ISSN: 2690-912X

Case Report

Journal of Genetic Engineering and Biotechnology Research

Video-Assisted Thoracoscopic Left Cardiac Sympathetic Denervation in a Young Child with Jervell and Lange-Nielsen Syndrome and a Family History of Sudden Infant Death: A Case Report

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Submitted: 2026, Sep 14; **Accepted:** 2026, Oct 26; **Published:** 2026, Nov 05

Citation: Rehman, S., Najeeb, K., Rashid. (2026). Video-Assisted Thoracoscopic Left Cardiac Sympathetic Denervation in a Young Child with Jervell and Lange-Nielsen Syndrome and a Family History of Sudden Infant Death: A Case Report. *J Gene Engg Bio Res, 8*(3), 01-05.

Abstract

Introduction: *Jervell and Lange-Nielsen syndrome (JLNS) is a rare autosomal recessive channelopathy caused by biallelic loss-of-function variants in KCNQ1 (or, less commonly, KCNE1), producing profound congenital sensorineural deafness together with a markedly prolonged QT interval and a very high risk of torsade's de pointes and sudden cardiac death from early childhood. Beta-blockade is first-line therapy, but a large proportion of patients remain symptomatic, and left cardiac sympathetic denervation (LCSD) is recommended as an adjunct in high-risk or breakthrough disease.*

Case Presentation: We describe a 4-year-1-month-old girl from rural Punjab, Pakistan, with genetically confirmed KCNQ1-related JLNS, who presented with recurrent seizure-like episodes from one year of age, profound bilateral sensorineural deafness with absent expressive speech, divergent squint, and a family history of a sibling who died at 5.5 months of age during an uncontrolled seizure/collapse. Despite oral propranolol and antiepileptic therapy, she remained at high arrhythmic risk on multidisciplinary review and underwent elective video-assisted thoracoscopic (VATS) left cardiac sympathetic denervation.

Clinical Discussion: *Baseline transthoracic echocardiography confirmed a structurally normal heart with preserved biventricular function, in keeping with a pure electrical (channelopathy) substrate. A dedicated perioperative pathway was used: tight target ranges for serum potassium (4.0–5.5 mmol/L) and magnesium (2.0–2.25 mg/dL), continuous ECG telemetry with defibrillator pads applied throughout the admission, bedside emergency antiarrhythmic drugs, and confirmed availability of pediatric thoracoscopic instrumentation and back-up temporary pacing/defibrillation. Surgery — resection of the lower third of the left stellate ganglion together with the upper thoracic sympathetic chain — was completed via a three-port VATS approach without intraoperative complication, inotropic support, or need for conversion.*

Conclusion: *This case illustrates that VATS-LCSD can be performed safely in a small child (13 kg) with high-risk JLNS when supported by careful multidisciplinary perioperative planning, and reinforces the importance of genetic counselling and cascade screening in families with unexplained infant deaths.*

Keywords: Jervell and Lange-Nielsen Syndrome, Long Qt Syndrome, Kcnq1, Left Cardiac Sympathetic Denervation, Video-Assisted Thoracoscopic Surgery, Sudden Cardiac Death, Pediatric Arrhythmia, Case Report

Highlights

- Jervell and Lange-Nielsen syndrome carries a substantially higher risk of cardiac events than isolated (Romano–Ward) long QT syndrome and often responds incompletely to beta-blockade alone.
- A family history of unexplained infant death with seizure-like

activity should prompt consideration of an underlying cardiac channelopathy, not epilepsy alone.

- VATS left cardiac sympathetic denervation is technically feasible and safe even in very young, low weight children when a structured, electrolyte-focused perioperative pathway is followed.
- Multidisciplinary planning — cardiology, cardiac surgery, anesthesia and nursing — with continuous monitoring and back-up defibrillation is central to safe outcomes in this high-risk population.

