World Journal of *Clinical Cases*

World J Clin Cases 2021 April 26; 9(12): 2696-2950





Published by Baishideng Publishing Group Inc

W J C C World Journal of Clinical Cases

Contents

Thrice Monthly Volume 9 Number 12 April 26, 2021

MINIREVIEWS

- 2696 Standardization of critical care management of non-critically ill patients with COVID-19 Wang CS, Gao Y, Kang K, Fei DS, Meng XL, Liu HT, Luo YP, Yang W, Dai QQ, Gao Y, Zhao MY, Yu KJ
- 2703 Mediastinal lymphadenopathy in COVID-19: A review of literature Taweesedt PT, Surani S
- Polycystic ovary syndrome: Pathways and mechanisms for possible increased susceptibility to COVID-19 2711 Ilias I, Goulas S, Zabuliene L

ORIGINAL ARTICLE

Clinical and Translational Research

2721 Circulating tumor cells with epithelial-mesenchymal transition markers as potential biomarkers for the diagnosis of lung cancer

Jiang SS, Mao CG, Feng YG, Jiang B, Tao SL, Tan QY, Deng B

Retrospective Study

2731 Management and implementation strategies of pre-screening triage in children during coronavirus disease 2019 pandemic in Guangzhou, China

Shi X, Cai YT, Cai X, Wen XL, Wang JY, Ma WC, Shen J, Wu JX, Liu HY, Sun J, He PQ, Lin Y, Zhao DY, Li PQ

2739 Clinicopathological features of superficial CD34-positive fibroblastic tumor Ding L, Xu WJ, Tao XY, Zhang L, Cai ZG

2751 Application of a rapid exchange extension catheter technique in type B2/C nonocclusive coronary intervention via a transradial approach

Wang HC, Lu W, Gao ZH, Xie YN, Hao J, Liu JM

SYSTEMATIC REVIEWS

Paradoxical relationship between proton pump inhibitors and COVID-19: A systematic review and meta-2763 analysis

Zippi M, Fiorino S, Budriesi R, Micucci M, Corazza I, Pica R, de Biase D, Gallo CG, Hong W

META-ANALYSIS

2778 Predictive risk factors for recollapse of cemented vertebrae after percutaneous vertebroplasty: A metaanalysis

Ma YH, Tian ZS, Liu HC, Zhang BY, Zhu YH, Meng CY, Liu XJ, Zhu QS



| Co | nte | ٥n | tc |
|----|-----|----|----|
| υJ | пu | | ιs |

CASE REPORT

| 2791 | Malignant pheochromocytoma with cerebral and skull metastasis: A case report and literature review | |
|------|---|--|
| | Chen JC, Zhuang DZ, Luo C, Chen WQ | |
| 2801 | Unresectable esophageal cancer treated with multiple chemotherapies in combination with chemoradiotherapy: A case report | |
| | Yura M, Koyanagi K, Hara A, Hayashi K, Tajima Y, Kaneko Y, Fujisaki H, Hirata A, Takano K, Hongo K, Yo K, Yoneyama K, Tamai Y, Dehari R, Nakagawa M | |
| 2811 | Role of positron emission tomography in primary carcinoma ex pleomorphic adenoma of the bronchus: A case report | |
| | Yang CH, Liu NT, Huang TW | |
| 2816 | Positive reverse transcription-polymerase chain reaction assay results in patients recovered from COVID- 19: Report of two cases | |
| | Huang KX, He C, Yang YL, Huang D, Jiang ZX, Li BG, Liu H | |
| 2823 | Laryngeal myxoma: A case report | |
| | Yu TT, Yu H, Cui Y, Liu W, Cui XY, Wang X | |
| 2830 | Prostate stromal tumor with prostatic cysts after transurethral resection of the prostate: A case report | |
| | Zhao LW, Sun J, Wang YY, Hua RM, Tai SC, Wang K, Fan Y | |
| 2838 | Intramuscular hematoma in rhabdomyolysis patients treated with low-molecular-weight heparin: Report of two cases | |
| | Yuan SY, Xie KF, Yang J | |
| 2845 | Partial response to Chinese patent medicine Kangliu pill for adult glioblastoma: A case report and review of the literature | |
| | Sun G, Zhuang W, Lin QT, Wang LM, Zhen YH, Xi SY, Lin XL | |
| 2854 | Behcet's disease manifesting as esophageal variceal bleeding: A case report | |
| | Xie WX, Jiang HT, Shi GQ, Yang LN, Wang H | |
| 2862 | Successful endoscopic surgery for emphysematous pyelonephritis in a non-diabetic patient with autosomal dominant polycystic kidney disease: A case report | |
| | Jiang Y, Lo R, Lu ZQ, Cheng XB, Xiong L, Luo BF | |
| 2868 | Robotically assisted removal of pelvic splenosis fifty-six years after splenectomy: A case report | |
| | Tognarelli A, Faggioni L, Erba AP, Faviana P, Durante J, Manassero F, Selli C | |
| 2874 | Pulmonary alveolar proteinosis complicated with nocardiosis: A case report and review of the literature | |
| | Wu XK, Lin Q | |
| 2884 | Detection of EGFR-SEPT14 fusion in cell-free DNA of a patient with advanced gastric cancer: A case report | |
| | Kim B, Kim Y, Park I, Cho JY, Lee KA | |



