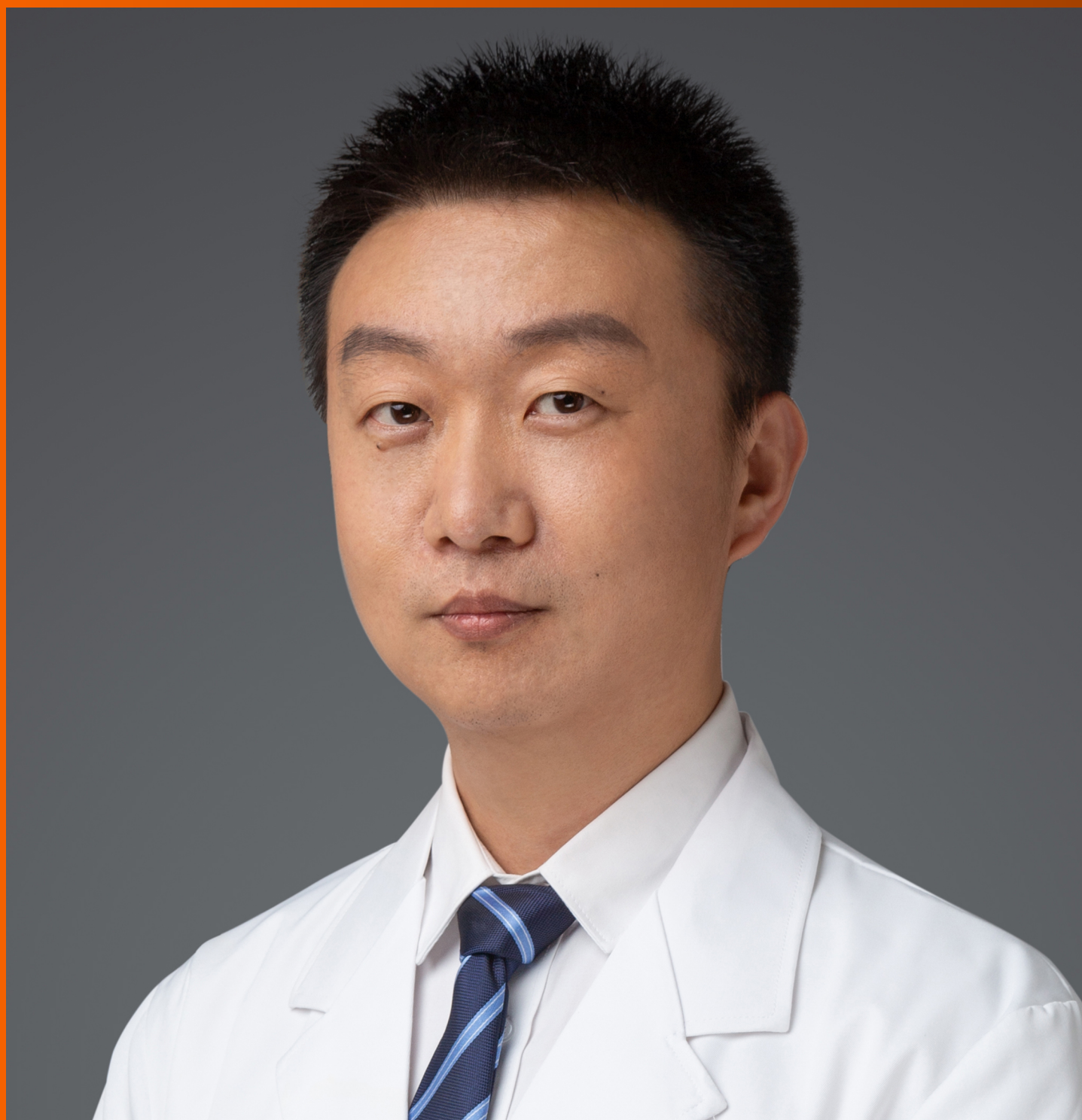


World Journal of *Clinical Cases*

World J Clin Cases 2021 April 16; 9(11): 2419-2695



MINIREVIEWS

- 2419 Current status of radical laparoscopy for treating hepatocellular carcinoma with portal hypertension
Shen ZF, Liang X

ORIGINAL ARTICLE**Retrospective Cohort Study**

- 2433 Impact of type 2 diabetes on adenoma detection in screening colonoscopies performed in disparate populations
Joseph DF, Li E, Stanley III SL, Zhu YC, Li XN, Yang J, Ottaviano LF, Bucobo JC, Buscaglia JM, Miller JD, Veluvolu R, Follen M, Grossman EB

- 2446 Early colonoscopy and urgent contrast enhanced computed tomography for colonic diverticular bleeding reduces risk of rebleeding
Ochi M, Kamoshida T, Hamano Y, Ohkawara A, Ohkawara H, Kakinoki N, Yamaguchi Y, Hirai S, Yanaka A

Retrospective Study

- 2458 Relationship between mismatch repair protein, *RAS*, *BRAF*, *PIK3CA* gene expression and clinicopathological characteristics in elderly colorectal cancer patients
Fan JZ, Wang GF, Cheng XB, Dong ZH, Chen X, Deng YJ, Song X

Clinical Trials Study

- 2469 Possible effect of blonanserin on gambling disorder: A clinical study protocol and a case report
Shiina A, Hasegawa T, Iyo M

