Transient Complete Atrioventricular Block in a Preterm Neonate with Congenital Myotonic Dystrophy: Case Report

Hee Na Kim,* Young Kuk Cho,* Joo Hyun Cho, Eun Mi Yang, Eun Song Song, and Young Youn Choi

Department of Pediatrics, Chonnam National University Medical School, Gwangju, Korea

*Hee Na Kim and Young Kuk Cho contributed equally to this work.

Received: 5 August 2013
Accepted: 19 November 2013

Address for Correspondence:
Eun Song Song, MD
Department of Pediatrics, Chonnam National University Medical School, 42 Jebong-ro, Dong-gu, Gwangju 501-757, Korea
Tel: +82.62-220-6649, Fax: +82.62-222-6103
E-mail: essong@jnu.ac.kr

INTRODUCTION

Congenital myotonic dystrophy (CMD) is an inherited neuromuscular disorder with major features including muscular hypotonia, respiratory failure, feeding problems, developmental delay, and less commonly systemic involvement including congenital cataract, thyroid dysfunction, and abnormal cardiac manifestation (1, 2). Cardiac involvement displays the following: conduction system disturbances, atrial and ventricular arrhythmias, hypertrophic cardiomyopathy, and sudden cardiac death (2-4). In patients with CMD, conduction disturbances including atrioventricular (AV) block generally occur during adulthood (5). There has been no recorded report of complete AV block in a neonate with CMD. We present a case of transient complete AV block completely managed with temporary transcutaneous pacing in a preterm neonate with CMD.

CASE DESCRIPTION

A 1,630 g female baby was born at 31+4 weeks of gestation by emergency Cesarean section to a spontaneously pregnant, nulli-, non-abortion, 33-yr-old mother due to persistent bradycardia of the fetal heart rate of 50 beats/min (bpm) on 11 December 2011. On pregnancy history, polyhydramnios (amniotic fluid index was 35) had been noted at 30+6 weeks of gestation at a local clinic. The result of amniocentesis for the fetal karyotype was normal. A decrease in fetal movements was detected from 31+2 weeks of gestation by 31+3-2 weeks of gestation. At birth, the baby’s Apgar score was 2 at 1 min, and she showed no respiration and movement, with a heart rate of 50-60 bpm. She was treated with ventilator assistance. At that time, the vital signs were as blood pressure (BP) of 57/34 (mean 41) mmHg, heart rate of 60 bpm, and O2 saturation (SpO2) of 50%. The initial venous blood gas analysis showed a pH of 7.099, pCO2 of 88.4 mmHg, and HCO3- of 27.4 mM. Despite administration of 100% oxygen, SpO2 was 60% and the heart rate was 60 bpm. As a result, cardiopulmonary resuscitation with intravenous epinephrine was performed. However, bradycardia of 60 bpm was persistent. A chest X-ray revealed respiratory distress syndrome with cardiacmegaly (cardiothoracic ratio 0.6), thin ribs, peripheral thinning, and floating clavicles (Fig. 1A). Surfactant was administered intratracheally. At 1 hr after birth, BP was decreased to 42/19 (mean 29) mmHg. Continuous intravenous infusion of dopamine, dobutamine, and epinephrine was started.

Electrocardiography (ECG) showed a complete AV block with an atrial rate of 150 bpm and a ventricular rate of 54 bpm (Fig. 2A). Echocardiography showed normal ventricular function with patent ductus arteriosus (PDA). For the treatment of the complete AV block, atropine and isoproterenol were administered, but there was no improvement. We applied temporary transcutaneous pacing (Responder 2000, Cardiac Science Corporation, USA) under sedation with midazolam at 4 hr after birth (Fig. 1B). The BP increased to 51/37 (mean 44) mmHg and the SpO2 gradually increased up to 100% on FiO2 of 0.21. Thirty two hours after transcutaneous pacing, the ECG completely recovered to a normal sinus rhythm of 135 bpm (Fig. 2B).

Conventional myotonic dystrophy (CMD) is an inherited neuromuscular disorder with cardiac rhythm abnormalities that may occur as a child grows. No report has described complete atrioventricular (AV) block detected in a neonate with CMD. We report a floppy infant of 31+4 weeks gestation with complete AV block at birth, who was diagnosed with CMD by Southern analysis. She recovered from complete AV block 32 hr after temporary transcutaneous pacing was applied. To the best of our knowledge, this is the first recorded case of a complete AV block accompanied by CMD during the neonatal period. When a newborn has a complete AV block, the physician should consider the possibility of the CMD and conduct a careful physical examination.

Keywords: Complete Atrioventricular Block; Congenital Myotonic Dystrophy; Pacing
The temporary transcutaneous pacing was removed at that time. However, there was a 3 × 1 cm-sized third degree contact burn, and escharotomy was done by a plastic surgeon at the removal site of the pacing at the apex (Fig. 3A). The burn wound healed by one month after birth.

