister, two younger sisters and one younger brother had similar progressive deterioration of vision. The parents had a consanguineous marriage, as the patient's mother was married to her maternal uncle. No family history of Congenital Heart Disease (CHD) was reported. The pedigree pattern, with multiple affected siblings of both sexes born to consanguineous parents, was suggestive of a possible autosomal recessive inheritance pattern for the retinal disorder [Table/Fig-1].



[Table/Fig-1]: Pedigree chart of the patient for the consanguinity.

Clinical Examination

On examination, the patient was conscious and oriented. General examination revealed pallor and severe dehydration; no icterus, cyanosis, clubbing, lymphadenopathy, or pedal oedema were present. Vital signs on admission showed a Blood Pressure (BP) of 140/80 mmHg, Pulse Rate (PR) of 78/minute, Respiratory Rate (RR) of 24/minute and SpO_2 of 100% on room air; Capillary Blood Glucose (CBG) was 114 mg/dL. Cardiovascular examination revealed normal heart sounds without any audible murmur. Respiratory and abdominal examinations were unremarkable. Neurological examination showed a Glasgow Coma Scale (GCS) score of 15/15 with normal tone and power (5/5 in all four limbs) and flexor plantar responses bilaterally. VA was limited to light perception in both eyes.

Laboratory Investigations

Serial laboratory findings across the admission are summarised in [Table/Fig-2]. Initial investigations revealed leukocytosis with

neutrophilia {White Blood Cell (WBC) 9800cells/mm^3, neutrophils 85.5%}, normocytic anaemia (Hb 11.3 g/dL), significant dyselectrolytaemia (sodium 126 mEq/L, potassium 3.2 mEq/L), elevated hepatic transaminases (SGOT 105 U/L, SGPT 62 U/L) and a markedly elevated CRP (>300 mg/L). Serum creatinine was 1.18 mg/dL. Urinalysis demonstrated haematuria (blood 3+) with 6-8 pus cells/high-power field (hpf). Blood and urine cultures yielded no growth. Arterial Blood Gas (ABG) analysis (Day 6) showed pH 7.44, pCO_2 41 mmHg, pO_2 32 mmHg and HCO_3 27.8 mEq/L. Biochemical parameters improved progressively with treatment.

Parameters	Day 1 (30/11/25)	Day 6 (05/12/25)	Day 8 (Discharge)	Reference range*
Haemoglobin (g/dL)	11.3	8.7	9.9	12-15
Total WBC (cells/cumm)	9800	5900	6800	4000-10000
Neutrophils (%)	85.5	31.9	33.2	40-80
Platelets (/cumm)	163000	274000	388000	150000-450000
Serum Sodium (mEq/L)	126	143	140	136-143
Serum Potassium (mEq/L)	3.2	3.4	3.6	3.5-5.1
Serum Creatinine (mg/dL)	1.18	0.40	0.55	0.6-1.2
SGOT/AST (U/L)	105	20	—	<31
SGPT/ALT (U/L)	62	23	—	<34
CRP (mg/L)	>300	Not done	—	<10
Serum Albumin (g/dL)	3.9	3.1	—	3.5–5.2

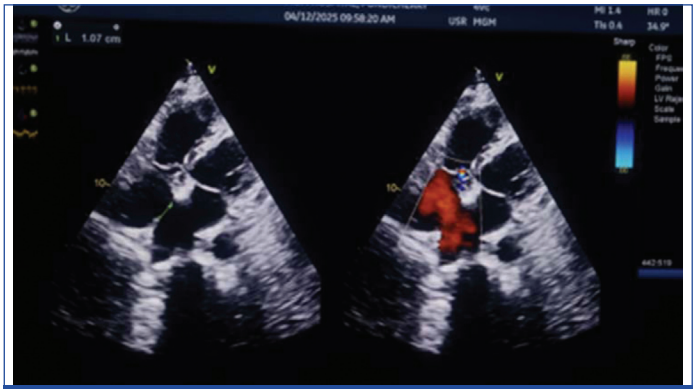
[Table/Fig-2]: Serial laboratory investigations during admission.
*The reference ranges mentioned are derived from the institutional Central Laboratory reporting standards of Mahatma Gandhi Medical College and Research Institute, Pondicherry. These ranges may vary slightly between laboratories depending on analyser methodology, calibration, reagent systems and population reference intervals.
Abbreviations: WBC: White blood cells; CRP: C-reactive protein; SGOT: Serum glutamic oxaloacetic transaminase (AST); SGPT: Serum glutamic pyruvic transaminase (ALT). Not repeated at that time point

