

# World Journal of *Clinical Cases*

*World J Clin Cases* 2020 January 6; 8(1): 1-244





### REVIEW

- 1 Role of oxysterol-binding protein-related proteins in malignant human tumours  
*Liu H, Huang S*

### ORIGINAL ARTICLE

#### Case Control Study

- 11 Oncogenic role of Tc17 cells in cervical cancer development  
*Zhang ZS, Gu Y, Liu BG, Tang H, Hua Y, Wang J*

#### Retrospective Study

- 20 Acute distal common bile duct angle is risk factor for post-endoscopic retrograde cholangiopancreatography pancreatitis in beginner endoscopist  
*Han SY, Kim DU, Lee MW, Park YJ, Baek DH, Kim GH, Song GA*
- 29 Three-dimensional computed tomography mapping of posterior malleolar fractures  
*Su QH, Liu J, Zhang Y, Tan J, Yan MJ, Zhu K, Zhang J, Li C*
- 38 Application of a modified surgical position in anterior approach for total cervical artificial disc replacement  
*Hou WX, Zhang HX, Wang X, Yang HL, Luan XR*
- 46 Potential role of the compound Eucommia bone tonic granules in patients with osteoarthritis and osteonecrosis: A retrospective study  
*Hu CX, Hu KY, Wang JF*
- 54 Prognostic factors for overall survival in prostate cancer patients with different site-specific visceral metastases: A study of 1358 patients  
*Cui PF, Cong XF, Gao F, Yin JX, Niu ZR, Zhao SC, Liu ZL*
- 68 Application of multiple Roux-en-Y hepaticojejunostomy reconstruction by formation of bile hilar duct lake in the operation of hilar cholangiocarcinoma  
*Yang XJ, Dong XH, Chen SY, Wu B, He Y, Dong BL, Ma BQ, Gao P*

#### Observational Study

- 76 Relationship between  $\beta$ -amyloid protein 1-42, thyroid hormone levels and the risk of cognitive impairment after ischemic stroke  
*Mao L, Chen XH, Zhuang JH, Li P, Xu YX, Zhao YC, Ma YJ, He B, Yin Y*

**Prospective Study**

- 88 Can the wet suction technique change the efficacy of endoscopic ultrasound-guided fine-needle aspiration for diagnosing autoimmune pancreatitis type 1? A prospective single-arm study  
*Sugimoto M, Takagi T, Suzuki R, Konno N, Asama H, Sato Y, Irie H, Watanabe K, Nakamura J, Kikuchi H, Takasumi M, Hashimoto M, Kato T, Hikichi T, Notohara K, Ohira H*

**CASE REPORT**

- 97 Pembrolizumab - emerging treatment of pulmonary sarcomatoid carcinoma: A case report  
*Cimpeanu E, Ahmed J, Zafar W, DeMarinis A, Bardarov SS, Salman S, Bloomfield D*
- 103 Sclerosing angiomatoid nodular transformation of the spleen, a rare cause for splenectomy: Two case reports  
*Chikhladze S, Lederer AK, Fichtner-Feigl S, Wittel UA, Werner M, Aumann K*
- 110 Postpartum pubic symphysis diastasis-conservative and surgical treatment methods, incidence of complications: Two case reports and a review of the literature  
*Norvilaite K, Kezeviciute M, Ramasauskaite D, Arlauskienė A, Bartkeviciene D, Uvarovas V*
- 120 Use of omental patch and endoscopic closure technique as an alternative to surgery after endoscopic full thickness resection of gastric intestinal stromal tumors: A series of cases  
*Sachdev AH, Iqbal S, Ribeiro IB, de Moura DTH*
- 126 Primary maxillary chondrosarcoma: A case report  
*Cuevas-González JC, Reyes-Escalera JO, González JL, Sánchez-Romero C, Espinosa-Cristóbal LF, Reyes-López SY, Tovar Carrillo KL, Donohue Cornejo A*
- 133 Hyalinizing clear cell carcinoma-a rare entity in the oral cavity: A case report  
*Donohue-Cornejo A, Paes de Almeida O, Sánchez-Romero C, Espinosa-Cristóbal LF, Reyes-López SY, Cuevas-González JC*
- 140 Jejunal cavernous lymphangioma manifested as gastrointestinal bleeding with hypogammaglobulinemia in adult: A case report and literature review  
*Tan B, Zhang SY, Wang YN, Li Y, Shi XH, Qian JM*
- 149 Large pelvic mass arising from the cervical stump: A case report  
*Zhang K, Jiang JH, Hu JL, Liu YL, Zhang XH, Wang YM, Xue FX*
- 157 Mechanical intestinal obstruction due to isolated diffuse venous malformations in the gastrointestinal tract: A case report and review of literature  
*Li HB, Lv JF, Lu N, Lv ZS*
- 168 Two-level percutaneous endoscopic lumbar discectomy for highly migrated upper lumbar disc herniation: A case report  
*Wu XB, Li ZH, Yang YF, Gu X*

