

A novel RYR2 variant associated with catecholaminergic polymorphic ventricular tachycardia requiring extracorporeal cardiopulmonary resuscitation and complicated by cervical vascular injury: a case report

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Case Report

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Abstract

Background

Catecholaminergic polymorphic ventricular tachycardia (CPVT) is an inherited arrhythmia syndrome that can cause sudden cardiac arrest. Its recognition after resuscitation may be obscured by troponin elevation and transient ventricular dysfunction, resembling fulminant myocarditis. Extracorporeal cardiopulmonary resuscitation (ECPR) can provide temporary circulatory support during refractory cardiac arrest; however, cervical cannulation carries a risk of vascular thrombosis and bleeding. We report a case of a child with ryanodine receptor 2 (*RYR2*)-associated CPVT, complicated by a compressive cervical haematoma after decannulation.

Case presentation

A 9-year-old boy with two previous syncopal episodes sustained a startle-triggered out-of-hospital cardiac arrest and developed recurrent polymorphic ventricular tachycardia after admission to the paediatric intensive care unit. Right cervical veno-arterial extracorporeal membrane oxygenation was established after continuous cardiopulmonary resuscitation without sustained return of spontaneous circulation, with an estimated resuscitation-to-flow interval of approximately 60 minutes. Serum troponin I was measured at 5.11 µg/L and left ventricular ejection fraction was 42–44%, prompting treatment for suspected diagnosis of fulminant myocarditis; the ejection fraction subsequently fell to 33%. Whole-exome sequencing identified a novel heterozygous *RYR2* variant, c.11836G > A (p.Gly3946Ser), classified as pathogenic by the reporting laboratory according to ACMG/AMP criteria. The variant is absent from ClinVar, relevant population reference databases, and the published literature, and its identification, together with the patient's characteristic clinical phenotype, supported the diagnosis of *RYR2*-associated CPVT. During anticoagulation for cervical vascular thrombosis after decannulation, an enlarging haematoma with active extravasation and tracheal compression required surgical evacuation, carotid repair, jugular thrombectomy, and venorrhaphy. The child was discharged after 41 days with a significant improvement ejection fraction of 67%, persistent internal jugular occlusion, and propranolol and aspirin therapy.

Conclusions

CPVT should be reconsidered after an adrenergic cardiac arrest, even when post-arrest findings suggest myocarditis. ECPR can provide temporary rescue support. A rare *RYR2* variant detection may support the clinical diagnosis and intervention.

Background

Catecholaminergic polymorphic ventricular tachycardia (CPVT) is an inherited arrhythmia syndrome in which adrenergic stimulation precipitates bidirectional or polymorphic ventricular tachycardia, usually without structural heart disease [1–3]. Pathogenic *RYR2* variants disrupt cardiac calcium handling and account for most genetically confirmed cases of CPVT [1, 4]. Cardiac arrest may be the first recognised presentation, although exertional- or emotion-triggered syncope may precede the diagnosis [2, 5]. After resuscitation, serum troponin elevation and transient ventricular dysfunction are non-specific and may mimic fulminant myocarditis [6–9]. Recognition is important because CPVT requires antiadrenergic therapy and clinical and genetic evaluation of first-degree relatives [10–13]. Extracorporeal cardiopulmonary resuscitation (ECPR) can support selected children with refractory arrest; however, the timing and patient selection vary among centres [14, 15]. In addition, cervical cannulation may complicate carotid or internal jugular thrombosis and bleeding, for which surveillance and reconstruction strategies remain heterogeneous [16, 17].

To date, paediatric experience with CPVT requiring extracorporeal support remains limited, with the available evidence largely restricted to isolated case reports and small case series [18–22]. In these reports, extracorporeal membrane oxygenation (ECMO) was used mainly as a temporary rescue strategy to provide haemodynamic and cardiopulmonary support while ventricular arrhythmias were controlled or the underlying diagnosis was established [18–22]. Furthermore, published paediatric cases of CPVT associated with rare *RYR2* variants remain scarce, highlighting the limited clinical experience with this combination [18–22]. Herein, we report a case of *RYR2*-associated CPVT, complicated by cervical vascular thrombosis and active bleeding after decannulation, and discuss the rarity and clinical implications of the *RYR2* variant.

Case presentation

Chief complaints and history of illness

A 9-year-old boy was referred after suddenly collapsing, becoming cyanotic and apnoeic, and being found pulseless. Before admission, he was abruptly startled by a rat running past him at home, became cyanotic and apnoeic within seconds, and was detected pulseless. Family members initiated cardiopulmonary resuscitation (CPR). At a provincial hospital, he was intubated and resuscitation continued, with an estimated pre-hospital resuscitation interval of approximately 30 minutes before the return of cardiac activity. The initial rhythm, no-flow interval, defibrillation details, and exact low-flow duration were not documented. He was transferred directly to our paediatric intensive care unit (PICU) while receiving vasopressor support.