Observational Study

- 2478 Parents' experience of caring for children with type 1 diabetes in mainland China: A qualitative study
Tong HJ, Qiu F, Fan L
- 2487 Differences in dietary habits of people with vs without irritable bowel syndrome and their association with symptom and psychological status: A pilot study
Meng Q, Qin G, Yao SK, Fan GH, Dong F, Tan C

SCIENTOMETRICS

- 2503 Prognostic nomograms for predicting overall survival and cause-specific survival of signet ring cell carcinoma in colorectal cancer patients
Kou FR, Zhang YZ, Xu WR

CASE REPORT

- 2519** Cerebellar artery infarction with sudden hearing loss and vertigo as initial symptoms: A case report
Wang XL, Sun M, Wang XP
- 2524** Three-dimensional-printed custom-made patellar endoprosthesis for recurrent giant cell tumor of the patella: A case report and review of the literature
Wang J, Zhou Y, Wang YT, Min L, Zhang YQ, Lu MX, Tang F, Luo Y, Zhang YH, Zhang XL, Tu CQ
- 2533** Gastrointestinal-type chemotherapy prolongs survival in an atypical primary ovarian mucinous carcinoma: A case report
Wang Q, Niu XY, Feng H, Wu J, Gao W, Zhang ZX, Zou YW, Zhang BY, Wang HJ
- 2542** Neoadjuvant chemoradiotherapy followed by laparoscopic distal gastrectomy in advanced gastric cancer: A case report and review of literature
Liu ZN, Wang YK, Li ZY
- 2555** Extraosseous spinal epidural plasmocytoma associated with multiple myeloma: Two case reports
Cui JF, Sun LL, Liu H, Gao CP
- 2562** Endoscopic diagnosis of early-stage primary esophageal small cell carcinoma: Report of two cases
Er LM, Ding Y, Sun XF, Ma WQ, Yuan L, Zheng XL, An NN, Wu ML
- 2569** Nemaline myopathy with dilated cardiomyopathy and severe heart failure: A case report
Wang Q, Hu F
- 2576** Immunoglobulin D- λ/λ biclonal multiple myeloma: A case report
He QL, Meng SS, Yang JN, Wang HC, Li YM, Li YX, Lin XH
- 2584** Point-of-care ultrasound for the early diagnosis of emphysematous pyelonephritis: A case report and literature review
Xing ZX, Yang H, Zhang W, Wang Y, Wang CS, Chen T, Chen HJ
- 2595** Minimally invasive treatment of forearm double fracture in adult using Acumed forearm intramedullary nail: A case report
Liu JC, Huang BZ, Ding J, Mu XJ, Li YL, Piao CD
- 2602** *Klebsiella pneumoniae* infection secondary to spontaneous renal rupture that presents only as fever: A case report
Zhang CG, Duan M, Zhang XY, Wang Y, Wu S, Feng LL, Song LL, Chen XY
- 2611** Eltrombopag-related renal vein thromboembolism in a patient with immune thrombocytopenia: A case report
Wu C, Zhou XM, Liu XD
- 2619** *Cryptococcus* infection with asymptomatic diffuse pulmonary disease in an immunocompetent patient: A case report
Li Y, Fang L, Chang FQ, Xu FZ, Zhang YB

- 2627** Triple administration of osimertinib followed by chemotherapy for advanced lung adenocarcinoma: A case report
Hu XY, Fei YC, Zhou WC, Zhu JM, Lv DL
- 2634** Anesthetic management of a child with double outlet right ventricle and severe polycythemia: A case report
Tan LC, Zhang WY, Zuo YD, Chen HY, Jiang CL
- 2641** Combined immune checkpoint inhibitors of CTLA4 and PD-1 for hepatic melanoma of unknown primary origin: A case report
Cheng AC, Lin YJ, Chiu SH, Shih YL
- 2649** Cholangiojejunostomy for multiple biliary ducts in living donor liver transplantation: A case report
Xiao F, Sun LY, Wei L, Zeng ZG, Qu W, Liu Y, Zhang HM, Zhu ZJ
- 2655** Surgical therapy for hemangioma of the azygos vein arch under thoracoscopy: A case report
Wang ZX, Yang LL, Xu ZN, Lv PY, Wang Y
- 2662** Calcium pyrophosphate deposition disease of the temporomandibular joint invading the middle cranial fossa: Two case reports
Tang T, Han FG
- 2671** Rare histological subtype of invasive micropapillary carcinoma in the ampulla of Vater: A case report
Noguchi H, Higashi M, Idichi T, Kurahara H, Mataka Y, Tasaki T, Kitazono I, Ohtsuka T, Tanimoto A
- 2679** Contrast-enhanced ultrasound using SonoVue mixed with oral gastrointestinal contrast agent to evaluate esophageal hiatal hernia: Report of three cases and a literature review
Wang JY, Luo Y, Wang WY, Zheng SC, He L, Xie CY, Peng L
- 2688** Melatonin for an obese child with MC4R gene variant showing epilepsy and disordered sleep: A case report
Ge WR, Wan L, Yang G

ABOUT COVER

Editorial Board Member of *World Journal of Clinical Cases*, Hong-Tao Xu, MD, PhD, Chief Physician, Professor, Department of Pathology, The First Affiliated Hospital and College of Basic Medical Sciences of China Medical University, Shenyang 110001, Liaoning Province, China. xuht@cmu.edu.cn

AIMS AND SCOPE

The primary aim of *World Journal of Clinical Cases* (WJCC, *World J Clin Cases*) is to provide scholars and readers from various fields of clinical medicine with a platform to publish high-quality clinical research articles and communicate their research findings online.