- 175 Successful treatment of congenital palate perforation: A case report  
*Zhang JF, Zhang WB*
- 179 Calcitonin-negative neuroendocrine tumor of the thyroid with metastasis to liver-rare presentation of an unusual tumor: A case report and review of literature  
*Cai HJ, Wang H, Cao N, Huang B, Kong FL, Lu LR, Huang YY, Wang W*
- 188 Giant exophytic cystic adenomyosis with a levonorgestrel containing intrauterine device out of the uterine cavity after uterine myomectomy: A case report  
*Zhou Y, Chen ZY, Zhang XM*
- 194 Unusual presentation of bladder neuroblastoma in a child: A case report  
*Cai JB, Wang JH, He M, Wang FL, Xiong JN, Mao JQ, Li MJ, Zhu K, Liang JW*
- 200 Value of dynamic plasma cell-free DNA monitoring in septic shock syndrome: A case report  
*Liu JP, Zhang SC, Pan SY*
- 208 Sarcomatoid intrahepatic cholangiocarcinoma mimicking liver abscess: A case report  
*Wang Y, Ming JL, Ren XY, Qiu L, Zhou LJ, Yang SD, Fang XM*
- 217 Clinical characteristics on manifestation and gene mutation of a transient neonatal cyanosis: A case report  
*Yuan J, Zhu XP*
- 222 Six families with balanced chromosome translocation associated with reproductive risks in Hainan Province: Case reports and review of the literature  
*Chen YC, Huang XN, Kong CY, Hu JD*
- 234 Primary intestinal extranodal natural killer/T-cell lymphoma, nasal type: A case report  
*Dong BL, Dong XH, Zhao HQ, Gao P, Yang XJ*

**LETTER TO THE EDITOR**

- 242 Cluster headache as a manifestation of a stroke-like episode in a carrier of the MT-ND3 variant m.10158T>C  
*Finsterer J*

**ABOUT COVER**

Editorial Board Member of *World Journal of Clinical Cases*, Maddalena Zippi, MD, PhD, Doctor, Unit of Gastroenterology and Digestive Endoscopy, Sandro Pertini Hospital, Rome 00157, Italy

**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 PubMed, PubMed Central, Science Citation Index Expanded (also known as SciSearch®), and Journal Citation Reports/Science Edition. The 2019 Edition of Journal Citation Reports cites the 2018 impact factor for WJCC as 1.153 (5-year impact factor: N/A), ranking WJCC as 99 among 160 journals in Medicine, General and Internal (quartile in category Q3).