| Carta | World Journal of Clinical Cases |
|--------|---|
| Conter | Thrice Monthly Volume 9 Number 12 April 26, 2021 |
| 2890 | Timing of convalescent plasma therapy-tips from curing a 100-year-old COVID-19 patient using convalescent plasma treatment: A case report |
| | Liu B, Ren KK, Wang N, Xu XP, Wu J |
| 2899 | Torsades de pointes episode in a woman with high-grade fever and inflammatory activation: A case report |
| | Qiu H, Li HW, Zhang SH, Zhou XG, Li WP |
| 2908 | Salivary duct carcinoma of the submandibular gland presenting a diagnostic challenge: A case report |
| | Uchihashi T, Kodama S, Sugauchi A, Hiraoka S, Hirose K, Usami Y, Tanaka S, Kogo M |
| 2916 | Allogeneic hematopoietic stem cell transplantation in a 3-year-old boy with congenital pyruvate kinase deficiency: A case report |
| | Ma ZY, Yang X |
| 2923 | Congenital bilateral cryptorchidism in an infant conceived after maternal breast cancer treatment: A case report |
| | Hu WK, Liu J, Liu RX, Liu XW, Yin CH |
| 2930 | Sclerosing polycystic adenosis of the submandibular gland: Two case reports |
| | Wu L, Wang Y, Hu CY, Huang CM |
| 2937 | Budd-Chiari syndrome associated with liver cirrhosis: A case report |
| | Ye QB, Huang QF, Luo YC, Wen YL, Chen ZK, Wei AL |
| 2944 | Separated root tip formation associated with a fractured tubercle of dens evaginatus: A case report |
| | Wu ZF, Lu LJ, Zheng HY, Tu Y, Shi Y, Zhou ZH, Fang LX, Fu BP |
| | |
| | |
| | |



Contents

Thrice Monthly Volume 9 Number 12 April 26, 2021

ABOUT COVER

Editorial Board Member of World Journal of Clinical Cases, Jing Liu, MD, PhD, Chief Doctor, Professor, Department of Neonatology and NICU, Beijing Chaoyang District Maternal and Child Healthcare Hospital, Beijing 100021, China. liujingbj@live.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: Ji-Hong Lin; Production Department Director: Xiang Li; Editorial Office Director: Jin-Lei Wang.

| NAME OF JOURNAL | |
|---|---|
| World Journal of Clinical Cases | https://www.wjgnet.com/bpg/gerinfo/204 |
| ISSN | GUIDELINES FOR ETHICS DOCUMENTS |
| ISSN 2307-8960 (online) | https://www.wjgnet.com/bpg/GerInfo/287 |
| LAUNCH DATE | GUIDELINES FOR NON-NATIVE SPEAKERS OF ENGLISH |
| April 16, 2013 | https://www.wjgnet.com/bpg/gerinfo/240 |
| FREQUENCY | PUBLICATION ETHICS |
| Thrice Monthly | https://www.wjgnet.com/bpg/GerInfo/288 |
| EDITORS-IN-CHIEF | PUBLICATION MISCONDUCT |
| Dennis A Bloomfield, Sandro Vento, Bao-Gan Peng | https://www.wjgnet.com/bpg/gerinfo/208 |
| EDITORIAL BOARD MEMBERS | ARTICLE PROCESSING CHARGE |
| https://www.wjgnet.com/2307-8960/editorialboard.htm | https://www.wjgnet.com/bpg/gerinfo/242 |
| PUBLICATION DATE | STEPS FOR SUBMITTING MANUSCRIPTS |
| April 26, 2021 | https://www.wjgnet.com/bpg/GerInfo/239 |
| COPYRIGHT | ONLINE SUBMISSION |
| © 2021 Baishideng Publishing Group Inc | https://www.f6publishing.com |