Personal and family history

He was the second child of non-consanguineous parents, with no family history of premature sudden cardiac death, unexplained drowning, recurrent syncope, or known inherited arrhythmia syndromes.

Parental testing for the identified *RYR2* variant was not performed; therefore, its inheritance status remained undetermined. The admission records consistently documented two previous episodes of sudden syncope, although the available documentation was inconsistent regarding whether cardiopulmonary resuscitation had been required or whether spontaneous recovery had occurred. Prior electrocardiographic and echocardiographic evaluations were reportedly normal, and no antiarrhythmic therapy had been initiated.

Physical examination and initial investigation

On admission, the patient weighed 25 kg and had impaired consciousness, prolonged capillary refill, and hypotension (70/40 mmHg) despite vasopressor support with noradrenaline, adrenaline, and milrinone. Bilateral crackles and blood-stained endotracheal secretions were noted, and the abdominal examination was unremarkable.

A bedside 12-lead electrocardiogram demonstrated sinus rhythm with narrow QRS complexes, without apparent baseline QT prolongation, diagnostic repolarisation abnormalities, or acute ST-segment changes (**Additional File 1, Figure S1**). Continuous bedside electrocardiographic monitoring documented recurrent polymorphic ventricular tachycardia (**Additional File 1, Figure S2**). Subsequent Holter monitoring revealed frequent multiform ventricular ectopy and recurrent episodes of nonsustained ventricular tachycardia, with occasional brief episodes of supraventricular tachycardia, indicating a substantial ventricular arrhythmic burden, while the corrected QT interval remained within the reported range of 400–440 ms (Fig. 1).

Given the recurrent haemodynamic instability and ventricular arrhythmia, preparation for venoarterial extracorporeal membrane oxygenation (VA-ECMO) via right cervical cannulation began approximately 25 minutes after admission. Approximately 45 minutes after admission, the child developed refractory cardiac arrest. CPR was continued throughout cannulation, and sustained return of spontaneous circulation was not achieved. Two unsynchronised shocks at 4 J/kg, together with adrenaline, did not prevent recurrent polymorphic ventricular tachycardia. Extracorporeal flow was established approximately 1 hour and 45 minutes after admission, corresponding to an estimated interval of approximately 60 minutes from initiation of CPR to establishment of extracorporeal flow.

Further investigations

Cardiac troponin I was measured at 5.11 µg/L at admission (reference value < 0.07 µg/L), increased to 11.78 µg/L the next day, and declined to 0.32 µg/L in the second week. The arterial pH was 7.196, with a serum lactate level of 11.6 mmol/L. The first anti-factor Xa activity was 0.38 IU/mL, sampled approximately 4.5 hours after the scheduled enoxaparin dose. Blood and respiratory cultures and multiplex polymerase chain reaction (PCR) did not detect any pathogens.

Whole-exome sequencing (WES) of DNA from a buccal swab identified a heterozygous *RYR2* variant (NM_001035.3:c.11836G > A; NP_001026.2:p.Gly3946Ser), classified as pathogenic by the reporting laboratory using ACMG/AMP criteria [23]. ClinVar database independently listed the same variant as pathogenic in relation to catecholaminergic polymorphic ventricular tachycardia, with concordant assessments from multiple submitters and no classification conflict (Variation ID 201315; accessed 13 August 2026). The WES report also detected *SLC4A3* c.2071C > T (p.Arg691Cys) and *F8* c.3279G > A (p.Met1093Ile), both variants of uncertain significance confirmed by Sanger sequencing (Table 1). Additional copy number analysis revealed no abnormalities within the assay limits.

Table 1
Genetic variants identified by whole-exome sequencing and clinical relevance.

Gene (inheritance)	Variant	Zygosity	Classification and relevance
<i>RYR2</i> (autosomal dominant)	NM_001035.3:c.11836G > A; NP_001026.2:p.Gly3946Ser	Heterozygous	Pathogenic by the reporting laboratory and ClinVar (Variation ID 201315; multiple submitters, no conflicts). Phenotype concordant with CPVT. Absent from the Genome Aggregation Database (gnomAD).
<i>SLC4A3</i>	NM_201574.3:c.2071C > T; p.Arg691Cys	Heterozygous	Variant of uncertain significance (Sanger-confirmed); not used to establish the diagnosis or guide treatment.
<i>F8</i>	NM_000132.4:c.3279G > A; p.Met1093Ile	Hemizygous	Variant of uncertain significance (Sanger-confirmed); not used to explain bleeding or guide treatment.
Notes. CPVT: catecholaminergic polymorphic ventricular tachycardia. Variant nomenclature follows the Human Genome Variation Society recommendations for the GRCh38 reference assembly. ClinVar was accessed on 22 August 2026.			