Blood analysis revealed CK-MB of 15.6 U/L, troponin-I of 0.01 ng/mL, and N-terminal pro b-type natriuretic peptide of 5,575 pg/mL. In the serum autoimmune test of the mother, antinuclear antibody (ANA) and lupus anticoagulant were negative. Likewise, in the patient’s test, ANA, anti-Ro/SSA, and anti-Ra/SSB antibodies were negative.

On physical examination, the baby showed typical features of CMD, such as frontal bossing, weak facial expression with tented upper lip, high arched palate, generalized hypotonia with hyporeflexia, and bilateral talipes equinovarus (Fig. 3B). Genetic study for CMD revealed a congenital form of more than 1,000 copies of cytosine-thiamine-guanine (CTG) trinucleotide repeat at the 3´ untranslated end in the myotonic dystrophy protein kinase gene (DMPK) on chromosome 19 (Fig. 4A). According to the genetic study of the parents, the father was normal and the mother showed 150 copies of the CTG repeat, which was compatible with CMD carrier status (Fig. 4B).

For nutrition, gavage feeding was started at 9 days of birth. Additional parenteral nutrition was given for 2 months due to the persistence of the respiratory difficulties, weak sucking, and swallowing difficulties. On the 88th day after birth, she began to be fed orally. She was successfully weaned from the ventilator at 115 days of birth despite poor respiratory effort and copious secretions. At 137 days of birth, she was discharged from the hospital (Fig. 3C). Currently, she is 16 months old in corrected age and has a weak facial expression, along with poor respiratory effort, and has not had any respiratory difficulties or arrhythmia since discharge.

**DISCUSSION**

CMD is a multisystem disorder characterized by neonatal hypotonia, joint contractures, facial diplegia, fatal respiratory failure, feeding problems, cardiac conduction abnormalities, and developmental delay (1, 6). It presents at birth or during the first year after birth (1, 2). In the neonatal period, mortality is high with a rate of 17% to 41%. A common cause of death is respiratory insufficiency (6). Prenatally, reduced fetal movements or polyhydramnios that reflect intrauterine hypotonia may induce concerns of CMD (7). The genetic mechanism is understood to be caused by expansion of the CTG repeat in the 3´ untranslated region of the DMPK gene on chromosome 19 (8-10). There are possible correlations between the presence and proceeding of ECG abnormalities, the timing of cardiac complications, and the risk of major cardiac events and the number of CTG repeats (1). The pathology of cardiac involvement is known as interstitial fibrosis, hypertrophy of mycardiocytes, fatty infiltration, and focal myocarditis (11). Cardiac evaluation consisting of a
basal ECG, echocardiogram, and Holter monitoring should routinely be done in all patients with CMD (1, 2).

Conduction system abnormalities are the major cardiac abnormalities observed in CMD (1, 5). A recent study (12) reported the prevalence of conduction abnormality in patients with myotonic dystrophy including CMD between 1980 and 2010. The prevalence of conduction abnormalities were grade 1 AV block of 28.2%, grade 2 AV block of 2.1%, frequent premature ventricular contractions of 14.6%, right/left bundle branch block of 4.4%/5.7%, atrial fibrillation or flutter of 5%, and non-sustained ventricular tachycardia of 4.1%. The prevalence of a complete AV block was rare (0.3%), and did not occur in the neonatal pe-

Fig. 2. Electrocardiograms (ECG) of the patient. (A) ECG at birth showed a complete atioventricular block with an atrial rate of 150 bpm and a ventricular rate of 54 bpm. (B) Post-recovery ECG after applying the temporary transcutaneous pacing showed no conduction abnormality.
Kim HN, et al. • Transient Complete AV Block in Congenital Myotonic Dystrophy

Fig. 3. Gross appearance of the patient. (A) There was a third degree burn at the removal site of the pacing pad at the cardiac apex. (B and C) Gross appearance showed bilateral talipes equinovarus, joint contracture, frontal bossing, tented upper lip at 2.5 months and 4 months of age.

Fig. 4. Southern blot analysis of the patient and her mother. (A) The analysis of the patient showed more than 1,000 copies of cytosine-thiamine-guanine (CTG) repeat in the myotonic protein kinase gene (black arrow). (B) The analysis of her mother showed about 150 copies of the CTG repeat (white arrow).

A congenital complete AV block may occur due to maternal autoimmune diseases (14). In our case, there was no clinical or laboratory evidence of maternal or fetal autoimmune disease. The 2008 guidelines of the American Heart Association, the American College of Cardiology Foundation, and the Heart Rhythm Society recommend that a permanent pacemaker be implant-
a burn injury on the chest in our case, the temporary transcutaneous pacing was removed after 32 hr without any major problems.