© 2021 Baishideng Publishing Group Inc. All rights reserved. 7041 Koll Center Parkway, Suite 160, Pleasanton, CA 94566, USA E-mail: bpgoffice@wjgnet.com https://www.wjgnet.com



W J C C World Journal of Clinical Cases

World Journal of

Submit a Manuscript: https://www.f6publishing.com

World J Clin Cases 2021 April 26; 9(12): 2916-2922

DOI: 10.12998/wjcc.v9.i12.2916

ISSN 2307-8960 (online)

CASE REPORT

Allogeneic hematopoietic stem cell transplantation in a 3-year-old boy with congenital pyruvate kinase deficiency: A case report

Zhong-Yang Ma, Xue Yang

ORCID number: Zhong-Yang Ma 0000-0002-1365-3647; Xue Yang 0000-0002-5020-8550.

Author contributions: Yang X conceived and coordinated the study, designed, performed, and analyzed the experiments, and wrote the paper; Ma ZY carried out the data collection and analysis, and revised the paper; all authors reviewed the results and approved the final version of the manuscript.

Informed consent statement:

Informed written consent was obtained from the patient's parents for publication of this report.

Conflict-of-interest statement: The authors declare that they have no conflict of interest to report.

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

Zhong-Yang Ma, Xue Yang, Department of Pediatrics, Key Laboratory of Birth Defects and Related Diseases of Women and Children, West China Second University Hospital, Sichuan University, Chengdu 610041, Sichuan Province, China

Corresponding author: Xue Yang, MS, Doctor, Department of Pediatrics, Key Laboratory of Birth Defects and Related Diseases of Women and Children, West China Second University Hospital, Sichuan University, No. 20 Section 3, Renmin South Road, Chengdu 610041, Sichuan Province, China. xueyang@scu.edu.cn

Abstract

BACKGROUND

The understanding regarding genetic variation, pathophysiology, and complications associated with pyruvate kinase deficiency (PKD) in red blood cells has been explained largely, and supportive treatment is currently the main management strategy. Etiotropic managements, including transplantation and genome editing, supplying for substitute dugs of the pyruvate kinase, are all under research.

CASE SUMMARY

We herein report a 3-year-old boy with severe transfusion-dependent PKD cured by unrelated identical peripheral blood stem cell transplantation (PBSCT). Hemoglobin was corrected to a normal level by gene correction after PBSCT, with no complication related to the transplantation.

CONCLUSION

Hematopoietic stem cell transplantation could be a substitute for transfusiondependent PKD.

Key Words: Pyruvate kinase deficiency; Transfusions; Hematopoietic stem cell transplantation; Peripheral blood stem cells; Peripheral blood stem cell transplantation; Case report

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

Core Tip: We herein report a 3-year-old boy with severe transfusion-dependent



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: htt p://creativecommons.org/License s/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): C Grade D (Fair): 0 Grade E (Poor): 0

Received: December 14, 2020 Peer-review started: December 14. 2020 First decision: January 7, 2021 Revised: January 21, 2021 Accepted: February 24, 2021 Article in press: February 24, 2021 Published online: April 26, 2021

P-Reviewer: Segovia JC, Sun K S-Editor: Zhang H L-Editor: Wang TQ P-Editor: Li X



pyruvate kinase deficiency (PKD) cured by unrelated identical peripheral blood stem cell transplantation. Hematopoietic stem cell transplantation could be a substitute for severe transfusion-dependent PKD, and should be carried out in the early stage of life. If there are no identical siblings available, unrelated identical peripheral blood stem cells might also be an alternative option.

Citation: Ma ZY, Yang X. Allogeneic hematopoietic stem cell transplantation in a 3-year-old boy with congenital pyruvate kinase deficiency: A case report. World J Clin Cases 2021; 9(12): 2916-2922

URL: https://www.wjgnet.com/2307-8960/full/v9/i12/2916.htm DOI: https://dx.doi.org/10.12998/wjcc.v9.i12.2916

INTRODUCTION

Pyruvate kinase deficiency (PKD) is the most frequent glycolytic enzyme defect that causes hereditary non-spherocytic hemolytic anemia^[1]. Glycolysis is the only way for red blood cells to synthesize ATP, which subsequently affects the longevity of red blood cells^[2]. Too much destruction of premature red blood cells, exceeding the compensatory ability of bone marrow hematopoiesis, leads to hemolytic anemia. The degree of hemolysis varies widely, from fully compensated forms to transfusiondependent anemia. With no relationship to the severity of hemolysis, neonates with PKD would suffer from severe or extreme hyperbilirubinemia, and some of whom even require exchange transfusion to avoid the occurrence of kernicterus^[2].