Transthoracic echocardiography performed at admission showed global hypokinesia, a left ventricular ejection fraction (LVEF) of 42–44%, and no pericardial effusion. The LVEF decreased to 33% during extracorporeal support before it recovered. No structural cardiac abnormalities were detected, and the resting corrected QT intervals were 420–425 ms. Brain magnetic resonance imaging (MRI) after decannulation revealed symmetrical restricted diffusion and T2/FLAIR hyperintensity in the caudate nuclei and putamina, consistent with hypoxic-ischaemic injury (Fig. 2).

The patient’s medical history, clinical examination findings, investigations, and treatments are summarised chronologically in **Additional File 2**.

Diagnosis

The main diagnosis was *RYR2*-associated CPVT presenting with out-of-hospital cardiac arrest, precipitated by the startle trigger, two previously reported syncopal episodes, recurrent polymorphic ventricular tachycardia, no structural cardiac lesions on echocardiography, and the pathogenic *RYR2* variant. Additional diagnoses included transient post-arrest left ventricular dysfunction, bilateral basal ganglia hypoxic-ischaemic injury, and post-decannulation right internal jugular vein (RIJV) thrombosis with an expanding cervical haematoma. Fulminant myocarditis remained an unconfirmed differential diagnosis, as it could not be excluded without cardiac MRI or endomyocardial biopsy, neither of which was performed [6–9].

Treatment

Extracorporeal cardiopulmonary resuscitation was initiated through right cervical cannulation with a 19 Fr venous cannula advanced to the right atrium and a 14 Fr arterial cannula directed towards the aortic arch. The initial VA-ECMO settings were 2800 rpm, blood flow 2.0 L/min (80 mL/kg/min), sweep gas flow 4 L/min, and FiO₂ 100%; circuit temperature increased from 35°C to 37°C over the admission. Activated clotting time was monitored hourly, and continuous veno-venous haemodiafiltration was connected in series. Noradrenaline and adrenaline were infused at 0.6 µg/kg/min with milrinone 1.0 µg/kg/min; additional adrenaline boluses were administered during arrest before CPVT was suspected, and the patient was subsequently weaned from these agents on extracorporeal support. Amiodarone (5 µg/kg/min) and lidocaine (25 mg bolus, then 50 µg/kg/min) were administered, and intravenous immunoglobulin (2 g/kg) was administered for suspected fulminant myocarditis. Empirical antimicrobials were adjusted during recurrent fever and discontinued after negative microbiological investigations.

VA-ECMO was discontinued approximately 6 days after admission, with primary repair of the right carotid artery and RIJV at decannulation. The child was extubated approximately 10 days after admission. After CPVT was recognised, propranolol was started at 1.6 mg/kg/day, temporarily withheld for bradycardia (40–44 beats/min), and resumed at 0.8 mg/kg/day. Post-decannulation Doppler ultrasonography revealed non-occlusive thrombi in the right common carotid artery and RIJV; hence, therapeutic enoxaparin was initiated and titrated to anti-factor Xa activity. The carotid thrombus resolved, whereas the RIJV thrombosis progressed to complete occlusion, and a progressively enlarging cervical swelling developed. Computed tomography angiography revealed a 40 × 33 × 46 mm haematoma with active extravasation, complete RIJV occlusion, and compression of the carotid artery and upper trachea (Fig. 3). Enoxaparin was withheld, and urgent surgical re-exploration was performed. The haematoma was evacuated, a small actively bleeding defect adjacent to the intact carotid arteriorrhaphy was repaired, an organised RIJV thrombus was removed, and the vein was reconstructed by primary venorrhaphy. Enoxaparin was readministered but discontinued after recurrent complete RIJV occlusion and enlargement of the residual cervical mass. At discharge, oral aspirin 81 mg once daily was prescribed to maintain patency of the repaired carotid artery, rather than as treatment for the right internal jugular vein thrombosis.