WJCC mainly publishes articles reporting research results and findings obtained in the field of clinical medicine and covering a wide range of topics, including case control studies, retrospective cohort studies, retrospective studies, clinical trials studies, observational studies, prospective studies, randomized controlled trials, randomized clinical trials, systematic reviews, meta-analysis, and case reports.

INDEXING/ABSTRACTING

The WJCC is now indexed in Science Citation Index Expanded (also known as SciSearch®), Journal Citation Reports/Science Edition, Scopus, PubMed, and PubMed Central. The 2020 Edition of Journal Citation Reports® cites the 2019 impact factor (IF) for WJCC as 1.013; IF without journal self cites: 0.991; Ranking: 120 among 165 journals in medicine, general and internal; and Quartile category: Q3. The WJCC's CiteScore for 2019 is 0.3 and Scopus CiteScore rank 2019: General Medicine is 394/529.

RESPONSIBLE EDITORS FOR THIS ISSUE

Production Editor: Jia-Hui Li; Production Department Director: Yu-Jie Ma; Editorial Office Director: Jin-Lai Wang.

NAME OF JOURNAL

World Journal of Clinical Cases

ISSN

ISSN 2307-8960 (online)

LAUNCH DATE

April 16, 2013

FREQUENCY

Thrice Monthly

EDITORS-IN-CHIEF

Dennis A Bloomfield, Sandro Vento, Bao-Gan Peng

EDITORIAL BOARD MEMBERS

<https://www.wjnet.com/2307-8960/editorialboard.htm>

PUBLICATION DATE

April 16, 2021

COPYRIGHT

© 2021 Baishideng Publishing Group Inc

INSTRUCTIONS TO AUTHORS

<https://www.wjnet.com/bpg/gerinfo/204>

GUIDELINES FOR ETHICS DOCUMENTS

<https://www.wjnet.com/bpg/GerInfo/287>

GUIDELINES FOR NON-NATIVE SPEAKERS OF ENGLISH

<https://www.wjnet.com/bpg/gerinfo/240>

PUBLICATION ETHICS

<https://www.wjnet.com/bpg/GerInfo/288>

PUBLICATION MISCONDUCT

<https://www.wjnet.com/bpg/gerinfo/208>

ARTICLE PROCESSING CHARGE

<https://www.wjnet.com/bpg/gerinfo/242>

STEPS FOR SUBMITTING MANUSCRIPTS

<https://www.wjnet.com/bpg/GerInfo/239>

ONLINE SUBMISSION

<https://www.f6publishing.com>



Nemaline myopathy with dilated cardiomyopathy and severe heart failure: A case report

Qian Wang, Fan Hu

ORCID number: Qian Wang 0000-0002-0382-9872; Fan Hu 0000-0002-2583-4058.

Author contributions: Wang Q and Hu F provided the concept for the study; Wang Q drafted the manuscript; Hu F performed the review; all authors have read and approved the content of the manuscript.

Informed consent statement:

Informed written consent was obtained from the patient for publication of this report and any accompanying images.

Conflict-of-interest statement: We declare that we have no conflicts of interest related to this work.

CARE Checklist (2016) statement:

The authors have read the CARE Checklist (2016), and the manuscript was prepared and revised according to the CARE Checklist (2016).

Open-Access: This article is an open-access article that was selected by an in-house editor and fully peer-reviewed by external reviewers. It is distributed in accordance with the Creative Commons Attribution NonCommercial (CC BY-NC 4.0) license, which permits others to distribute, remix, adapt, build upon this work non-commercially,

Qian Wang, Department of Pediatric Neurology, West China Second University Hospital, Sichuan University, Chengdu 610041, Sichuan Province, China

Fan Hu, Department of Pediatric Cardiology, West China Second University Hospital, Sichuan University, Chengdu 610041, Sichuan Province, China

Corresponding author: Fan Hu, MD, Associate Chief Physician, Department of Pediatric Cardiology, West China Second University Hospital, Sichuan University, No. 20, 3rd Section, South Renmin Road, Chengdu 610041, Sichuan, China. heracleshu@sina.com

Abstract

BACKGROUND

Nemaline myopathy (NM) is a rare type of congenital myopathy, with an incidence of 1:50000. Patients with NM often exhibit hypomyotonia and varying degrees of muscle weakness. Skeletal muscles are always affected by this disease, while myocardial involvement is uncommon. However, with improvements in genetic testing technology, it has been found that NM with a mutation in the myopalladin (*MYPN*) gene not only causes slow, progressive muscle weakness but also results in dilated or hypertrophic cardiomyopathy.

CASE SUMMARY

A 3-year-old pre-school boy was admitted to our hospital with cough, edema, tachypnea, and an increased heart rate. The patient was clinically diagnosed with severe dilated cardiomyopathy and heart failure, and subsequent gene examination confirmed the diagnosis of NM with a mutation in *MYPN*. Captopril, diuretics, low-dose digoxin, and dobutamine were administered. After 22 d of hospitalization, the patient was discharged due to the improvement of clinical symptoms. During the follow-up period, the patient died of refractory heart failure.