1. Introduction

Jervell and Lange-Nielsen syndrome (JLNS) was first described in 1957 in a Norwegian family with congenital deaf-mutism, a strikingly prolonged QT interval, and sudden death in childhood [1]. It is now understood to be an autosomal recessive channelopathy caused by homozygous or compound heterozygous pathogenic variants in KCNQ1 (approximately 90% of cases) or KCNE1, the genes encoding the two subunits of the slow delayed-rectifier potassium current (IKs) [2,3]. Because the same channel is expressed in both the cardiac myocyte and the stria vascularis of the inner ear, loss of IKs produces the syndrome's two defining features: profound bilateral sensorineural hearing loss present from birth, and a severely prolonged corrected QT interval (mean QTc in reported cohorts of approximately 557ms) [2]. JLNS is rare, with an estimated prevalence of roughly 1 in 200,000 to 1 in 1,000,000 live births, and is more frequent in populations with higher rates of consanguinity [3]. Its natural history is considerably more malignant than that of the autosomal dominant Romano–Ward form of long QT syndrome: in the largest published cohort, 86% of patients experienced at least one cardiac event, half were already symptomatic by three years of age, and 27% suffered cardiac arrest or sudden death despite beta-blocker therapy, which was found to have only partial efficacy [2]. These features place JLNS among the highest-risk long QT syndrome genotypes, for which international consensus guidelines identify left cardiac sympathetic denervation (LCSD) — and, in selected patients, implantable defibrillator therapy — as important adjuncts to beta-blockade [4]. LCSD reduces sympathetically mediated triggering of ventricular arrhythmia by resecting the lower part of the stellate ganglion (to minimise the risk of Horner syndrome) together with the upper thoracic sympathetic chain, typically from T2–T4 [4,5]. A large multicenter series demonstrated a fall in the proportion of long QT syndrome patients experiencing cardiac events from 99% to 45% after LCSD [5]. Increasingly, the procedure is performed thoracoscopically rather than through open supraclavicular or axillary incisions, which reduces morbidity and is feasible even in small children [6]. We report the case of a young child with genetically confirmed KCNQ1-related JLNS, a strong family history of sudden unexplained infant death, and incompletely controlled symptoms, who underwent elective VATS-LCSD, to illustrate the clinical recognition of this syndrome and the perioperative pathway used to manage it safely.

2. Case Presentation

2.1. History

The patient was a girl aged 4 years 1 month at the time of admission, from a rural district of Punjab, Pakistan, referred to a tertiary paediatric cardiology and cardiac surgery unit for consideration of cardiac sympathetic denervation. She was born at term by normal vaginal delivery following an uneventful pregnancy (a normal fetal anomaly scan was recorded, with no antenatal infection) and had an unremarkable neonatal course. Her early development was reported as normal until one year of age, when she began to have recurrent seizure like episodes. Neurological assessment at that time, including electroencephalography and brain magnetic resonance imaging, was reported as normal. Genetic testing performed in the course of this neurological work-up identified a pathogenic KCNQ1 variant, consistent with LQT1/JLNS when present in the homozygous or compound heterozygous state. She was started on antiepileptic therapy (sodium valproate and levetiracetam) and, following cardiology referral, oral propranolol, she was subsequently referred to a national cardiac centre and then to our institution specifically for evaluation for cardiac sympathetic denervation. Her family history was strongly relevant: she had three siblings (two brothers and one sister), and one brother had died at 5.5 months of age during an episode of uncontrolled seizure-like activity. In retrospect, and in view of the index patient's diagnosis, this raises the possibility that the deceased sibling may also have been affected by an unrecognized cardiac channelopathy rather than a primary seizure disorder, although this could not be confirmed. Vaccinations were up to date, and there was no history of other chronic illness.

2.2. Examination

On examination the child was alert, active and comfortable in room air, with a Glasgow Coma Scale score of 15/15. She had profound bilateral sensorineural hearing loss with no expressive speech, communicating only through vocalisations and gestures, and a divergent squint. Growth parameters were appropriate for age (weight approximately 12.8–13 kg, height approximately 96–100 cm). Vital signs on the initial cardiology assessment were blood pressure 109/55 mmHg, heart rate 69 beats per minute, respiratory rate within normal limits, and oxygen saturation 98% on room air, during the subsequent admission, heart rate ranged from 58 to 90 beats per minute (higher values coinciding with periods of distress) and oxygen saturation remained 96–100% on room air throughout. Cardiovascular examination revealed normal heart sounds without murmur, the chest showed equal bilateral air entry, the abdomen was soft and non-tender, and there was no skin rash or wound.

2.3. Investigations

Transthoracic echocardiography showed a structurally normal heart: normal aortic root and ascending aorta and arch (aortic valve annulus, root and sinotubular junction z-scores within normal range), a left-sided aortic arch with no coarctation and no patent ductus arteriosus, normal pulmonary arteries, and no pericardial effusion. Left ventricular systolic function was preserved