Outcome and follow-up

The patient's left ventricular ejection fraction improved to 61% shortly after ECMO decannulation and further increased to 67% by hospital discharge. At discharge, the child was alert and interactive, followed commands appropriately, and demonstrated spontaneous movement of all four limbs, without focal neurological deficits. Although brain MRI showed bilateral basal ganglia abnormalities consistent with hypoxic ischaemic injury (Fig. 2), no clinically apparent neurological deficit was documented at discharge. The repaired right carotid artery remained patent. Partial recanalization of the right internal jugular vein was initially observed but was followed by complete re-occlusion. After a 41-day hospitalization, the patient was discharged in sinus rhythm, breathing spontaneously on room air, with a residual 40-mm cervical mass. Discharge medications included oral propranolol 10 mg twice daily and aspirin 81 mg once daily. Vascular follow-up was scheduled for one week after discharge. At the one-month post-discharge follow-up, cardiac rhythm (on ECG), vascular examination, and neurodevelopmental assessment revealed no clinically apparent abnormalities.

Discussion

This case report describes a rare presentation of previously unrecognised catecholaminergic polymorphic ventricular tachycardia (CPVT) in a 9-year-old boy who developed refractory polymorphic ventricular tachycardia (VT) and cardiac arrest following a sudden startle event. Despite initial resuscitation and conventional antiarrhythmic therapy, recurrent life-threatening ventricular arrhythmias prompted further investigation into whole-exome analysis revealing an underlying inherited arrhythmia syndrome.

The diagnosis of catecholaminergic polymorphic ventricular tachycardia (CPVT) was supported by the close concordance between the clinical phenotype and genetic findings. The patient had a history of recurrent unexplained syncope, a startle-triggered cardiac arrest, recurrent adrenergically mediated polymorphic/bidirectional ventricular tachyarrhythmias, and no evidence of underlying structural heart disease, all of which are characteristic features of CPVT [1–5, 13]. Genetic testing identified a heterozygous *RYR2* c.11836G > A (p.Gly3946Ser) variant (Table 1). Notably, the *RYR2* encodes the cardiac ryanodine receptor, the principal sarcoplasmic-reticulum calcium-release channel involved in excitation–contraction coupling. Pathogenic *RYR2* variants can promote abnormal diastolic calcium release, delayed after depolarisations, and catecholamine-dependent ventricular arrhythmogenesis [1, 4]. The p.Gly3946Ser variant was classified as pathogenic for CPVT by the reporting laboratory according to ACMG/AMP criteria and has concordant pathogenic classifications from multiple ClinVar submitters without conflicting interpretations (ClinVar Variation ID 201315) [23]. It was also absent from both the exome and genome datasets of the Genome Aggregation Database (gnomAD), providing supporting evidence of extreme population rarity [24, 25]. However, absence from a population database alone is insufficient to establish pathogenicity and should be interpreted in conjunction with phenotype–genotype concordance, segregation, functional, computational, and other variant-level evidence. Furthermore, precise population prevalence cannot be inferred without locus-specific allele-count and

callability data. In this patient, the characteristic CPVT phenotype together with the pathogenic *RYR2* finding strongly supported CPVT as the primary arrhythmic substrate. This distinction was clinically important because severe post-resuscitation myocardial injury and transient ventricular dysfunction could initially mimic fulminant myocarditis. Accordingly, inherited arrhythmia syndromes should be considered in children presenting with otherwise unexplained polymorphic or bidirectional ventricular tachycardia, syncope, or cardiac arrest precipitated by emotional or physical stress, particularly when structural cardiac abnormalities are absent. Establishing the diagnosis informed subsequent management, including non-selective β -blockade, avoidance of adrenergic triggers, and consideration of cascade genetic testing for first-degree relatives [10–13].

To date, published paediatric experience with extracorporeal circulatory support for catecholaminergic polymorphic ventricular tachycardia (CPVT) remains limited to isolated case reports and small case series (Table 2) [18–22]. Cafferkey and McMahon reported a 12-year-old boy with refractory pulseless polymorphic ventricular tachycardia who was treated with intravenous propranolol followed by esmolol and required four days of extracorporeal life support before undergoing left cardiac sympathetic denervation and implantable cardioverter-defibrillator (ICD) implantation [18]. Bellamy *et al.* subsequently described three children with CPVT presenting with recurrent ventricular tachycardia or ventricular fibrillation. Two required extracorporeal membrane oxygenation (ECMO) after prolonged electrical and haemodynamic instability in the setting of repeated epinephrine administration, whereas the third was stabilised without extracorporeal support after earlier recognition of CPVT and avoidance of further epinephrine exposure [19]. Additional reports have similarly described ECMO as rescue circulatory support in severe CPVT presentations. Li *et al.* reported a 4-year-old boy with refractory ventricular arrhythmias, cardiogenic shock, and severely impaired left ventricular systolic function (LVEF 23%) who was successfully weaned from veno-arterial ECMO (VA-ECMO) after 10 days [20]. Song *et al.* described an 18-month-old boy who developed cardiac arrest following minor trauma, required VA-ECMO support, and was subsequently diagnosed with *RYR2*-associated CPVT [21]. By contrast, Sadagopan and Yue reported a patient in whom ECMO was made available as procedural backup during electrophysiological testing but ultimately was not required [22]. Collectively, these reports indicate that extracorporeal support has been used primarily as a temporary rescue strategy to maintain systemic perfusion while facilitating haemodynamic stabilisation, arrhythmia control, diagnostic clarification, and transition to definitive antiarrhythmic or interventional therapy. However, the available evidence is entirely observational and is derived from a very small number of clinically heterogeneous cases. Consequently, no conclusions can be drawn regarding the efficacy, optimal timing, duration, or patient-selection criteria for extracorporeal cardiopulmonary resuscitation (ECPR) or ECMO in CPVT.