CONCLUSION

Decreased muscular tone and dilated cardiomyopathy are common features of *MYPN*-mutated NM. Heart transplantation may be a solution to this type of cardiomyopathy.

Key Words: Nemaline myopathy; Myopalladin; Dilated cardiomyopathy; Heart failure; Whole-exome sequencing; Case report

and license their derivative works on different terms, provided the original work is properly cited and the use is non-commercial. See: <http://creativecommons.org/licenses/by-nc/4.0/>

Manuscript source: Unsolicited manuscript

Specialty type: Pediatrics

Country/Territory of origin: China

Peer-review report's scientific quality classification

Grade A (Excellent): 0

Grade B (Very good): B

Grade C (Good): 0

Grade D (Fair): 0

Grade E (Poor): 0

Received: December 25, 2020

Peer-review started: December 25, 2020

First decision: January 10, 2021

Revised: January 23, 2021

Accepted: February 8, 2021

Article in press: February 8, 2021

Published online: April 16, 2021

P-Reviewer: Dzięwiecka E

S-Editor: Zhang H

L-Editor: Wang TQ

P-Editor: Ma YJ



©The Author(s) 2021. Published by Baishideng Publishing Group Inc. All rights reserved.

Core Tip: Nemaline myopathy (NM) is a rare kind of congenital myopathy, with an incidence of 1:50000. The pathological characteristic is accumulated “rod” shaped structures in the muscle biopsies observed by light or electron microscopy. NM patients often have hypomyotonia and different degrees of muscle weakness. Skeletal muscles are always involved in this disease, while myocardial involvement is uncommon. However, it has been recognized that NM with mutation in myopalladin (*MYPN*) gene also results in dilated cardiomyopathy or hypertrophic cardiomyopathy. Here, we report the case of a 3-year-old boy with NM who was admitted with dilated cardiomyopathy and heart failure followed by genetic confirmation of NM with an *MYPN* mutation.

Citation: Wang Q, Hu F. Nemaline myopathy with dilated cardiomyopathy and severe heart failure: A case report. *World J Clin Cases* 2021; 9(11): 2569-2575

URL: <https://www.wjgnet.com/2307-8960/full/v9/i11/2569.htm>

DOI: <https://dx.doi.org/10.12998/wjcc.v9.i11.2569>

INTRODUCTION

Nemaline myopathy (NM) is a rare type of congenital myopathy, with an incidence of 1:50000^[1]. A distinct pathological characteristic of NM is the accumulation of “rod” shaped structures observed by light or electron microscopy in muscle biopsies^[2]. Patients with NM often have hypomyotonia and varying degrees of muscle weakness. Skeletal muscles are always affected by this disease, while myocardial involvement is uncommon. However, with improvements in genetic testing technology, it has been recognized that NM with a mutation in the myopalladin (*MYPN*) gene not only causes slow, progressive muscle weakness but also results in dilated or hypertrophic cardiomyopathy^[3].

Here, we report the case of a 3-year-old boy with NM who was admitted with dilated cardiomyopathy and heart failure followed by the genetic confirmation of NM with an *MYPN* mutation. The ethics committee of the West China Second University Hospital of Sichuan University approved this study. Informed consent was obtained from the patient for publication of this case report.

CASE PRESENTATION

Chief complaints

A 3-year-old boy was referred to our hospital with cough, edema, tachypnea, and tachycardia.

History of present illness

One week before admission, the patient began to exhibit a paroxysm of coughing with phlegm, accompanied with fatigue and plummeting level of physical activity. Soon afterwards, he exhibited edema all over the body, particularly on the face and both lower limbs. Half a day before admission, the patient developed tachypnea and tachycardia.

History of past illness

It is worth noting that the patient had a history of delayed physical growth development. He only learned to sit when he was 10 mo old, and he still could not crawl or stand at 1 year of age. He was sent to a hospital and diagnosed with growth retardation. Doctors guided the boy in a rehabilitation training program for 1 year, after which he appeared to walk and run with no significant difference compared with his peers. However, the parents found that his muscle tension was low, and that he fell over easily. In addition, the patient had strephenopodia after birth, which improved after the use of orthotics.

Personal and family history

The family history was unremarkable.

Physical examination

When the patient was admitted to our hospital, he had symmetrical edema in the face and lower extremities. The pulmonary respiratory sounds were rough with a few coarse rales. There was no protuberance in the precordial region. The apical impulse of the heart was diffused. Heart amplification was identified, and the apical beat was at the sixth intercostal space, 4.5 cm outside the middle line of the left clavicle. Neither thrill nor pericardium friction was found. The heart rhythm was regular with a gallop rhythm and low cardiac sound. The abdomen was supple with the liver 4 cm below the costal margin and 6 cm below the xiphoid, with the spleen impalpable. Moreover, the boy had an elongated face, inhibited facial expressions, a high palate arch (*Figure 1*), clawfoot, normal muscular strength, decreased muscular tone, and decreased bilateral knee reflexes. The Gower's sign was positive, and the meningeal irritation sign was negative.