**RESPONSIBLE EDITORS FOR THIS ISSUE**

Responsible Electronic Editor: *Yan-Xia Xing*

Proofing Production Department Director: *Yun-Xiaojuan Wu*

**NAME OF JOURNAL**

*World Journal of Clinical Cases*

**ISSN**

ISSN 2307-8960 (online)

**LAUNCH DATE**

April 16, 2013

**FREQUENCY**

Semimonthly

**EDITORS-IN-CHIEF**

Dennis A Bloomfield, Bao-Gan Peng, Sandro Vento

**EDITORIAL BOARD MEMBERS**

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

**EDITORIAL OFFICE**

Jin-Lei Wang, Director

**PUBLICATION DATE**

January 6, 2020

**COPYRIGHT**

© 2020 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 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>

# Clinical characteristics on manifestation and gene mutation of a transient neonatal cyanosis: A case report

Jing Yuan, Xue-Ping Zhu

**ORCID number:** Jing Yuan (0000-0003-4803-3832); Xue-Ping Zhu (0000-0002-3502-7655).

**Author contributions:** Yuan J and Zhu XP wrote and edited the final manuscript; both authors have read and approved the final manuscript.

**Supported by** National Natural Science Foundation of China, No. 81771626.

**Informed consent statement:** Written informed consent was obtained from the patient's legal guardian(s) for publication of this case report and any accompanying images.

**Conflict-of-interest statement:** The authors declare that they have no conflicts of interest in relation to this manuscript.

**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 which was selected by an in-house editor and fully peer-reviewed by external reviewers. It is distributed in accordance with the Creative Commons Attribution Non Commercial (CC BY-NC 4.0) license, which permits others to distribute, remix, adapt, build upon this work non-commercially, and license their derivative works on different terms, provided the original work is properly cited and the use is non-commercial. See: <https://creativecommons.org/licenses/by-nc/4.0/>

**Jing Yuan, Xue-Ping Zhu,** Department of Neonatology, Children's Hospital of Soochow University, Suzhou 215025, Jiangsu Province, China

**Corresponding author:** Xue-Ping Zhu, MD, Chief Doctor, Department of Neonatology, Children's Hospital of Soochow University, 92 Zhongnan Street, Industrial Park, Suzhou 215025, Jiangsu Province, China. [zhuxueping4637@hotmail.com](mailto:zhuxueping4637@hotmail.com)

## Abstract

### BACKGROUND

We analyzed the main features of an infant diagnosed with temporary neonatal cyanosis in order to strengthen our understanding of the disease.

### CASE SUMMARY

We report a patient diagnosed with temporary neonatal cyanosis. The main clinical characteristics, gene mutation and treatment are discussed and a review of related literature was conducted. The neonate aged 1 d and 5 h was admitted to hospital due to cyanosis after birth. The main clinical manifestation was cyanosis, which was not improved by auxiliary ventilation and the patient showed no obvious shortness of breath or methemoglobinemia. Gene mutation analysis showed a heterozygous c.190C>T mutation in the *HBG2* gene associated with transient neonatal cyanosis, which was derived from his mother. Symptomatic supportive treatment was given for 2 mo. The neonate was discharged and gradually improved with oral administration of vitamin C and vitamin B2 for 2 wk.

### CONCLUSION

There is no special treatment for temporary neonatal cyanosis caused by heterozygous mutation of the *HBG2* gene.

**Key words:** Temporary neonatal cyanosis; *HBG2*; Genetic mutation; Methemoglobinemia; Cyanosis; Case report

©The Author(s) 2020. Published by Baishideng Publishing Group Inc. All rights reserved.

**Core tip:** We report a case diagnosed with temporary neonatal cyanosis. The main characteristics of the clinical manifestations and gene mutation, and treatments were assessed and a review of the related literature was performed, which may help to improve the understanding of the disease.



