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Survival of a Newborn with 2:1 Atrioventricular Block, Long QT Syndrome, and Torsades de Pointes

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ABSTRACT

Long QT syndrome is a rare disorder that can manifest as syncope, Torsades de Pointes, or sudden cardiac death. We report a newborn with asymptomatic bradycardia, 2:1 atrioventricular block, long QT syndrome, and episodes of Torsades de Pointes. The patient was managed with mexiletine and propranolol and continued to have episodes of Torsades de Pointes, so she underwent epicardial pacemaker implantation. No further episodes of Torsades de Pointes were noted prior to discharge.

INTRODUCTION

Congenital, prolonged QT interval syndrome (LQTS) is a relatively uncommon cardiac disorder. There are a number of forms of LQTS, with each form affecting the patient in a different manner. To date, a few cases have been reported of infants with LQTS and 2:1 atrioventricular (AV) block (1–12). Treatment options for this form of LQTS include beta-blockers and mexiletine if long QT 3 is suspected; nevertheless, the prognosis remains poor. Few reports of the implantation of a pacemaker as a treatment for LQTS currently exist (3,6,7). To date, there is only a single known published case report regarding a neonate with LQTS, 2:1 AV block, and Torsades de Pointes (TdP) responding to pacemaker implantation (3). We report the case of an infant with LQTS, 2:1 AV block, and numerous episodes of TdP who responded to antiarrhythmic medications and implantation of a pacemaker.

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CASE REPORT

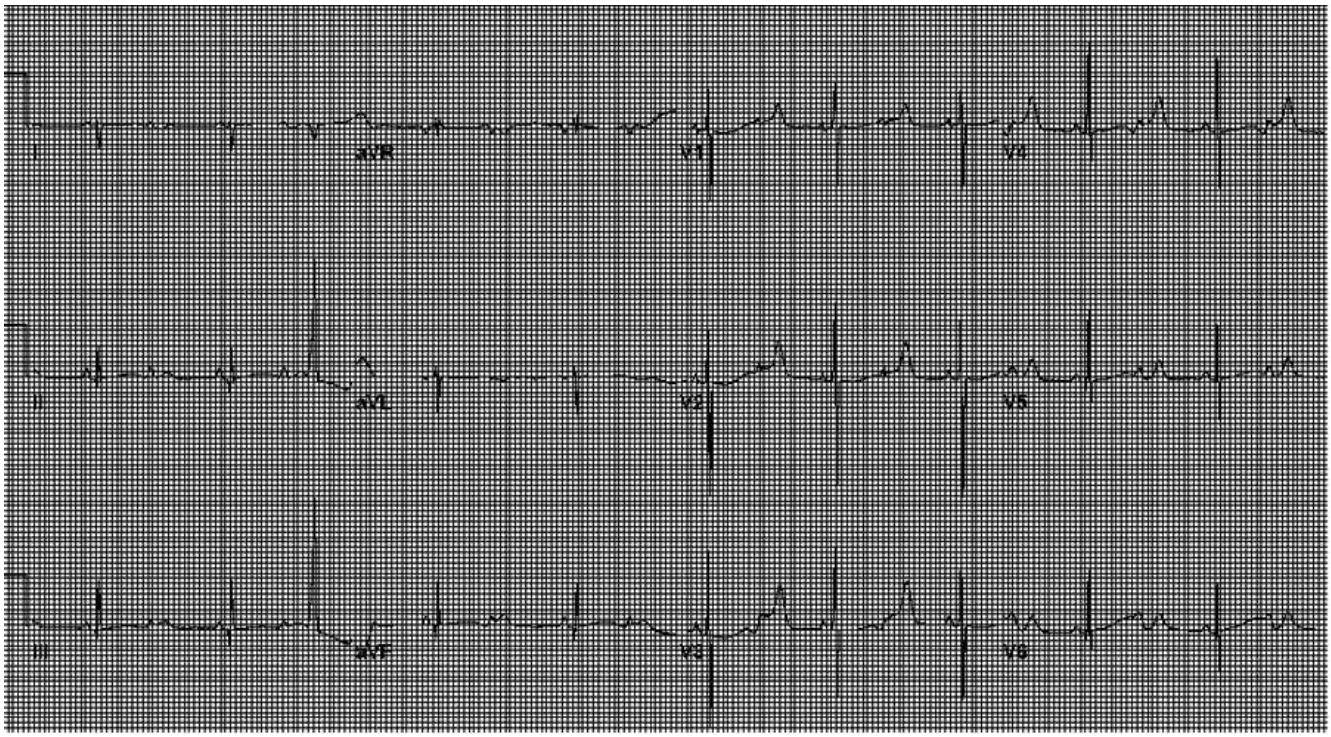
A 1-day-old female had been delivered to a seronegative, 25-year-old G1 P0-1 mother at 37-week gestation via emergent cesarean section secondary to a decreased fetal heart tone. At delivery, the infant's heart rate varied between 80 and 160 beats per minute (bpm). A pediatric cardiology consult found the patient to have a normal cardiac examination and echocardiogram, but an ECG showed sinus rhythm with a prolonged corrected QT interval and pseudo 2:1 AV block (Fig. 1). The patient was transferred to our institution for advanced electrophysiologic care.