Laboratory examinations

The serum cardiac troponin I level was 0.211 µg/L (normal < 0.034 µg/L). The level of brain natriuretic peptide reached up to 27500 pg/mL (normal < 215 pg/mL). The alanine aminotransferase level was 458 U/L (normal < 72 U/L), and aspartate aminotransferase level was 671 U/L (normal < 59 U/L). However, there was no significant increase in creatine kinase (99 U/L; normal < 170 U/L), creatine kinase myocardial band (2.29 µg/L; normal < 3.38 µg/L), and myoglobin (133.1 µg/L; normal < 121 µg/L).

Imaging examinations

After one week in the hospital, Holter showed about 16.6% of ventricular premature beats (VPBs) of the total number of beats (*Figure 2*). Echocardiogram showed that the patient had enlarged, weakened left and right ventricles with decreased systolic function. The heart chamber sizes were as follows: Left atrium, 27 mm (normal < 18 mm); left ventricle, 58 mm (normal < 31 mm); right atrium, 40 mm (normal < 32 mm); right ventricle, 22 mm (normal < 11 mm). The ejection fraction was 18%, and the fraction of shortening was 8%. The systolic excursion of the tricuspid annular plane was 13 mm (*Figure 3*). Cardiac magnetic resonance imaging showed enlarged ventricles, reduced systolic function, and focal delayed enhancement (*Figure 4*).

FINAL DIAGNOSIS

Based on the above findings, NM was suspected.

TREATMENT

Initially, we thought that the condition in question might be myocarditis or idiopathic cardiomyopathy. The patient received spironolactone (2 mg/kg), hydrochlorothiazide (2 mg/kg), captopril (1 mg/kg), and digoxin (6 µg/kg). In addition, intravenous methylprednisolone was administered for 3 d followed by oral prednisone for 4 d. The degree of heart failure appeared to be very severe, so dobutamine (6 µg/kg) was also administered on the day of admission. The edema and cough improved, with the disappearance of rales after 1 wk of treatment, and we stopped using dobutamine (*Table 1*).

We continued using spironolactone, hydrochlorothiazide, captopril, digoxin, and dobutamine (we stopped using dobutamine when the patient was in stable condition). After 22 d of treatment, the edema completely regressed and the gallop rhythm disappeared. The patient still exhibited sinus tachycardia with a reduced VPB of 8.9%. Echocardiography showed that the ejection fraction was 23% and the fraction of shortening was 11% after treatment.

Table 1 Summary of the clinical course

	Symptoms			Signs				Drug administration
	Edema	Cough	Tachypnea	Tachycardia	Rale	Gallop rhythm	Hepatomegaly	
Day 1	+	+	++	++	+	+	4 cm below costal margin	Spironolactone, hydrochlorothiazide, captopril, digoxin, dobutamine, methylprednisolone
Day 4	+	+	++	++	+	+	4 cm below costal margin	Spironolactone, hydrochlorothiazide, captopril, digoxin, dobutamine, prednisolone
Day 8	-	-	++	++	-	+	4 cm below costal margin	Spironolactone, hydrochlorothiazide, captopril, digoxin
Day 18	-	-	+	+	-	-	3 cm below costal margin	Spironolactone, hydrochlorothiazide, captopril, digoxin
Day 22	-	-	+	+	-	-	3 cm below costal margin	Spironolactone, hydrochlorothiazide, captopril, digoxin

**Figure 1** High palate arch of the patient: A typical sign of nemaline myopathy.

OUTCOME AND FOLLOW-UP

The patient was discharged due to improvement in heart failure symptoms. He continued to be administered with spironolactone, hydrochlorothiazide, captopril, and digoxin at home. During the follow-up period, the patient suffered from repeated and worsened heart failure and was hospitalized. Ultimately, he died of uncontrollable heart failure 4 mo after the first admission.

DISCUSSION

NM is a sporadic, rare congenital myopathy^[4]. Genetic mutations that cause skeletal muscle involvement lead to this disease. Clinical manifestations vary in different patients. NM was divided into six types by the European Commission for Neuromuscular Diseases^[5]: (1) Severe congenital NM. Patients exhibit clinical symptoms at birth, with feeding difficulties, hypotonia, myasthenia, respiratory failure, and no independent activities. Most patients die at an early stage, usually no more than one year of age; (2) Typical form of NM. This type has the highest incidence. Children get sick at an early age, with obvious muscle weakness in the face,