Echocardiographic Findings

Two-Dimensional Echocardiography (2D-ECHO) [Table/Fig-3] demonstrated ostium secundum ASD with a defect measuring 1.07 cm showing left-to-right shunt on colour Doppler interrogation. Additional findings included dilated right atrium, right ventricle and main pulmonary artery, with a PASP of 43 mmHg indicating mild-to-moderate pulmonary arterial hypertension. Left Ventricular Systolic Function (LVSF) was preserved {Ejection Fraction (EF) 55%}; diastolic dysfunction was also noted. The ECHO findings are summarised in [Table/Fig-4].

Neurological Event and MRI Brain Findings

During the hospital course, the patient experienced a single episode of left upper limb weakness that was resolved spontaneously within minutes, consistent with a TIA. Magnetic Resonance Imaging (MRI) brain with Magnetic Resonance Angiography (MRA) and



[Table/Fig-3]: Two-Dimensional Echocardiography (2D-ECHO) (4-chamber view) with colour Doppler demonstrating ostium secundum ASD (defect size 1.07 cm) with left-to-right shunt (red jet). Dilated right atrium and right ventricle are evident. (Abbreviations: ASD: Atrial septal defect; PASP: Pulmonary arterial systolic pressure; LV: Left ventricle; EF: Ejection fraction)

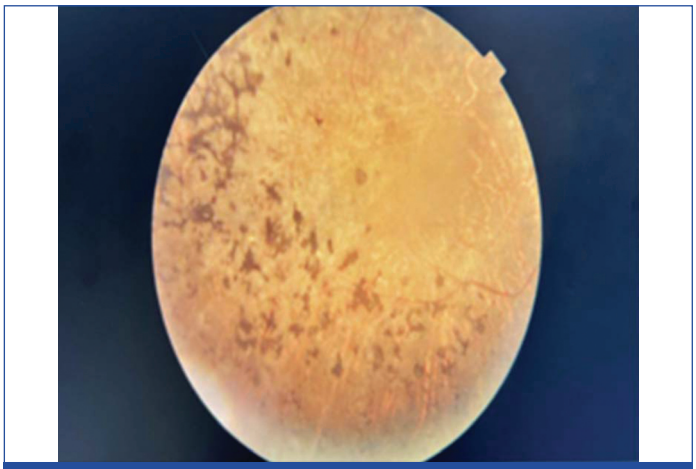
Parameters	Finding	Clinical significance
ASD type	Ostium secundum	Most common adult ASD variant
Defect size	1.07 cm	Moderate-sized; haemodynamically significant
Shunt direction (colour doppler)	Left-to-right	Volume overload of right heart and pulmonary vasculature
Right atrium	Dilated	Consequence of chronic volume overload
Right ventricle	Dilated	Consequence of chronic volume overload
Main pulmonary artery	Dilated	Pulmonary vascular remodelling
PASP (mmHg)	43 mmHg	Mild-moderate pulmonary arterial hypertension
LV systolic function/EF	Preserved / 55%	Normal LV function maintained
Diastolic dysfunction	Present	Likely related to hypertension/age

[Table/Fig-4]: Echocardiographic findings.