<http://creativecommons.org/licenses/by-nc/4.0/>

**Manuscript source:** Unsolicited manuscript

**Received:** September 30, 2019

**Peer-review started:** September 30, 2019

**First decision:** October 23, 2019

**Revised:** November 16, 2019

**Accepted:** November 30, 2019

**Article in press:** November 30, 2019

**Published online:** January 6, 2020

**P-Reviewer:** Akbulut S, Skok P

**S-Editor:** Dou Y

**L-Editor:** Webster JR

**E-Editor:** Liu JH



**Citation:** Yuan J, Zhu XP. Clinical characteristics on manifestation and gene mutation of a transient neonatal cyanosis: A case report. *World J Clin Cases* 2020; 8(1): 217-221

**URL:** <https://www.wjnet.com/2307-8960/full/v8/i1/217.htm>

**DOI:** <https://dx.doi.org/10.12998/wjcc.v8.i1.217>

## INTRODUCTION

Temporary neonatal cyanosis<sup>[1]</sup> is characterized by varying degrees of bruising on the body, around the mouth, or at the extremities, with or without other symptoms or signs. Fetal hemoglobin (Hb) chains are defective, and the affinity for oxygen is weakened, resulting in neonatal cyanosis. In some patients, anemia occurs due to abnormal red blood cells (RBCs). Cyanosis is usually self-healing after 5 to 6 mo or earlier when the adult  $\beta$ -globin chain replaces the fetal  $\gamma$ -globin chain. There are few similar reports both at home and abroad, and scholars such as Celeste Bento reported that the disease is gene-related<sup>[2]</sup>. This article reports the clinical features, diagnosis and treatment of a child with temporary neonatal cyanosis associated with *HBB2* gene mutation. The child was diagnosed in the neonatal intensive care unit of the Children's Hospital of Soochow University, and the purpose of this article is to improve our understanding of this disease.

## CASE PRESENTATION

### Chief complaints

Discovering bruises for 12 h.

### History of present illness

Proband Male, born 29 h at the time of the visit. He was treated for "discovering bruises for 12 h".

### History of past illness

G1P1, gestational age 38<sup>+</sup>4 wk, born by cesarean section at Taicang First People's Hospital on October 18, 2017, birth weight 3140 g, Apgar score at 1 min 10 points, Apgar score at 5 min 10 points, amniotic fluid clear; however, the quantity is unknown. The placenta status is unknown, and a history of rescue suffocation is denied.

### Personal and family history

Parents are not close relatives, and his mother had similar symptoms when she was a baby.

### Physical examination upon admission

His body temperature was 36.5 °C, pulse 147 beats/min, respiratory rate 56 breaths/min, the oxygen pulse of hood oxygen was 81%, body weight 3.02 kg, length 48 cm, head circumference 34 cm, and chest circumference 32 cm. The neonate had full-term appearance, color cyan, scalp edema, anterior sputum soft, size approximately 1.5 cm × 1.5 cm tension was not high, perioral cyanosis, no spitting, soft neck, no polypnea, no obvious inspiratory three concave sign, the lungs showed thick breath sounds, no rales were heard in the lungs, heart rhythm showed no obvious murmur in the precordium, abdomen soft, liver and spleen under the ribs, cold extremities, nerve reflex could be induced.

### Laboratory examinations

White blood cell count  $7.75 \times 10^9/L$ , RBC  $4.00 \times 10^{12}/L$ , Hb 136 g/L, platelet distribution width 9.40%, blood gas analysis showed  $Ca^{2+}$  1.13 mmol/L,  $Cl^-$  111 mmol/L,  $BE^-$  3.5 mmol/L, lactic acid 6.0 mmol/L, total bilirubin 182 mmol/L, whole blood oxygen content 6.5 mmol/L, oxygen partial pressure 140 mmHg, blood pH 7.438, carbon dioxide partial pressure 21.6 mmHg, methemoglobin (MetHb) fluctuation between 2.6% and 5.7%. Glucose-6-phosphate dehydrogenase activity was 1.45, which was within the normal range. Complete hemolysis examination showed Hb F > 40%, Hb A2 0%, Hb electrophoresis showed no abnormal zone, and a small abnormal S window peak was observed by HPLC.

### Imaging examinations

Echocardiography demonstrated an atrial septal defect (2.7 mm) and patent ductus arteriosus. Chest and abdominal imaging showed that the texture of both lungs was darkened and blurred. Hepatobiliary, pancreatic, spleen, kidney and cranial ultrasound were normal, and scrotum ultrasound showed a right testicular hydrocele.