(fractional shortening 36%, ejection fraction 67% by the Teichholz method), with normal right ventricular function (tricuspid annular plane systolic excursion 20 mm) and normal chamber dimensions for body size (body surface area 0.6 m^2). These findings excluded structural congenital heart disease and were consistent with a pure electrical (ion-channel) substrate. Baseline laboratory investigations were unremarkable: haemoglobin 8.8 g/dL, total leukocyte count 6.3 $\times 10^9$/L, platelets 266 $\times 10^9$/L, prothrombin time 11.4 s (INR 1.09), activated partial thromboplastin time 26.2 s, urea 14 mg/dL, creatinine 0.3 mg/dL, sodium 140 mmol/L, potassium 4.7 mmol/L, chloride 105 mmol/L, corrected calcium 9.6 mg/dL and magnesium 2.0 mg/dL, hepatitis B surface antigen and anti-hepatitis C IgG were non-reactive. Serial electrocardiography during the admission was used to confirm the previously documented marked QT prolongation characteristic of her known diagnosis and to screen for any new ectopy, continuous telemetry was maintained throughout her stay.

2.4. Multidisciplinary Decision-Making

The case was discussed at a joint paediatric cardiology–cardiac surgery meeting. Given the JLNS phenotype recognised as conferring substantially higher arrhythmic risk than isolated long QT syndrome — incompletely controlled seizure-like activity despite antiepileptic therapy, and a family history of sudden death in infancy, she was classified as high perioperative and long-term risk. The multidisciplinary team elected to proceed with left (rather than bilateral) cardiac sympathetic denervation via VATS as an adjunct to continued beta-blocker therapy. Specific precautions agreed in advance included: maintaining serum potassium and magnesium within tight target ranges (potassium 4.0–5.5 mmol/L, magnesium 2.0–2.25 mg/dL) throughout the perioperative period, having back-up temporary pacing and percutaneous defibrillation immediately available, and confirming, before proceeding, the availability of paediatric-sized thoracoscopic instrumentation.

2.5. Perioperative Management

The patient was admitted electively two days before surgery. Home medications (propranolol and antiepileptic therapy) were continued under direct nursing supervision, and continuous cardiac monitoring with defibrillator pads applied was maintained from admission. A structured pre-operative checklist was completed, including confirmation of patient identity and consented procedure, medical history and allergy review, availability of cross-matched blood products, chlorhexidine skin preparation and nasal mupirocin application, weight-based prophylactic intravenous antibiotic (cefazolin), and a final surgical time-out. Written informed consent for the operation was obtained from the patient's father. All scheduled medications were cross-checked against a QT-drug safety reference list before administration, and emergency antiarrhythmic drugs (epinephrine, atropine, adenosine and amiodarone) together with airway equipment were kept at the bedside throughout the admission, consistent with a high-risk long QT syndrome perioperative pathway.

2.6. Operative Findings and Procedure

Surgery was performed on the second hospital day under general anesthesia, with the patient positioned in the lateral decubitus position. Three thoracoscopic port incisions of 2–3 mm (one camera port and two working ports) were made in a triangular configuration. The thoracic cavity was inspected, and the sympathetic chain and stellate ganglion were identified and visualised from the first to the fourth intercostal space. The lower one-third of the stellate ganglion was resected together with the sympathetic chain, which was divided at the level of the fifth intercostal space, in keeping with standard left cardiac sympathetic denervation technique. No significant intraoperative abnormal findings were noted, hemostasis was secured, the lung was re-expanded under direct vision, and the ports were removed and closed with 4-0 poliglecaprone (Monocryl) sutures. The patient did not require inotropic support, tolerated the procedure without arrhythmia, and was extubated in theatre before transfer to the paediatric cardiac intensive care unit in stable condition.

2.7. Postoperative Course and Outcome

The immediate postoperative course was uneventful. The patient remained hemodynamically stable on continuous ECG telemetry with defibrillator pads in place, without ventricular ectopy or arrhythmia, and did not require inotropic or ventilatory support beyond the perioperative period, postoperative echocardiography was not required. A mild reduction in serum magnesium was identified on routine postoperative bloods and was corrected with intravenous magnesium repletion in line with the pre-agreed electrolyte protocol. No wound complications, pneumothorax or clinical features of Horner syndrome (ptosis, miosis or facial anhidrosis) were documented. She was discharged on the following day tolerating oral intake, on her established regimen of propranolol and antiepileptic therapy, with cardiology and neurology follow-up arranged. Total inpatient stay was two days.

3. Discussion

This case highlights two clinically important themes: the recognition of JLNS as a distinct and unusually severe long QT syndrome subtype, and the safe conduct of thoracoscopic cardiac sympathetic denervation in a very young child.