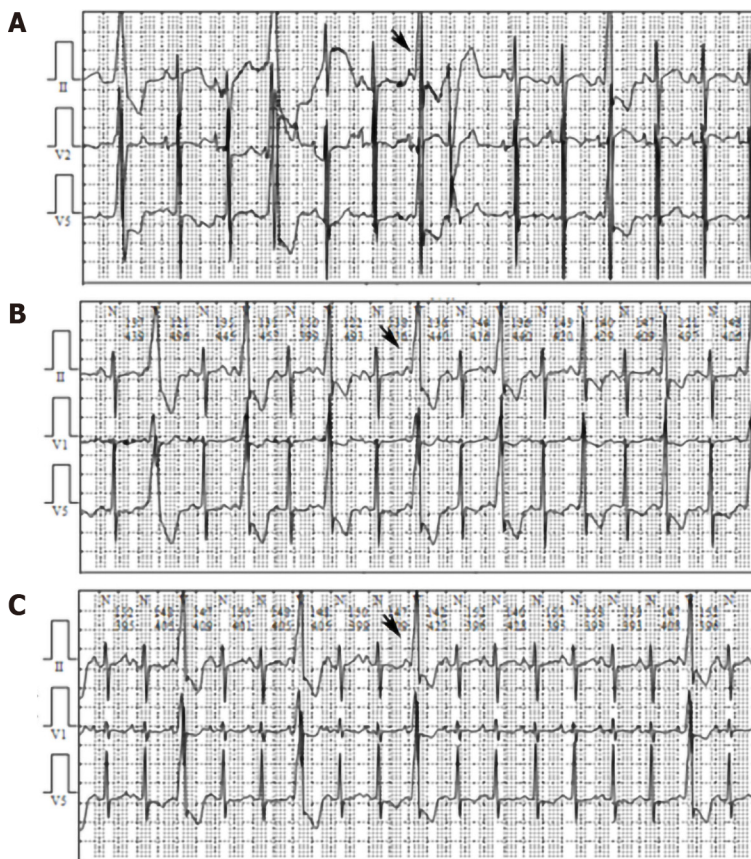


Figure 2 A total of 28352 ventricular premature beats in Holter monitoring. A: Four instances of paired ventricular premature beats; B: 2069 instances of ventricular bigeminy; C: 245 instances of ventricular trigeminy.

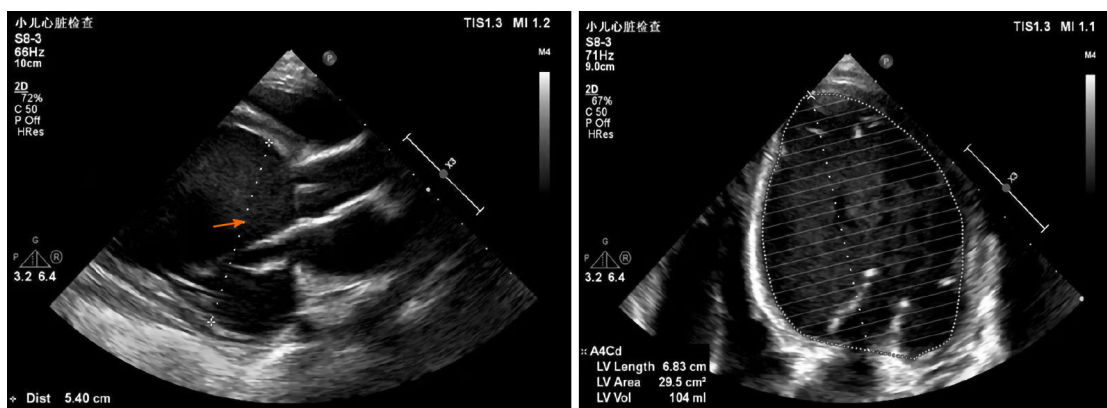


Figure 3 Echocardiogram. The heart chamber sizes showed an enlarged heart, especially the left heart.

medulla oblongata, and respiratory system. Muscle strength possibly increases with age. The quality of life in most patients is not affected. Patients have normal intelligence and myocardial contractility; (3) Intermediate congenital NM. This form has an onset in early infancy, spontaneous respiratory movement may occur at birth, but in early childhood. Patients may develop an inability to breathe autonomously, and walk and stand independently. Muscle contractures may occur during early childhood. The clinical symptoms of this type of NM are congenital severe syndromes and congenital mild syndromes; (4) Mild, childhood-, or juvenile-onset NM. The clinical manifestations are similar to mild syndromes, except that the onset age is late childhood or adolescence; (5) Adult form of NM. The age of onset is 30 to 60 years, and this form is incredibly diverse, with vast differences in clinical manifestations and disease progression. Cases are sporadic and have no family history. The onset is acute or subacute, and the respiratory muscles are easily involved. The disease has a quick

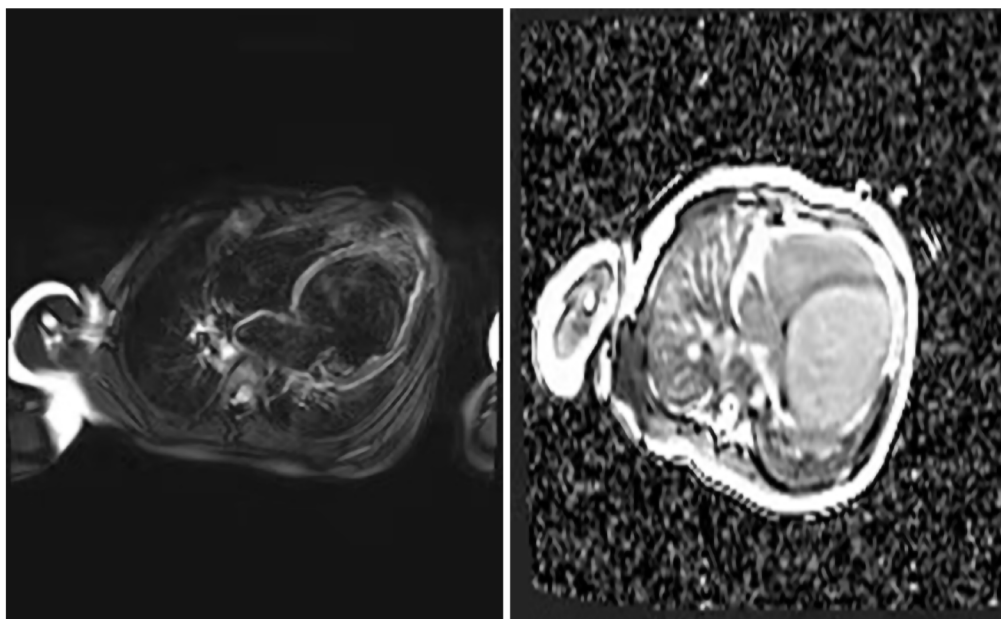


Figure 4 Magnetic resonance images. Cardiac magnetic resonance imaging showed enlarged ventricles, reduced systolic function, and focal delayed enhancement.