### **Hb-related gene test**

The patient (SH171214132), chr11:5275647, showed a heterozygous mutation of c.190C>T; the father (SH171214133), chr11:5275647 showed no mutation; the mother (SH171214134), chr11:5275647 showed a heterozygous mutation of c.190C>T (Figure 1).

---

## **FINAL DIAGNOSIS**

Neonatal methemoglobinemia? Neonatal pneumonia; congenital foramen ovale; congenital atrial septal defect; neonatal hyperbilirubinemia; right testicular hydrocele.

---

## **TREATMENT**

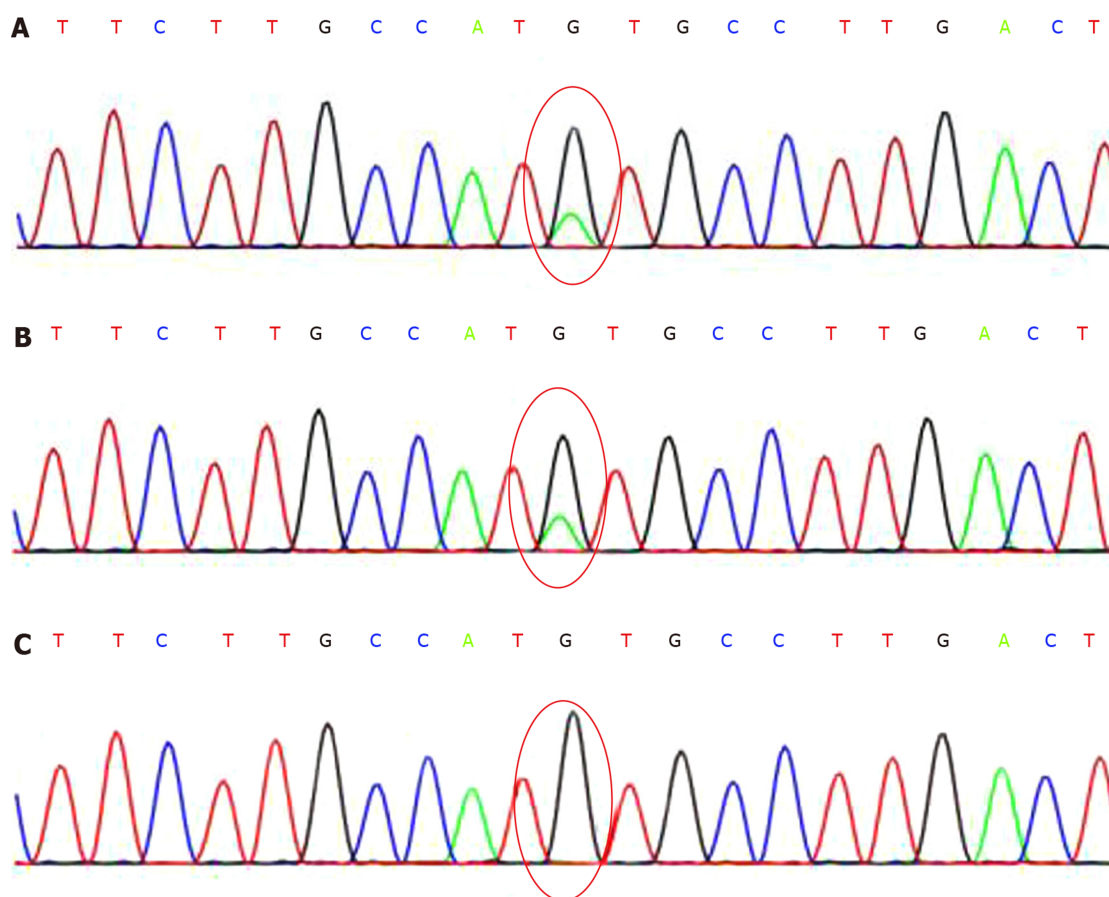
When the patient was admitted to hospital, the oxygen pulse of hood oxygen was maintained at 75%-78%, perioral and facial region cyanosis was observed, lowest blood pressure was 54/37 mmHg, saline was expanded, assisted ventilation was administered, and the patient was actively treated. The percutaneous oxygen pulse still fluctuated around 72%-85%, and the blood oxygen saturation following arterial blood gas analysis was 96.0%-100%, and the oxygen partial pressure was maintained in the normal range. After hospitalization, the MetHb content was high, fluctuating between 2.6% and 5.7%. After clinical diagnosis, vitamin C and vitamin B2 treatment was given, but there was no significant improvement in percutaneous oxygen pulse saturation, and Hb-related gene detection was performed. The patient was diagnosed with temporary neonatal cyanosis. The clinical phenotype showed anemia, cyanosis, and methemoglobinemia. The oxygen pulse during assisted ventilation did not increase significantly. The child did not have breathing difficulties, no obvious inspiratory three concave sign, breathing was stable, and arterial blood gas analysis showed that oxygen partial pressure and oxygen saturation were maintained in the normal range. The existence of genetic problems was considered. Gene examination was carried out and a transfusion to correct anemia was administered; however, percutaneous oxygen saturation showed no obvious increase. After 3 d, assisted ventilation was discontinued and replaced with hood oxygen. The percutaneous oxygen pulse was maintained at approximately 75%, and his respiration rate was 47 breaths/min. When hood oxygen was stopped, percutaneous oxygen saturation was maintained between 70% and 80%, with no obvious dyspnea, no dysphoria or other clinical manifestations. Seventeen days after admission, he was discharged on vitamin C and vitamin B2 therapy. Hyperbilirubinemia occurred during the disease course, and phototherapy was administered due to jaundice, and the ganglioside nutrient nervous system was used for rehabilitation. Vitamin C and vitamin B2 were discontinued 2 wk after discharge. Perioral and facial cyanosis gradually subsided. After 189 d of follow-up, the child's growth and development were normal, and his body weight was 9 kg. No recurrence was observed.