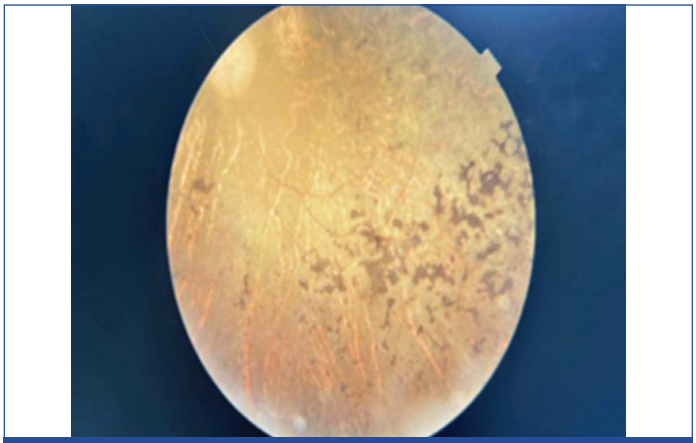
Magnetic Resonance Venography (MRV) revealed age-related neuroparenchymal changes with dilated ventricles and prominent sulcal spaces, but no evidence of acute infarct, haemorrhage, or space-occupying lesion. MRA demonstrated a hypoplastic right vertebral artery; all other cerebral vessels were normal. MRV showed no venous thrombosis. Neuromedicine opinion was obtained and dual antiplatelet therapy (Aspirin 75 mg + Clopidogrel 75 mg) along with Atorvastatin 40 mg were initiated.

Ophthalmological Findings - Retinitis Pigmentosa (RP)

Fundus examination of both eyes [Table/Fig-5,6] revealed the classical triad of RP: peripheral bone-spicule pigmentary deposits distributed throughout the peripheral retina, attenuated retinal vasculature bilaterally and waxy pallor of the optic disc with early disc



[Table/Fig-5]: Fundus photograph- right eye. Diffuse peripheral bone-spicule pigmentary deposits with attenuated vessels and relative macular preservation, consistent with Retinitis Pigmentosa (RP).



[Table/Fig-6]: Fundus photograph - left eye. Dense bone-spicule pigmentation particularly in the inferonasal quadrant, markedly attenuated vessels and early disc pallor indicating advanced Retinitis Pigmentosa (RP).

pallor more advanced on the left. Relative macular preservation was noted on the right eye; more advanced changes were evident on the left. VA was limited to light perception bilaterally. No superimposed diabetic retinopathy or papilloedema was identified.

Audiological and Ear, Nose and Throat (ENT) Evaluation

Otorhinolaryngological consultation was sought to systematically exclude Usher syndrome (the most common syndrome of RP with sensorineural hearing loss) and to evaluate the complaint of giddiness. Pure Tone Audiometry (PTA) was performed on the same day of ophthalmic assessment. The complete ENT findings are documented in [Table/Fig-7,8].

Parameters	Right ear	Left ear	Interpretation
PTA threshold	15 dBHL	15 dBHL	Within normal limits (<25 dBHL)
Rinne test	Positive (AC > BC)	Positive (AC > BC)	No conductive hearing loss
Weber test	No lateralisation	No lateralisation	No asymmetric SNHL
EAC	Normal	Normal	No external ear pathology
Tympanic membrane	Grade I (normal)	Grade I (normal)	No middle ear pathology
Facial nerve (clinical)	Intact	Intact	No facial nerve involvement
Overall impression	Normal hearing	Normal hearing	Usher syndrome excluded

[Table/Fig-7]: Pure Tone Audiometry (PTA) findings.
Abbreviations: PTA: Pure tone audiometry; AC: Air conduction; BC: Bone conduction; SNHL: Sensorineural hearing loss; dBHL: Decibels hearing level; EAC: External auditory canal; Grade I TM = Normal tympanic membrane without perforation

ENT structure/symptom	Right	Left	Comment
Hearing loss (reported)	Nil	Nil	No subjective hearing complaint
Ear discharge	Nil	Nil	—
Pain in the ear	Nil	Nil	—
Ear fullness/block	Nil	Nil	—
Tinnitus	Nil	Nil	—
Giddiness (at ENT review)	Nil	Nil	Resolved by time of ENT review
External auditory canal	Normal	Normal	—
Tympanic membrane	Grade I (intact)	Grade I (intact)	No perforation
Facial nerve (clinical)	Intact	Intact	—
Nasal cavity/Septum	Mild DNS to right	Clear	Incidental finding
Oral cavity/ Oropharynx	Clear	Clear	—