course, and most patients have a poor prognosis; and (6) Other forms of NM. These are rare types, presenting as cardiomyopathy, ophthalmoplegia, abnormal muscle weakness distribution, and internuclear rods.

The presence of rod-like bodies is a characteristic change observed in muscle biopsies, and there are many eosinophilic rod-like structures under the myofibril or in the myoplasm^[6-8]. However, due to various limitations, muscle biopsies are not easy to perform. Besides, with the development of genetic testing, muscle biopsies are no longer the only gold standard. Our patient did not undergo a muscle biopsy due to severe heart failure and refusal from the parents.

So far, at least 13 gene mutations have been recognized to be associated with NM, including *TPM3*, *NEB*, *ACTA1*, *TPM2*, *TNNT1*, *KBTD13*, *CFL2*, *KLHL40*, *KLHL41*, *LMOD3*, *MYO18B*, *TNNT3*, and *MYPN*^[9]. *MYPN* is a sarcoma component. It is generally found in the sarcomere, cardiac muscle, and skeletal muscle nucleus. The central regions and C-terminus of *MYPN* can bind to both α -actinin and nebulin in the skeletal muscle or nebulin in the cardiac muscle at the Z-line^[9]. On the other hand, its N-terminal region binds to cardiac ankyrin repeat protein^[10]. In an animal model, *MYPN* has been shown to potentially promote muscle growth^[11]. Patients with *MYPN* mutations show manifestations of myopathy and dilated or hypertrophic cardiomyopathy^[12,13].

The patient in question had normal intelligence, but his motor developmental milestones were delayed after birth. In addition, the facial and skeletal characteristics, including an elongated face, high palate arch, and clawfoot, are typical signs of NM^[14,15]. Decreased muscular tone, a positive Gower's sign, and dilated cardiomyopathy are common features of *MYPN*-mutated NM. The typical clinical manifestations and genetic tests supported the diagnosis. Unfortunately, the heart failure was too severe to be controlled by drugs. β -blockers are another treatment option for cardiomyopathy but the patient had such poor heart function that we did not use it after careful evaluation of the risk. Heart transplantation may be a solution to this type of cardiomyopathy.

CONCLUSION

In some cases, life-threatening cardiomyopathy with severe heart failure is a manifestation of inherited myopathy. When diagnosing cardiomyopathy, the development of striated muscles should be detected.

ACKNOWLEDGEMENTS

We thank all of the participants in this case report and people involved in diagnosis. We thank the Sichuan Province Science and Technology Support Program of China. We also thank the parents of the patient, who were always cooperative with our doctors.