---

## **OUTCOME AND FOLLOW-UP**

Gene mutation analysis identified a heterozygous mutation in the exon region of the *HBG2* gene: c.190C>T (nucleotide number 190 in the coding region was changed from cytosine to thymine), resulting in amino acid change p.H64Y (No. 64) in the patient. The amino acid was changed from histidine to tyrosine, which is a missense mutation<sup>[3]</sup>. This mutation does not belong to the polymorphic site, and the frequency of occurrence in the population is low. The Human Gene Mutation Database Professional Edition has reported an association with the Hb variant<sup>[4]</sup>. After verification by the family, the father of the subject showed no variation at this site, and the mother of the subject showed a heterozygous variation at the same site<sup>[5]</sup>. This site is a suspected mutation. If the mother had similar symptoms, it should theoretically cause illness; if the mother had no similar symptoms, the possibility of the site causing mutation is very small. In this case, no specific disease-related copy number variation was found at the exon level.





**Figure 1 Hemoglobin-related gene test.** A: The patient (SH171214132), chr11:5275647 had a heterozygous mutation of c.190C>T; B: The father (SH171214133), chr11:5275647 had no mutations; C: The mother (SH171214134), chr11:5275647 had a heterozygous mutation of c.190C>T.

## DISCUSSION

Neonatal cyanosis is a common sign in the early neonatal period, and it is one of the most serious clinical manifestations<sup>[6]</sup>. Whether it is a gas exchange disorder caused by cardiopulmonary disease or a localized reduction in Hb around the tissue capillaries which can cause cyanosis is still unknown<sup>[7]</sup>. Skin mucosa in a child after birth which shows persistent cyanosis and percutaneous oxygen saturation less than 90%, with the lowest at 60%, is classified as pathological cyanosis. The current reasons for cyanosis are complicated. After hospitalization, the patient received oxygen, continuous positive airway pressure assisted ventilation, and mechanical ventilation. Repeated blood gas analysis showed that arterial oxygen saturation and oxygen partial pressure were similar to non-oxygenation, indicating that assisted ventilation did not improve hypoxia, and heart color Doppler ultrasound suggested congenital heart disease; however, there was no obvious pulmonary hypertension, so central cyanosis was excluded. The child's body temperature was normal, the temperature of the extremities was normal, the Hb content was not high, clinical manifestations of shock or heart failure were not obvious, and there was no obvious local circulation failure; thus, peripheral cyanosis was excluded. Therefore, considering other factors which may cause bruising, repeated blood gas analysis was performed after admission and showed that the MetHb concentration was significantly higher than the normal value, the highest being 8.2%. Blood tests showed that the child's blood color was brownish red, indicating that the heart status, B-ultrasound, chest X-ray and other related examinations, in addition to congenital cyanosis, low oxygen pulse and other factors, led to the diagnosis of neonatal methemoglobinemia. MetHb is a genetic factor and a toxic compound produced by RBCs. Hb is composed of globin and heme, which combine with oxygen to form oxygenated Hb. MetHb is a Hb derivative in which the iron ion of the heme group in deoxygenated or oxygenated Hb is oxidized from ferrous to ferric iron. The MetHb reduction process mainly depends on NADH-dependent Cytochrome 65 reductase, and the absence of this enzyme leads to abnormal accumulation of MetHb. When MetHb is overproduced, the ratio of MetHb and Hb is unbalanced and methemoglobinemia is formed<sup>[8]</sup>. Following hospitalization

for active oxygen therapy, the condition of the child improved and the diagnosis was clinically supported. Methylene blue is recommended for methemoglobinemia, but the child was also thought to have an Hb disease and possible genetic problems. Blood gas analysis after treatment showed that the content of MetHb was still high. So, the parents of the child were asked about the history of pregnancy. The mother of the child likes to eat spicy food, and was also born with a similar condition to the child; thus, it was considered that the child's cyanosis was closely related to MetHb content.

In this case of suspected methemoglobinemia, a gene mutation was identified and the patient was diagnosed with temporary neonatal cyanosis, and clinical follow-up showed that the condition gradually improved. The recurrence of unexplained cyanosis and the repeated rise in MetHb can be diagnosed by genetic testing. The *HBG2* gene is associated with transient neonatal cyanosis<sup>[9]</sup>, and is reported to occur *via* autosomal dominant inheritance. Theoretically, chromogenic pathogenic mutations can cause the disease<sup>[10]</sup>, and the clinical manifestations are similar to this case. In this patient, genetic testing detected a heterozygous mutation in the transient neonatal cyanosis-related gene *HBG2*, which was derived from his mother and is X-linked. The genetic characteristic, in this case, temporary neonatal cyanosis, was treated with vitamin C and vitamin B2, and after active oxygen therapy and anti-oxidation therapy, perioral and facial cyanosis gradually subsided. Usually cyanosis can take 5 to 6 mo to resolve with possible earlier self-healing, as the adult  $\beta$ -globin chain replaces the fetal  $\gamma$ -globin chain. In this case, although early treatment was administered, early genetic testing was not carried out. Temporary neonatal cyanosis should be diagnosed early with early treatment to avoid other diseases and improve the prognosis.

## CONCLUSION

Transient neonatal cyanosis caused by heterozygous mutation of the *HBG2* gene has no specific treatment and can resolve itself. With the development of genetic diagnosis, early diagnosis of neonatal diseases can be achieved relatively easily. The formation of a neonatal disease is a complex pathological process, and its prognosis will also be determined by the progress of pathological changes. The general condition of the patient should be comprehensively evaluated and early conservative treatment is recommended.