Based on evidences of over-destruction of red blood cells (jaundice, anemia, etc.) and increased hematopoiesis (increased immature reticulocyte count, enlargement of liver and spleen, etc.), the diagnosis of chronic hemolytic anemia could be conducted. Further evaluations of pyruvate kinase activity and mutations in the PKLR gene lead to the diagnosis of PKD.

The treatment for PKD is mainly supportive, which consists of regular red blood cell transfusions, splenectomy, and chelation therapy for iron overload^[2]. There are several methods that might cure PKD, including allogeneic hematopoietic stem cell transplantation (allo-HSCT)^[3-5], gene therapy^[6], and Mitapivat (a small-molecule allosteric activator of red-cell pyruvate kinase)^[7], but all are under researching.

We herein report a 3-year-old boy born with severe hyperbilirubinemia who had required exchange-transfusion to prevent the occurrence of bilirubin encephalopathy and received red blood cell transfusion monthly before the transplantation. Due to severe transfusion-dependent hemolytic anemia, peripheral blood stem cell transplantation (PBSCT) was performed at the age of 3 for him.

CASE PRESENTATION

Chief complaints

The patient was one month shy of his third birthday, and his parents visited the hematology outpatient department for consulting about allo-HSCT.

History of present illness

Three years ago, the patient's mother found decreased fetal movement in her 39+2 wk of menopause without any drug use, and soon she visited her doctor locally. Due to high fetal heart rate (170-180 beats per min), he was born by an emergency cesarean section with yellow sclera and skin, pallor, and tachycardias. Soon he was transferred to the neonatal intensive care unit in another hospital and some examinations were done, like total blood cell count, biochemistry, and blood grouping. The results revealed that the red blood cell count and hemoglobin were extremely low, while his total bilirubin and unconjugated bilirubin were too high. He was diagnosed with neonatal hemolytic jaundice, and underwent blood exchange transfusion as soon as he was admitted, after which he stayed in the hospital for phototherapy in the first 12 d.

His parents observed yellow sclera and skin, pallor, and tachycardia again when he was around 2 mo old, and they visited the outpatient department of our hospital. The



total blood cell count showed severe anemia again needing transfusion.

Personal and family history

The patient's parents had mild anemia without dizziness, syncope, fatigue, etc.

Physical examination

The patient had a mild anemia appearance, and superficial lymph nodes were not palpable. Sclera was slightly yellow. Physical examination of the heart and lung showed no abnormality. The abdomen was soft and the liver and spleen were not palpable under the ribs.

Laboratory examinations

The gene sequencing showed that there were compound heterozygous mutations of the PKLR gene, which were obtained one from his mother and the other from his father, contributing to severe hemolytic anemia. The details of the laboratory examinations before the hematopoietic stem cell transplantation are shown in Table 1.

FINAL DIAGNOSIS

The patient was diagnosed with congenital pyruvate kinase deficiency.

TREATMENT

After being diagnosed with PKD, the patient visited a hospital for transfusion every 35 d to maintain his hemoglobin level above 90 g/L. As he grew up, the amount of blood needed increased and the transfusion intervals became shorter, especially when he was attacked by cold. At the worst of time, he had to receive transfusions every 20 d. At the age of 1 year and 8 mo, his serum ferritin reached 1085.7 ng/L, and he started iron removal therapy. Due to huge financial burden for the family and inconvenience of regular transfusion, his parents applied for allo-HSCT for this patient. Luckily, there was a matched-human leukocyte antigen-identical hematopoietic stem cell donor for him. And PBSCT was done on this 3-year-old boy.

OUTCOME AND FOLLOW-UP

The transplantation was absolutely successful. Engraftment of granulocytes and platelets was on post-day 13 and post-day 16, respectively, and he was discharged on post-day 31. Follow-up for 1 year showed no acute or chronic graft vs host disease (GvHD) and severe infection. The total blood cell count post transplantation is shown in Table 2.

DISCUSSION

Regular transfusion is a usual means to maintain the life of patients with severe hemolytic anemia. However, there are several puzzling problems in the management of patients with PKD, for example, when to initiate the transfusion, at which level of pre-transfusion hemoglobin should be maintained, and how long interval between transfusions might be good for patients with PKD. What's more, HSCT has been reported as an effective treatment for severe transfusion-dependent hemolytic anemia, but the indication of HSCT in PKD patients is not exact. We herein discuss some problems that we met during the management of this patient.

Could physicians copy the therapeutic model of thalassemia in the management of PKD?