REFERENCES

- 1 **de Winter JM**, Ottenheijm CAC. Sarcomere Dysfunction in Nemaline Myopathy. *J Neuromuscul Dis* 2017; **4**: 99-113 [PMID: [28436394](#) DOI: [10.3233/JND-160200](#)]
- 2 **Cassandrini D**, Trovato R, Rubegni A, Lenzi S, Fiorillo C, Baldacci J, Minetti C, Astrea G, Bruno C, Santorelli FM; Italian Network on Congenital Myopathies. Congenital myopathies: clinical phenotypes and new diagnostic tools. *Ital J Pediatr* 2017; **43**: 101 [PMID: [29141652](#) DOI: [10.1186/s13052-017-0419-z](#)]
- 3 **Sewry CA**, Laitila JM, Wallgren-Pettersson C. Nemaline myopathies: a current view. *J Muscle Res Cell Motil* 2019; **40**: 111-126 [PMID: [31228046](#) DOI: [10.1007/s10974-019-09519-9](#)]
- 4 **Yeşilbaş O**, Şevketoglu E, Kılıtr HS, Ersoy M, Petmezci MT, Akkuş CH, Şahin Ö, Ceylaner S. A rare structural myopathy: Nemaline myopathy. *Turk Pediatri Ars* 2019; **54**: 49-52 [PMID: [31217710](#) DOI: [10.5152/TurkPediatriArs.2018.4402](#)]
- 5 **Wallgren-Pettersson C**, Laing NG. Report of the 70th ENMC International Workshop: nemaline myopathy, 11-13 June 1999, Naarden, The Netherlands. *Neuromuscul Disord* 2000; **10**: 299-306 [PMID: [10838258](#) DOI: [10.1016/s0960-8966\(99\)00129-7](#)]
- 6 **Schnitzler LJ**, Schreckenbach T, Nadaj-Pakleza A, Stenzel W, Rushing EJ, Van Damme P, Ferbert A, Petri S, Hartmann C, Bornemann A, Meisel A, Petersen JA, Tousseyn T, Thal DR, Reimann J, De Jonghe P, Martin JJ, Van den Bergh PY, Schulz JB, Weis J, Claeys KG. Sporadic late-onset nemaline myopathy: clinico-pathological characteristics and review of 76 cases. *Orphanet J Rare Dis* 2017; **12**: 86 [PMID: [28490364](#) DOI: [10.1186/s13023-017-0640-2](#)]
- 7 **D'Amico A**, Fattori F, Fiorillo C, Paglietti MG, Testa MBC, Verardo M, Catteruccia M, Bruno C, Bertini E. 'Amish Nemaline Myopathy' in 2 Italian siblings harbouring a novel homozygous mutation in Troponin-I gene. *Neuromuscul Disord* 2019; **29**: 766-770 [PMID: [31604653](#) DOI: [10.1016/j.nmd.2019.09.005](#)]
- 8 **Sztal TE**, Zhao M, Williams C, Oorschot V, Parslow AC, Giousoh A, Yuen M, Hall TE, Costin A, Ramm G, Bird PI, Busch-Nentwich EM, Stemple DL, Currie PD, Cooper ST, Laing NG, Nowak KJ, Bryson-Richardson RJ. Zebrafish models for nemaline myopathy reveal a spectrum of nemaline bodies contributing to reduced muscle function. *Acta Neuropathol* 2015; **130**: 389-406 [PMID: [25931053](#) DOI: [10.1007/s00401-015-1430-3](#)]
- 9 **Miyatake S**, Mitsuhashi S, Hayashi YK, Purevjav E, Nishikawa A, Koshimizu E, Suzuki M, Yatabe K, Tanaka Y, Ogata K, Kuru S, Shiina M, Tsurusaki Y, Nakashima M, Mizuguchi T, Miyake N, Saitsu H, Ogata K, Kawai M, Towbin J, Nonaka I, Nishino I, Matsumoto N. Biallelic Mutations in MYPN, Encoding Myopalladin, Are Associated with Childhood-Onset, Slowly Progressive Nemaline Myopathy. *Am J Hum Genet* 2017; **100**: 169-178 [PMID: [28017374](#) DOI: [10.1016/j.ajhg.2016.11.017](#)]
- 10 **Bang ML**, Mudry RE, McElhinny AS, Trombitás K, Geach AJ, Yamasaki R, Sorimachi H, Granzier H, Gregorio CC, Labeit S. Myopalladin, a novel 145-kilodalton sarcomeric protein with multiple roles in Z-disc and I-band protein assemblies. *J Cell Biol* 2001; **153**: 413-427 [PMID: [11309420](#) DOI: [10.1083/jcb.153.2.413](#)]
- 11 **Merlini L**, Sabatelli P, Antonietti M, Carinci V, Niro F, Monetti G, Torella A, Giugliano T, Faldini C, Nigro V. Congenital myopathy with hanging big toe due to homozygous myopalladin (MYPN) mutation. *Skelet Muscle* 2019; **9**: 14 [PMID: [31133047](#) DOI: [10.1186/s13395-019-0199-9](#)]
- 12 **Zhao Y**, Feng Y, Zhang YM, Ding XX, Song YZ, Zhang AM, Liu L, Zhang H, Ding JH, Xia XS. Targeted next-generation sequencing of candidate genes reveals novel mutations in patients with dilated cardiomyopathy. *Int J Mol Med* 2015; **36**: 1479-1486 [PMID: [26458567](#) DOI: [10.3892/ijmm.2015.2361](#)]
- 13 **Chen Y**, Barajas-Martinez H, Zhu D, Wang X, Chen C, Zhuang R, Shi J, Wu X, Tao Y, Jin W, Wang X, Hu D. Erratum to: Novel trigenic CACNA1C/DES/MYPN mutations in a family of hypertrophic cardiomyopathy with early repolarization and short QT syndrome. *J Transl Med* 2017; **15**: 101 [PMID: [28490369](#) DOI: [10.1186/s12967-017-1203-y](#)]
- 14 **Xue Y**, Magoulas PL, Wirthlin JO, Buchanan EP. Craniofacial Manifestations in Severe Nemaline Myopathy. *J Craniofac Surg* 2017; **28**: e258-e260 [PMID: [28468212](#) DOI: [10.1097/SCS.00000000000003483](#)]
- 15 **Yeung KS**, Yu FNY, Fung CW, Wong S, Lee HHC, Fung STH, Fung GPG, Leung KY, Chung WH, Lee YT, Ng VKS, Yu MHC, Fung JLF, Tsang MHY, Chan KYK, Chan SHS, Kan ASY, Chung BHY. The KLHL40 c.1516A>C is a Chinese-specific founder mutation causing nemaline myopathy 8: Report of six patients with pre- and postnatal phenotypes. *Mol Genet Genomic Med* 2020; **8**: e1229 [PMID: [32352246](#) DOI: [10.1002/mgg3.1229](#)]



Published by **Baishideng Publishing Group Inc**
7041 Koll Center Parkway, Suite 160, Pleasanton, CA 94566, USA

Telephone: +1-925-3991568

E-mail: bpgoffice@wjgnet.com

Help Desk: <https://www.f6publishing.com/helpdesk>

<https://www.wjgnet.com>