## REFERENCES

- 1 **Dainer E**, Shell R, Miller R, Atkin JF, Pastore M, Kutlar A, Zhuang L, Holley L, Davis DH, Kutlar F. Neonatal cyanosis due to a novel fetal hemoglobin: Hb F-Circleville [Gamma63(E7)His-->Leu, CAT>CTT]. *Hemoglobin* 2008; **32**: 596-600 [PMID: [19065339](#) DOI: [10.1080/03630260802507915](#)]
- 2 **Bento C**, Magalhães Maia T, Carvalhais I, Moita F, Abreu G, Relvas L, Pereira A, Fabela Neves J, Ribeiro ML. Transient neonatal cyanosis associated with a new Hb F variant: Hb F viseu. *J Pediatr Hematol Oncol* 2013; **35**: e77-e80 [PMID: [22935660](#) DOI: [10.1097/MPH.0b013e3182667be3](#)]
- 3 **Kedar PS**, Gupta V, Warang P, Chiddarwar A, Madkaikar M. Novel mutation (R192C) in CYB5R3 gene causing NADH-cytochrome b5 reductase deficiency in eight Indian patients associated with autosomal recessive congenital methemoglobinemia type-I. *Hematology* 2018; **23**: 567-573 [PMID: [29482478](#) DOI: [10.1080/10245332.2018.1444920](#)]
- 4 **Adachi K**, Asakura T, Schwartz E. Aggregation of hemoglobin S and hemoglobin CHarlem with nonsickle hemoglobin in concentrated phosphate buffer. *Blood* 1980; **55**: 494-500 [PMID: [7357080](#) DOI: [10.1182/blood.V55.3.494.bloodjournal553494](#)]
- 5 **Giordano SJ**, Kaftory A, Steggle AW. A splicing mutation in the cytochrome b5 gene from a patient with congenital methemoglobinemia and pseudohermaphroditism. *Hum Genet* 1994; **93**: 568-570 [PMID: [8168836](#) DOI: [10.1007/bf00202825](#)]
- 6 **Qureshi AU**, Latiff HA, Sivalingam S. Persistent valve of systemic venous sinus: a cause of neonatal cyanosis. *Cardiol Young* 2014; **24**: 756-759 [PMID: [24016801](#) DOI: [10.1017/S1047951113001200](#)]
- 7 **Ko HS**, Chen MR, Lin YC. A huge Chiari network presenting with persistent cyanosis in a neonate. *Pediatr Cardiol* 2011; **32**: 239-240 [PMID: [21210098](#) DOI: [10.1007/s00246-010-9857-8](#)]
- 8 **Wright RO**, Lewander WJ, Woolf AD. Methemoglobinemia: etiology, pharmacology, and clinical management. *Ann Emerg Med* 1999; **34**: 646-656 [PMID: [10533013](#) DOI: [10.1016/s0196-0644\(99\)70167-8](#)]
- 9 **Koga M**, Inada S, Yoshino K, Miyazaki A. IFCC-HbA1c May Be More Useful than NGSP-HbA1c in the Comparison between HbA1c Values in Variant Hemoglobin with a Mutation on  $\alpha$  Chain and  $\beta$  Chain. *Ann Clin Lab Sci* 2019; **49**: 650-655 [PMID: [31611209](#)]
- 10 **Yang L**, Liu X, Li Z, Zhang P, Wu B, Wang H, Hu L, Cheng G, Wang L, Zhou W. Genetic aetiology of early infant deaths in a neonatal intensive care unit. *J Med Genet* 2019 [PMID: [31501239](#) DOI: [10.1136/jmedgenet-2019-106221](#)]



Published By Baishideng Publishing Group Inc  
7041 Koll Center Parkway, Suite 160, Pleasanton, CA 94566, USA  
Telephone: +1-925-2238242  
E-mail: [bpgoffice@wjgnet.com](mailto:bpgoffice@wjgnet.com)  
Help Desk: <https://www.f6publishing.com/helpdesk>  
<https://www.wjgnet.com>