Table 2

Published paediatric cases of catecholaminergic polymorphic ventricular tachycardia involving extracorporeal life support and comparison with the present case.

Reports	Patient and trigger	Acute course and extracorporeal strategy	Diagnostic evidence and outcome
Cafferkey and McMahon, 2018 [18]	12-year-old boy	Refractory pulseless polymorphic VT; IV propranolol then esmolol; ECLS 4 days	Clinical CPVT diagnosis; LCSD then ICD; arrhythmia-free on atenolol at follow-up
Bellamy <i>et al</i> , 2019 [19]	Three children, aged 4, 5, 10 years; activity/immersion/running-related events	Recurrent VT/VF; two received ECMO, one stabilised without it after early recognition	De novo <i>RYR2</i> variants in all; beta-blocker plus flecainide and ICD; no recurrent arrest over follow-up
Li <i>et al</i> , 2025 [20]	4-year-old boy; recurrent syncope before collapse	Bidirectional VT/VF, cardiogenic shock, LVEF 23%; femoral VA-ECMO 10 days	De novo <i>RYR2</i> variant; recurrent VT despite metoprolol; later ICD; asymptomatic at 1 year
Song <i>et al</i> , 2026 [21]	18-month-old boy; arrest after minor fall	Post-ROSC heart failure and polymorphic VT; VA-ECMO	De novo missense <i>RYR2</i> variant; propranolol; no recurrent arrhythmia reported
Sadagopan and Yue, 2023 [22]	9-year-old girl; previous events labelled epilepsy	Cardiorespiratory arrest; ECMO standby for EPS, not deployed	Isoproterenol-induced VT; CPVT gene panel negative; arrhythmia-free at 4 months
Present case	9-year-old boy; sudden fright; two previous syncopal episodes	Recurrent polymorphic VT then refractory arrest; continuous CPR without sustained ROSC; right cervical ECPR/VA-ECMO 6 days; CPR-to-flow ~ 60 min	Pathogenic <i>RYR2</i> p.Gly3946Ser, absent from gnomAD; cervical thrombosis and active bleeding required vascular surgery; favourable outcome at discharge
Notes. CPVT, catecholaminergic polymorphic ventricular tachycardia; ECMO, extracorporeal membrane oxygenation; EPS, electrophysiological study; ICD, implantable cardioverter-defibrillator; LCSD, left cardiac sympathetic denervation; LVEF, left ventricular ejection fraction; ROSC, return of spontaneous circulation; VA-ECMO, venoarterial extracorporeal membrane oxygenation; VF, ventricular fibrillation; VT, ventricular tachycardia.			

The principal acute management challenge arose before catecholaminergic polymorphic ventricular tachycardia had been recognised. During profound circulatory collapse and subsequent cardiac arrest, adrenaline and other vasoactive agents were administered according to standard resuscitation priorities. Given the retrospective nature of this report, it is not possible to determine whether catecholamine exposure initiated, amplified, or prolonged the ventricular arrhythmias observed in this patient.