Upon arrival, her physical examination, initial screening laboratory values, complete blood count, and basic metabolic profile were normal, despite a persistently slow heart rate (80 bpm). The patient and her mother were on no medications, and her past medical and surgical history was non-contributory. The family history was negative for sudden unexplained death, deafness, syncope, drowning, long QT, or Brugada's syndrome. ECGs on both parents were normal, with normal QT intervals. A blood sample was drawn for genetic evaluation and the patient was started on propranolol.

After initiation of propranolol at 4 mg/kg/d, she was placed on telemetry for observation. Review of her telemetry revealed numerous non-sustained episodes of TdP (Fig. 2). Mexiletine was subsequently added at 5 mg/kg/dose every 8 hours, but she continued to have episodes of TdP. At this point, she was referred for implantation of a single chamber, epicardial ventricular pacemaker. After pacemaker placement, she remained stable without further episodes of TdP (Figs. 3 and 4).

In follow-up, she has had no further recorded episodes of TdP since implantation of her pacemaker. Her genetic testing revealed a novel genetic mutation associated with the familial arrhythmia-causing syndrome of type 3 LQTS. This genetic substitution, located on exon 28, is the result of a substitution of threonine for methionine in the trans-membrane spanning region of the SCN5A gene.

Figure 1. Electrocardiogram with sinus rhythm with a prolonged corrected QT interval and pseudo 2:1 AV block.



DISCUSSION

Since the original description of 2:1 AV block and long QT syndrome by Scott and Dick in 1987 (13), patients with this relatively rare inherited arrhythmia continue to have a poor prognosis (3,13,11). Numerous reports of attempts at stabilizing rhythm disturbance with either beta-blockers or mexiletine have met with mixed results (1,4). Even patients who have undergone pacemaker implantation for this arrhythmia have not fared much better (3,6,13).

This patient with 2:1 AV block and type 3 long QT syndrome had episodes of TdP on antiarrhythmic medications. Her rhythm eventually stabilized once a pacemaker was implanted.

She had a relatively rare form of LQT syndrome know as type 3 LQTS, which is a direct result of a genetic mutation in the SCN5A gene. The SCN5A gene is a cardiac ion channel that regulates transportation of sodium into the cell. Defects in this gene result in a gain of function of this sodium channel. This gain of function causes a sustained inward sodium current, leading to an imbalance between the normal inward and outward currents. This imbalance in the sodium current results in a prolonged cardiac action potential, and, therefore, prolongs the QT interval.

In this small newborn patient, who had failed to respond to either anti-arrhythmic medication, the decision was made to implant a single chamber

Figure 2. Episode of non-sustained Torsades de Pointes.

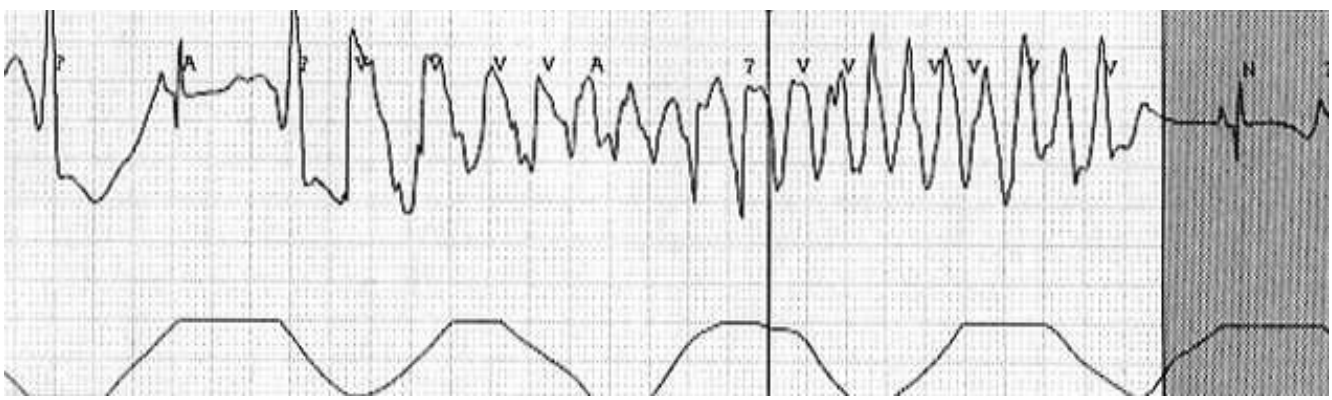


Figure 3. Electrocardiogram post-pacing.