First, JLNS should be actively considered whenever congenital sensorineural deafness coexists with syncope, seizure-like episodes, or a family history of sudden unexplained death, since seizure-like presentations of ventricular arrhythmia are easily misattributed to a primary neurological disorder, as very nearly occurred in this family. Both KCNQ1 and KCNE1 are inherited in an autosomal recessive fashion in JLNS, so parents are typically obligate heterozygous carriers who may themselves have a milder, or entirely absent, long QT phenotype [3]. This has two practical implications for the family described here: genetic counselling regarding the recurrence risk in future pregnancies, and cascade clinical and, where possible, genetic screening of surviving siblings and parents — particularly relevant given that a sibling of the index patient died in infancy during an episode that, in hindsight, may have represented an unrecognised cardiac event rather than isolated epilepsy.

Second, the natural history data available for JLNS indicate that beta-blockade alone is frequently insufficient: in the largest published series, just over half of patients had breakthrough cardiac events despite therapy, and more than a quarter suffered cardiac arrest or sudden death [2]. International consensus guidance therefore supports left cardiac sympathetic denervation as an important adjunctive therapy for high-risk long QT syndrome patients, including those with breakthrough symptoms on beta-blockers, those intolerant of adequate beta-blocker dosing, or as a bridge to or alternative for implantable defibrillator therapy in very young children in whom device-related morbidity is disproportionately high [4]. Mechanistically, LCSD reduces norepinephrine-mediated triggering of ventricular arrhythmia by removing the lower stellate ganglion and upper thoracic sympathetic outflow to the heart, and multicenter data show a marked reduction in cardiac events following the procedure, from 99% to 45% in one series, without abolishing risk altogether [5]. It is, therefore, understood as an adjunct to — not a replacement for — lifelong beta-blocker therapy, a point that was explicitly incorporated into this patient's discharge planning.

Thoracoscopic (VATS) LCSD, first described in children as an alternative to open supraclavicular or axillary approaches, offers reduced surgical morbidity, improved cosmesis and a shorter hospital stay, and has been shown to be feasible even in small children [6]. This patient, at 13 kg and four years of age, is illustrative of the lower end of the size range in which the procedure can be safely undertaken when supported by appropriate paediatric thoracoscopic instrumentation and an experienced surgical team. The perioperative pathway followed in this case — pre-emptive cross-checking of all medications against a QT-drug safety reference, tight electrolyte targets for potassium and magnesium, continuous ECG telemetry with defibrillator pads applied throughout the hospital stay, and bedside availability of emergency antiarrhythmic drugs — reflects accepted best practice for high-risk inherited arrhythmia patients undergoing any procedure under general anaesthesia, and arguably contributed to the uneventful course observed here.

Recognised complications of LCSD include Horner syndrome, from inadvertent injury to the upper (rather than lower) stellate ganglion, and compensatory facial or truncal sweating asymmetry, neither was observed in this patient. More recently, some high-volume centres have reported increasing use of bilateral, rather than unilateral, cardiac sympathetic denervation in selected high-risk patients, typically introduced in a staged fashion after LCSD when arrhythmic risk remains unacceptably high, this patient's genotype and clinical course will merit ongoing surveillance to determine whether such an escalation is ultimately required [7].
This report is limited by its retrospective, single-patient design and by gaps in some handwritten chart entries (for example, a precisely documented pre-operative QTc value was not available for direct citation here), which should be verified against the original record before any formal submission. Nonetheless, the case adds to a very small published experience of VATS-LCSD in JLNS specifically, as opposed to long QT syndrome more broadly, and underscores the value of a multidisciplinary, protocol-driven approach in this rare and high-risk paediatric population.

4. Conclusion

Jervell and Lange-Nielsen syndrome is a rare but highly malignant cause of long QT syndrome that should be suspected whenever congenital deafness accompanies syncope, seizure-like episodes, or a family history of sudden infant death. In this young child with genetically confirmed KCNQ1-related JLNS and ongoing symptoms despite beta-blocker and antiepileptic therapy, video-assisted thoracoscopic left cardiac sympathetic denervation was performed safely and without complication, supported by a structured multidisciplinary perioperative pathway with strict electrolyte control and continuous arrhythmia monitoring. The case reinforces both the importance of early recognition of this syndrome — with genetic counselling and family screening — and the feasibility of minimally invasive surgical sympathetic denervation as an adjunct to medical therapy even in very young, low-weight children.

Declarations

Consent: Written informed consent for the surgical procedure was obtained from the patient's father and is documented in the clinical record.

Ethical Approval: Consent taken from the patient. No IRB approval for only case report

Funding: Not applicable

Conflicts of Interest: None declared.

Author Contribution: Saad Rehman compiled the case and write the case report

Research Registration: Not applicable

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