[Table/Fig-8]: ENT clinical examination summary.
Abbreviations: DNS: Deviated nasal septum. ENT Impression: No syndromic ENT pathology identified. Bilateral hearing sensitivity within normal limits

The audiological evaluation conclusively demonstrated bilateral hearing sensitivity within normal limits, with PTA thresholds of 15 dBHL (decibel hearing level) bilaterally, positive Rinne test bilaterally and no Weber positive test. This effectively excluded Usher syndrome as a unifying diagnosis.

Syndromic Evaluation

The complete clinical profile was systematically evaluated against established syndromic associations of RP with cardiac disease. The findings are summarised in [Table/Fig-9].

Hospital Course and Treatment

The patient received intravenous fluid resuscitation with boluses for septic and hypovolemic shock and inotropic support was initiated for persistent hypotension; this was subsequently tapered and discontinued as haemodynamic stability was restored. Antibiotics

Syndrome	Mandatory features	Status in this patient	Conclusion
Usher syndrome	RP + Sensorineural hearing loss ± vestibular dysfunction	Hearing normal (PTA 15 dBHL bilaterally); no vestibular deficit	Excluded
Kearns-Sayre syndrome	RP + Cardiac conduction defect + progressive external ophthalmoplegia	No conduction defect on ECG; no ophthalmoplegia	Excluded
Bardet-Biedl syndrome	RP + obesity + polydactyly + renal anomaly + hypogonadism	None of these features present	Excluded
Refsum disease	RP + cerebellar ataxia + peripheral neuropathy + cardiac arrhythmia	No ataxia, neuropathy, or arrhythmia	Excluded
Non syndromic ASD + RP	Isolated co-occurrence without a unifying syndrome	All evaluations consistent with this	Most likely diagnosis

[Table/Fig-9]: Syndromic differential diagnosis – evaluation against known RP-cardiac associations.
Abbreviations: RP: Retinitis pigmentosa; ASD: Atrial septal defect; PTA: Pure tone audiometry; ECG: Electrocardiogram

comprised Injection Meropenem 1 g intravenous thrice daily for six days and Capsule Doxycycline 100 mg twice daily for five days. Antiemetic (Injection Ondansetron 4 mg), proton pump inhibitor (Injection Pantoprazole 40 mg), electrolyte correction, calcium supplementation (Tab Shelcal 500 mg) and nutritional support were provided. Following the TIA event, dual antiplatelet therapy (Tab Aspirin 75 mg + Tab Clopidogrel 75 mg) and Tab Atorvastatin 40 mg nightly were commenced on neurological advice.

At discharge, the patient was haemodynamically stable (BP 100/60 mmHg, PR 84/min, SpO_2 98% on room air, GCS 15/15). Discharge medications included Tab Faropenam 200 mg twice daily for four days, continuation of dual antiplatelets and statin, Midodrine 1 mg (tapered), Pantoprazole 40 mg, calcium supplementation and Tab Metformin 250 mg once daily. The patient was advised to follow-up in the General Medicine Outpatient Department (OPD), Cardiology OPD for ASD management planning and Ophthalmology OPD for RP monitoring.

DISCUSSION

The ASD is one of the common congenital cardiac lesions detected in adults and the ostium secundum type accounts for the majority of adult cases. Many patients remain asymptomatic for years and are diagnosed incidentally when complications such as right heart dilatation, pulmonary arterial hypertension, arrhythmia, or embolic events occur [1,2]. In the present case, the patient had no previous cardiac evaluation and no history suggestive of CHD. However, 2D-ECHO revealed an ostium secundum ASD measuring 1.07 cm with left-to-right shunt, dilated right atrium, dilated right ventricle, dilated main pulmonary artery, preserved LVSF and PASP of 43 mmHg. These findings suggest a haemodynamically significant interatrial communication with chronic right-sided volume overload and mild-to-moderate pulmonary arterial hypertension [1-3].