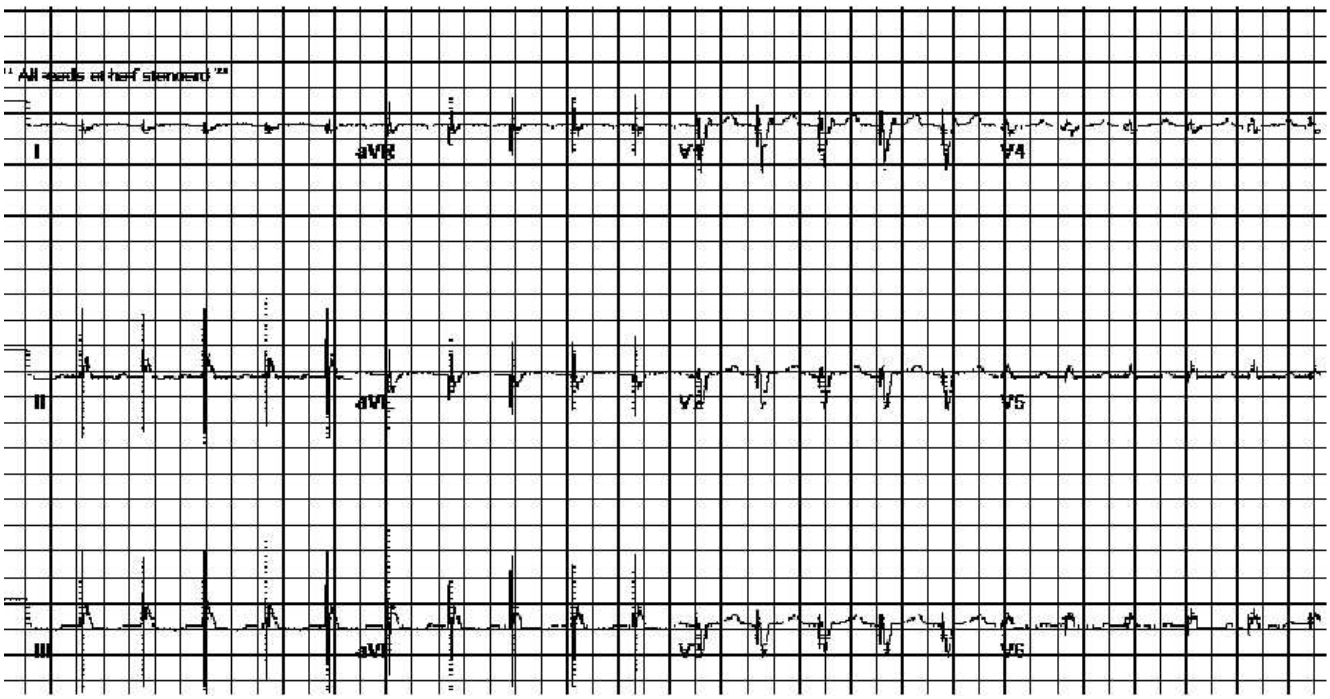
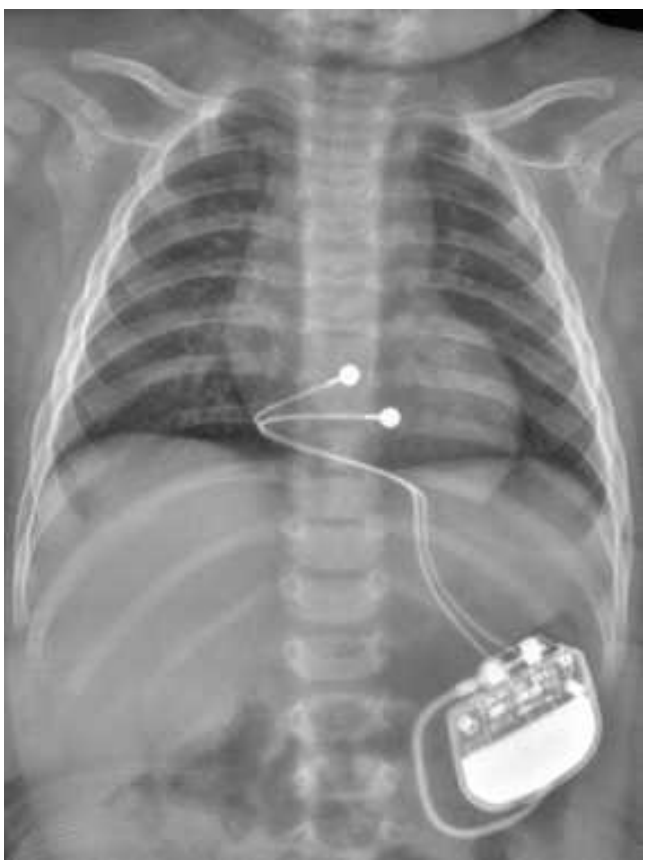


Figure 4. Chest X-ray after pacemaker implantation.



ventricular pacemaker. We opted not to implant an implantable cardioverter-defibrillator (ICD) based solely on the patient's size. Despite previously reported implants of ICDs in children, we felt this patient was too small even for a minimally invasive ICD implantation. We believed that permanent pacing might stabilize the rhythm, prevent episodes of bradycardia, and reduce or eliminate episodes of TdP (14,15).

CONCLUSION

We recommend that all newborn patients with 2:1 AV block and long QT syndrome receive both beta-blockers and mexiletine. Implantation of a pacing system should also be considered if TdP episodes occur. In addition, genetic testing should be performed on the proband and all first-degree relatives.

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