Nevertheless, previous paediatric reports provide biological and clinical support for catecholamine sensitivity in CPVT. Bellamy *et al.* described worsening ventricular arrhythmias during epinephrine exposure, including induction of polymorphic ventricular tachycardia during an epinephrine challenge while the patient was supported with extracorporeal membrane oxygenation [19], whereas Cafferkey and McMahon reported successful suppression of refractory ventricular arrhythmias with intravenous non-selective β -blockade during extracorporeal life support [18]. These observations are consistent with the adrenergically mediated pathophysiology of CPVT but, because they are derived from uncontrolled reports, do not establish causality or comparative treatment efficacy. Clinically, recurrent polymorphic or bidirectional ventricular arrhythmias precipitated by exercise, emotional stress, or sudden startle should therefore raise suspicion for CPVT, prompting minimisation of avoidable adrenergic stimulation and initiation of non-selective β -blockade once haemodynamic conditions permit [10, 12]. Notably, extracorporeal cardiopulmonary resuscitation (ECPR) should similarly be regarded as a rescue strategy for refractory cardiac arrest rather than as evidence of treatment efficacy in an individual patient. A meta-analysis of 28 observational paediatric studies comprising 1,348 patients undergoing ECPR reported a pooled survival rate of 46% (95% CI 43–48%) and survival with favourable neurological outcome of 30% (95% CI 27–33%) [26]. However, these estimates are derived from heterogeneous observational cohorts and cannot establish the causal benefit of ECPR or predict outcomes for an individual child. In the present case, preparation for cervical VA-ECMO cannulation began approximately 25 minutes after admission, refractory cardiac arrest occurred at approximately 45 minutes, and extracorporeal flow was established approximately 1 hour 45 minutes after admission. This indicated an estimated interval of approximately 60 minutes from initiation of CPR to establishment of extracorporeal flow. Given that CPR quality, coronary and cerebral perfusion, and the effectiveness of interim spontaneous circulation were not continuously quantified, this interval should not be interpreted as a precisely measured low-flow duration. Accordingly, the favourable neurological recovery observed in this patient neither validates prolonged CPR-to-flow intervals nor establishes that recovery was directly attributable to ECPR [14, 15]. Following recognition of CPVT, secondary-prevention management was initiated before discharge. Non-selective β -blockade was commenced; however, propranolol could be titrated only to 0.8 mg/kg/day because of treatment-associated bradycardia. Although this approach is consistent with guideline-based management, the adequacy of arrhythmia suppression could not be fully assessed because the follow-up Holter recording lasted less than 24 hours and did not systematically evaluate arrhythmic burden during exercise or emotional stress. The preference for non-selective β -blockade is supported by an international cohort of 329 children with CPVT, in which β 1-selective β -blocker therapy was associated with a higher risk of arrhythmic events than non-selective β -blockade [10, 27]. At discharge, several components of longer-term risk stratification and secondary prevention had not yet been completed or formally documented, including exercise or catecholamine provocation testing, assessment of the need for adjunctive flecainide, consideration of left cardiac sympathetic denervation or implantable cardioverter-defibrillator therapy, structured counselling regarding exercise and adrenergic triggers, development of an emergency action plan, and cascade genetic testing of first-degree relatives [10, 28, 29]. These unresolved issues emphasise the importance of close follow-up in a specialist inherited-

arrhythmia service to assess treatment response, optimise antiarrhythmic therapy, refine individual risk stratification, and facilitate family screening and genetic counselling.

Notably, the cervical vascular complication observed in this case illustrates the competing risks of thrombosis and bleeding during and after the ECMO. In a prospective cohort of 514 children receiving ECMO, bleeding occurred in 70.2% and patient-related thrombosis in 12.8%, although post-decannulation cervical vascular complications were not analysed separately [16]. In addition, a recent literature review of paediatric vascular reconstruction after cervical ECMO cannulation identified 18 studies and highlighted substantial variability in vascular reconstruction strategies and postoperative surveillance. The largest reported series included 51 patients, with postoperative arterial occlusion rates ranging from 11.8% to 17.8% and an overall postoperative vessel patency rate of 78.6% [17]. These findings underscore the importance of careful vascular assessment and follow-up after ECMO decannulation, particularly when cervical cannulation has been performed. In the present case, non-occlusive thrombi involving the carotid artery and internal jugular vein developed within 24 hours of vascular repair. The carotid thrombus subsequently resolved, but the internal jugular thrombus progressed to complete occlusion. The patient subsequently developed an enlarging cervical haematoma with active contrast extravasation adjacent to the arterial repair site. The coexistence of progressive venous thrombosis and active arterial bleeding represents a more complex post-decannulation vascular complication than isolated cannula-associated venous thrombosis alone. This posed a particular therapeutic challenge because escalation of anticoagulation may aggravate ongoing haemorrhage [30–32]. Hence, the combination of progressive neck swelling and anticoagulation should prompt urgent vascular imaging to assess thrombosis, active bleeding, or both. When active bleeding or an expanding haematoma is identified, urgent multidisciplinary assessment and prompt surgical or other haemostatic intervention may be required rather than anticoagulation therapy alone. This case highlights the importance of maintaining a high index of suspicion for cervical vascular injury after decannulation, particularly when neck swelling is progressive or associated with bleeding, airway compression, unexplained anaemia, or haemodynamic deterioration.