In conclusion, when a newborn has a complete AV block, the physician should consider the possibility of the CMD and conduct a careful physical examination.

**ORCID**

Hee Na Kim  [http://orcid.org/0000-0001-7079-9012](http://orcid.org/0000-0001-7079-9012)
Young Kuk Cho  [http://orcid.org/0000-0001-6811-2883](http://orcid.org/0000-0001-6811-2883)
Eun Song Song  [http://orcid.org/0000-0003-1056-2165](http://orcid.org/0000-0003-1056-2165)

**REFERENCES**

1. Pelargonio G, Dello Russo A, Sanna T, De Martino G, Bellocchi F. Myotonic dystrophy and the heart. Heart 2002; 88: 665-70.
2. Campbell C. Congenital myotonic dystrophy. J Neurol Neurophysiol 2012; 57: 001. doi:10.4172/2155-9562.57-001.
3. Bassez G, Lazarus A, Desguerre I, Varin J, Laforêt P, Bécane HM, Meune C, Arne-Bes MC, Ounnoughene Z, Radvanyi H, et al. Severe cardiac arrhythmias in young patients with myotonic dystrophy type 1. Neurology 2004; 63: 1939-41.
4. Igarashi H, Momoi MY, Yamagata T, Shiraishi H, Eguchi I. Hypertrophic cardiomyopathy in congenital myotonic dystrophy. Pediatr Neurol 1998; 18: 366-9.
5. Forberg H, Olofsson BO, Eriksson A, Andersson S. Cardiac involvement in congenital myotonic dystrophy. Br Heart J 1990; 63: 119-21.
6. Campbell C, Sherlock R, Jacob P, Blayney M. Congenital myotonic dystrophy: assisted ventilation duration and outcome. Pediatrics 2004; 113: 811-6.
7. Rudnik-Schöneborn S, Nicholson GA, Morgan G, Röhrl D, Zerres K. Different patterns of obstetric complications in myotonic dystrophy in relation to the disease status of the fetus. Am J Med Genet 1998; 80: 314-21.
8. Buxton J, Shelbourne P, Davies J, Jones C, Van Tongeren T, Aslanidis C, de Jong P, Jansen G, Anrret M, Riley B, et al. Detection of an unstable fragment of DNA specific to individuals with myotonic dystrophy. Nature 1992; 355: 547-8.
9. Aslanidis C, Jansen G, Amemiy C, Shaiter G, Mahadevan M, Tsiflidis C, Chen C, Alleman I, Worskamp NG, Vooljs M, et al. Cloning of the essential myotonic dystrophy region and mapping of the putative defect. Nature 1992; 355: 548-51.
10. Brook JD, McCurrah ME, Harley HG, Buckler AJ, Church D, Abaratani H, Hunter K, Stanton VP, Thirion JP, Hudson T, et al. Molecular basis of myotonic dystrophy: expansion of a trinucleotide (CTG) repeat at the 3′ end of a transcript encoding a protein kinase family member. Cell 1992; 68: 799-808.
11. Phillips ME, Harper PS. Cardiac disease in myotonic dystrophy. Cardiovasc Res 1997; 33: 13-22.
12. Petri H, Vissing J, Witting N, Bundgaard H, Kober L. Cardiac manifestations of myotonic dystrophy type 1. Int J Cardiol 2012; 160: 82-8.
13. Michaelsson M, Engle MA. Congenital complete heart block: an international study of the natural history. Cardiovasc Clin 1972; 4: 85-101.
14. Udink ten Cate FE, Sreeram N. Pacing therapy in infants and children with congenital and acquired complete atrioventricular block: optimal pacing strategies, management, and follow-up. In: Das MK, editor. Modern pacemakers – present and future. Intech Publisher, 2011, p89-117.
15. Epstein AE, DiMarco JP, Ellenbogen KA, Estes NA 3rd, Freedman RA, Gettes LS, Gillinov AM, Gregoratos G, Hammill SC, Hayes DL, et al. ACC/AHA/HRS 2008 guidelines for device-based therapy of cardiac rhythm abnormalities: a report of the American College of Cardiology/American Heart Association Task Force on Practice Guidelines (writing committee to revise the ACC/AHA/NASPE 2002 guideline update for implantation of cardiac pacemakers and antiarrhythmia devices) developed in collaboration with the American Association for Thoracic Surgery and Society of Thoracic Surgeons. J Am Coll Cardiol 2008; 51: e1-62.
16. Friedman RA, Fenrich AL, Kertesz NJ. Congenital complete atrioventricular block. Pacing Clin Electrophysiol 2001; 24: 1681-8.
17. Maginot KR, Mathewson JW, Bichell DP, Perry JC. Applications of pacing strategies in neonates and infants. Prog Pediatr Cardiol 2000; 11: 65-75.