The RP is a genetically heterogeneous inherited retinal dystrophy characterised by progressive photoreceptor degeneration [4,5]. The classical fundus findings include peripheral bone-spicule pigmentation, arteriolar attenuation and waxy optic disc pallor [5]. In this patient, ophthalmological evaluation showed bilateral advanced RP with VA limited to light perception in both eyes. Fundus examination demonstrated diffuse peripheral bone-spicule pigmentary deposits, attenuated retinal vessels and optic disc pallor, with more advanced changes in the left eye. Importantly, there was no evidence of superimposed diabetic retinopathy, papilloedema, or other retinal pathology despite the patient having T2DM. This helped establish RP as the primary cause of visual impairment.

The family history in the present case is clinically important. The patient reported progressive visual loss in one elder sister, two younger sisters and one younger brother. In addition, there was a history of consanguinity, as the patient's mother was married to her maternal uncle. The involvement of multiple siblings of both sexes in a consanguineous family background strongly suggests a possible autosomal recessive inheritance pattern for the retinal disorder [6,7]. Although genetic testing was not performed during admission, this pedigree pattern supports the need for genetic counselling and whole-exome sequencing of the proband and, if feasible, affected family members.

When RP co-exists with systemic disease, recognised syndromic associations must be considered and excluded. Usher syndrome is characterised by RP with sensorineural hearing loss, with or without vestibular dysfunction [4,8]. In the present patient, PTA showed hearing thresholds of 15 dB HL bilaterally, with positive Rinne test on both sides and no Weber laterisation, effectively excluding clinically significant sensorineural hearing loss. Kearns-Sayre syndrome, a mitochondrial cytopathy was unlikely because there was no progressive external ophthalmoplegia or cardiac conduction defect [9]. Bardet-Biedl syndrome was excluded because obesity, polydactyly, renal anomaly and hypogonadism were absent [10]. Refsum disease was also unlikely because there was no cerebellar ataxia, peripheral neuropathy, or arrhythmia [6,7]. Therefore, the clinical profile did not fulfil criteria for any established multisystem syndrome linking RP and cardiac disease.

Similar and atypical associations of RP have been described in various literature. Lobo S et al., in a case series, reported unusual associations such as posterior pole revascularisation, central serous chorioretinopathy and proliferative diabetic retinopathy in patients with RP [11]. These findings are atypical because central visual loss in RP is more commonly attributed to posterior subcapsular cataract, cystoid macular oedema, epiretinal membrane, or macular hole formation [11]. Another case series presented by Shivani M et al., reported RP with ocular and systemic associations, further supporting the need for complete ocular and systemic screening in patients with inherited retinal dystrophy [12]. These reports highlight that RP should not be viewed only as an isolated retinal disorder, especially when there are severe visual impairment, consanguinity, or additional systemic findings.

The present case differs from these previously described atypical associations. The present patient did not have diabetic retinopathy, cystoid macular oedema, cataract-related visual loss, keratoconus, glaucoma, hearing loss, polydactyly, obesity, renal abnormality, neuropathy, ataxia, or ophthalmoplegia. Instead, the additional major findings were an ostium secundum ASD with right-sided chamber dilatation and pulmonary arterial hypertension. This makes the present case clinically distinct, as the co-existence of RP with a congenital interatrial ASD, in the absence of a recognised syndrome, appears to be rare. The association may be coincidental; however, the strong consanguineous background and familial clustering of RP raise the possibility of an unidentified shared genetic or developmental pathway.