In addition, a nationwide study of pregnant women in Vietnam receiving routine prenatal care reported that 450 of 3,259 women (13.8%) were carriers of disease-associated variants in *G6PD*, *PAH*, and *GALT*, which are associated with glucose-6-phosphate dehydrogenase (G6PD) deficiency, phenylketonuria, and galactosemia, respectively [33]. Hence, these findings support the potential utility of incorporating genetic carrier screening into routine prenatal care programs. In the present case, there was no documented evidence of parental or familial screening for the *RYR2* variant. This highlights the importance of considering parental and familial genetic evaluations when a potentially pathogenic variant is identified. The integration of prenatal genetic screening with confirmatory molecular testing and appropriate cascade family screening may facilitate the early identification of at-risk individuals and enable timely clinical surveillance and intervention, potentially reducing the burden of inherited diseases.

The strengths of this report include the identification of an *RYR2* variant concordant with the clinical phenotype, together with serial documentation of cardiac rhythm, ventricular function, vascular

complications, and major therapeutic interventions during the index admission. However, several limitations should be acknowledged. The precise duration and quality of CPR, including the low-flow interval before extracorporeal support, could not be fully quantified. In this presented patient, cardiac magnetic resonance imaging and endomyocardial biopsy were not conducted, limiting definitive exclusion of myocarditis. Rhythm surveillance was incomplete, and there was no documented evidence of parental segregation testing or systematic family screening for the *RYR2* variant. As a single case, this report cannot establish causal relationships between clinical outcomes and ECPR, catecholamine exposure, antiarrhythmic therapy, or anticoagulation, nor can it determine the efficacy or safety of these interventions in children with CPVT.

In conclusion, startle-triggered cardiac arrest, recurrent syncope, and polymorphic or bidirectional ventricular tachycardia should raise suspicion for catecholaminergic polymorphic ventricular tachycardia (CPVT), even when post-arrest myocardial injury initially suggests an alternative diagnosis. Identification of a rare *RYR2* variant may support the diagnosis when interpreted within established ACMG/AMP criteria and in conjunction with the clinical phenotype; rarity alone is insufficient to establish pathogenicity. ECPR may provide temporary circulatory support in refractory cardiac arrest, but its efficacy cannot be inferred from individual cases. Progressive neck swelling after cervical decannulation warrants urgent vascular assessment because thrombosis and active bleeding may coexist and require prompt intervention.

Abbreviations

ACMG/AMP

American College of Medical Genetics and Genomics/Association for Molecular Pathology

CPR

Cardiopulmonary resuscitation

CPVT

Catecholaminergic polymorphic ventricular tachycardia

CVVHDF

Continuous veno-venous haemodiafiltration

ECLS

Extracorporeal life support

ECMO

Extracorporeal membrane oxygenation

ECPR

Extracorporeal cardiopulmonary resuscitation

ICD

Implantable cardioverter-defibrillator

LVEF

Left ventricular ejection fraction

PICU

Paediatric intensive care unit
RIJV
Right internal jugular vein
ROSC
Return of spontaneous circulation
RYR2
Ryanodine receptor 2
VA-ECMO
Venoarterial extracorporeal membrane oxygenation
VF
Ventricular fibrillation
VT
Ventricular tachycardia
WES
Whole-exome sequencing.

Declarations

Clinical trial number

not applicable

Declarations

Ethics approval and consent to participate

Formal institutional ethics committee approval was not required for the publication of this single anonymised case report, in accordance with our institution's policy. The patient's legal guardian provided informed consent for all treatments delivered as standard clinical care.

Consent for publication

Written informed consent for publication of this case report and the accompanying clinical images was obtained from the patient's parent (or legal guardian). A copy of the signed written informed consent form is held by the corresponding author and will be made available for review by the Editor-in-Chief of this journal upon request. The completed consent form will not be submitted unless specifically requested by the journal.

Competing interests

The authors declare no competing interests.

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Author Contribution

Authors' contributions TTBK and NTT conceived and designed the study. NVD, PTH, HKG, DTTD, TMTD were involved in the direct clinical care of the patient and acquisition of clinical data. VTL, NDTV, DMC and NTT contributed to the data interpretation, literature review, and manuscript drafting. TTBK, TTN supervised the clinical course, interpreted the genetic findings, and critically revised the manuscript for important, intellectual content. All authors have read and approved the final manuscript.

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Availability of data and materials

The datasets generated and/or analysed during the current case are not publicly available to protect patient privacy, but de-identified data are available from the corresponding author upon reasonable request.

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Figures



Figure 1

Holter electrocardiographic recordings revealed frequent multi-form ventricular ectopy, ventricular couplets and recurrent runs of nonsustained ventricular tachycardia with alternating QRS morphology and axis, compatible with bidirectional or polymorphic ventricular tachycardia. The corrected QT interval remained within the reported range of 400–440 ms.

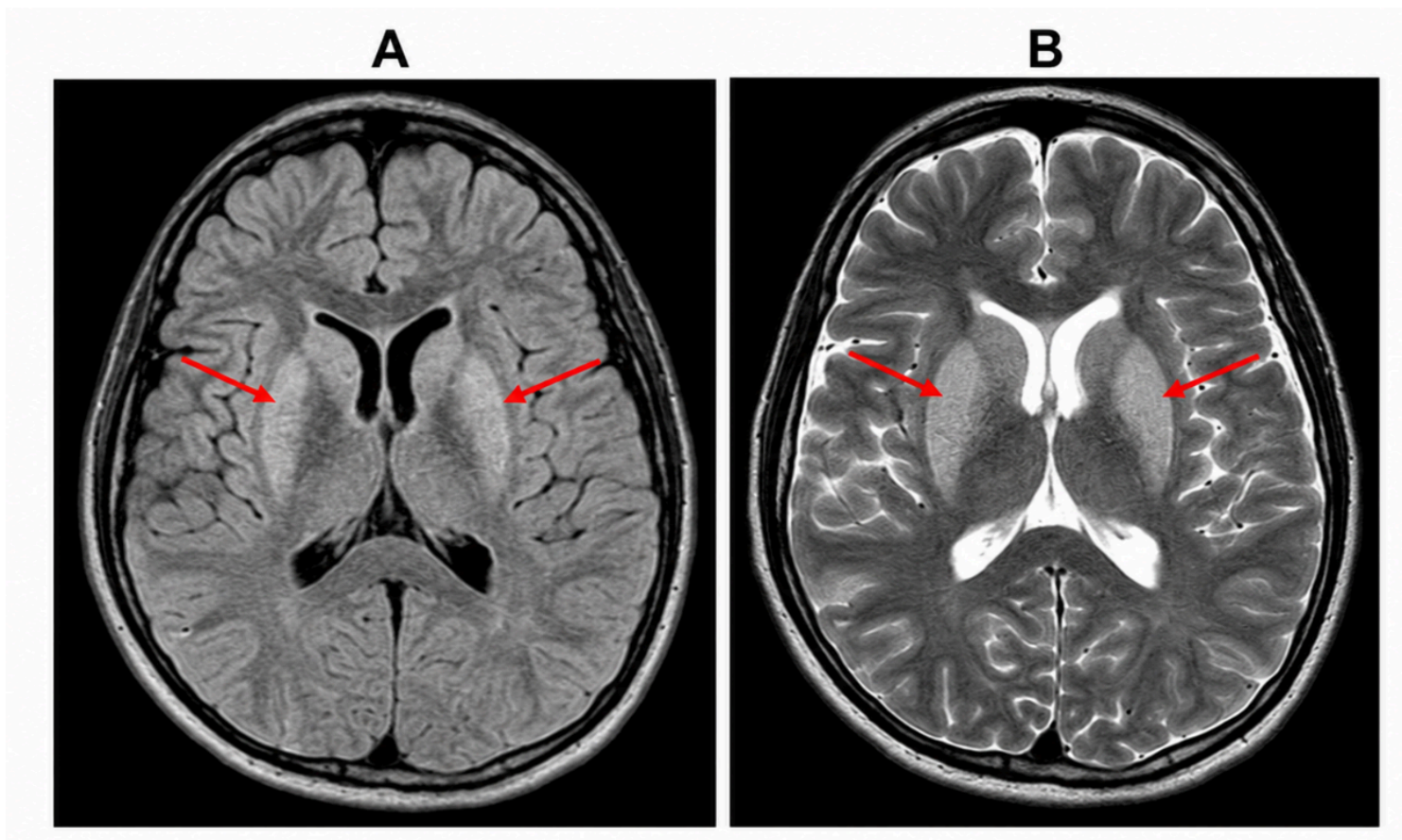


Figure 2

Brain magnetic resonance imaging was performed after decannulation. (A) Axial FLAIR image of the brain. (B) Axial T2-weighted image of the same patient. Both panels show symmetrical signal abnormalities involving the caudate nuclei and putamina (indicated by red arrows), consistent with hypoxic-ischaemic injury.



Figure 3

Contrast-enhanced computed tomography angiography of the neck showing post-decannulation vascular complications. (A) Axial image showing a 40 × 33 × 46 mm right cervical haematoma with active contrast extravasation, complete right internal jugular venous occlusion, and mass effect on the right common carotid artery and upper trachea. (B) Three-dimensional volume-rendered angiographic reconstruction demonstrating the cervical and great vessels and the region of vascular injury. Red arrows indicate cervical haematoma, active extravasation, venous occlusion, and associated vascular abnormalities.

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