A possible biological link between congenital cardiac septation defects and retinal dystrophy may involve genes with pleiotropic developmental roles. Mutations affecting cardiac transcription factors such as NKX2-5, GATA4 and TBX5 are known to be associated with CHD, including septal defects [13]. Inherited retinal dystrophies, including RP, are also genetically heterogeneous and may involve genes related to photoreceptor structure, ciliary function and retinal maintenance [5,14]. Although no single established syndrome was identified in the present case, the coexistence of familial RP and ASD supports the need for molecular evaluation. Whole-exome sequencing would help clarify whether this represents a coincidental association or a previously unrecognised genetic link.

The transient neurological event in the present patient is also clinically relevant. During admission, she developed a transient episode of left upper limb weakness that resolved spontaneously within minutes, consistent with a TIA. MRI of the brain did not show acute infarct or haemorrhage, while MRA showed a hypoplastic right vertebral artery. In patients with an interatrial communication, paradoxical embolism is a recognised mechanism for TIA events, particularly when transient right-to-left flow occurs during pressure changes [3,15,16]. Although the ECHO in this case demonstrated a left-to-right shunt, the presence of an unrepaired ASD makes cardiology follow-up important for assessment of closure suitability and prevention of future complications.

From a management perspective, this case emphasises the importance of multidisciplinary evaluation. The cardiac lesion requires formal cardiology assessment for device closure or surgical repair, considering the defect size, right-sided chamber dilatation, pulmonary artery dilatation and PASP [1,3]. The retinal disease requires ophthalmology follow-up, low-vision rehabilitation, surveillance for treatable complications such as cataract and cystoid macular oedema and counselling regarding the progressive nature of the disease [5]. Normal audiological findings helped exclude Usher syndrome, while neurological evaluation-guided treatment of the TIA event. Therefore, the novelty of the present case lies not merely in reporting two rare coexisting diagnoses but demonstrating the value of systematic multispecialty evaluation in an adult with CHD, familial retinal dystrophy, consanguinity and TIA.

CONCLUSION(S)

The authors report a rare adult case of ostium secundum ASD co-existing with bilateral RP in a 52-year-old female with a consanguineous family history of progressive visual loss, T2DM and a complicating TIA, without meeting criteria for any established multisystem syndrome. Systematic multidisciplinary evaluation and comprehensive audiological testing (PTA 15 dBHL bilaterally) effectively excluded all known syndromic associations. The co-occurrence of ASD and RP in this non syndromic context may reflect shared but as-yet-uncharacterised genetic pathways governing both cardiac septation and retinal photoreceptor development. Whole-exome sequencing is recommended. Clinicians should maintain a low threshold for ophthalmological and audiological assessment in adults diagnosed with CHD, particularly when a positive family history of visual loss or consanguinity is identified.

Authors' contribution: SK: Case identification, data collection, laboratory data compilation, literature review, manuscript preparation and drafting. VR: Clinical supervision, Echocardiography interpretation support, clinical inputs, conceptualisation, critical revision of the manuscript and final approval. All authors have read and approved the final manuscript.

Ethical statement: This case report was conducted in accordance with the ethical principles of the Declaration of Helsinki. Written informed consent was obtained from the patient for publication of the present case report and the accompanying clinical images. Patient identity has been de-identified in accordance with institutional policy. Ethical clearance for case report publication was obtained from the Institutional Ethics Committee, Mahatma Gandhi Medical College and Research Institute Hospital, Pondicherry (Reference: IEC/MGMCRI/CR/2025). No experimental procedures were performed; all investigations and treatments were carried out as part of routine clinical care.

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PARTICULARS OF CONTRIBUTORS:
1. Postgraduate Student, Department of General Medicine, Mahatma Gandhi Medical College and Research Institute, Puducherry, India.
2. Assistant Professor, Department of General Medicine, Mahatma Gandhi Medical College and Research Institute, Sri Balaji Vidyapeeth (Deemed to be University), Puducherry, India.

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:
Dr. Vignessh Raveekumaran,
Assistant Professor, Mahatma Gandhi Medical College and Research Institute,
Sri Balaji Vidyapeeth (Deemed to be University), Puducherry-607402, India.
E-mail: vignesshravee@gmail.com

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