Thalassemia is a kind of hemolytic anemia, in which the normal ratio of alpha-globin to beta-globin production is disrupted due to a disease-causing variant in one or more of the genes coding the hemoglobin. Patients with thalassemia major also need transfusion regularly. The similarity and difference between thalassemia and PKD are



WJCC | https://www.wjgnet.com

| Date | CBC | | |
|----------------------------|--------------------------------|-------------------------|--|
| 3 h and 56 min after birth | Blood cell count | | |
| | RBC | $1.6 \times 10^{9}/L$ | |
| | MCV | 132.5 fL | |
| | MCH | 40.6 pg | |
| | MCHC | 307 g/L | |
| | HGB | 65 g/L | |
| | Reticulocyte count | $0.477 \times 10^6 / L$ | |
| | Reticulocyte (%) | 29.78% | |
| | Bio-chemistry | | |
| | ALT | 55.0 U/L | |
| | AST | 273.0 U/L | |
| | Total bilirubin | 175.4 μmol/l | |
| | Unconjugated bilirubin | 159.8 μmol/L | |
| | Blood group | | |
| | Group O and Rh positive | | |
| | Blood group antibody screening | Negative | |
| | Others | | |
| | Coombs test | Negative | |
| | Free antibody test | Negative | |
| | Antibody release test | Negative | |
| 12 d old | | | |
| | RBC | $3.7 \times 10^{9}/L$ | |
| | HGB | 111 g/L | |
| 2 mo old | | | |
| | HGB | HB 57 g/L | |

CBC: Complete blood count; RBC: Red blood cells; MCV: Merkel cell polyomavirus; MCH: Melanin-concentrating hormone; MCHC: Mean corpuscular hemoglobin concentration; HGB: Blood hemoglobin; ALT: Alternative lengthening of telomeres; AST: Aspartate aminotransferase.

> shown in Table 3. There are numbers of practical guidelines on the management of thalassemia. "Hyper-transfusion" (in the United States) or "moderate-transfusion" (in Europe) was used to maintain the hemoglobin level above a set nadir with a goal of avoiding complications. Since both of PKD and thalassemia major are transfusiondependent chronic hemolytic anemias, we come up with the idea that the management of thalassemia might be suitable for patients with PKD.

> The patient in this case report developed severe hemolysis, and the HGB decreased to 57 g/L when he was two months old. Later he got 1 unit red blood cells monthly to maintain the level of HGB above 90 g/L. There were no obvious compensated signs of hematopoiesis increase and he obtained a good result from allo-HSCT later. But one fact is that, different from thalassemia, patients with PKD, with increased red cell 2,3-DPG content that enhances oxygen unloading from hemoglobin, might tolerate moderate severe anemia with few symptoms. Thus, whether physicians should copy the therapeutic model of thalassemia requires more research.

Could patients with PKD benefit from splenectomy?

Splenectomy is an effective way for eliminating or decreasing transfusion dependence. Splenectomy raises the baseline hemoglobin level and might improve the survival of transfused cells when splenomegaly is present.

British Society for Hematology has upsurged the indication of splenectomy: (1)



WJCC | https://www.wjgnet.com

| Table 2 Complete blood count of the patient before and after transplantation | | |
|--|------------------|------------------------|
| Date | CBC | |
| Before transplantation | Blood cell count | |
| | RBC | $5.7 \times 10^9 / L$ |
| | HGB | 84 g/L |
| | Bio-chemistry | |
| | ALT | 23 U/L |
| | AST | 46 U/L |
| | Total bilirubin | 28.2 μmol/L |
| 1 mo post transplantation | | |
| | WBC | $3.9 \times 10^{9}/L$ |
| | ANC | $1.23 \times 10^9 / L$ |
| | HGB | 126 g/L |
| | PLT | $109 \times 10^{9}/L$ |
| 2 mo post transplantation | | |
| | WBC | $2.7 \times 10^{9}/L$ |
| | ANC | $0.73 \times 10^{9}/L$ |
| | HGB | 118 g/L |
| | PLT | $195 \times 10^{9}/L$ |
| 6 mo post transplantation | | |
| | WBC | $4.3 \times 10^{9}/L$ |
| | ANC | $1.52 \times 10^9 / L$ |
| | HGB | 119 g/L |
| | PLT | $150 \times 10^{9}/L$ |
| 1 year post transplantation | | |
| | WBC | $5.6 \times 10^{9}/L$ |
| | ANC | $1.89 \times 10^9 / L$ |
| | HGB | 119 g/L |
| | PLT | $169 \times 10^{9}/L$ |

CBC: Complete blood count; RBC: Red blood cells; HGB: Blood hemoglobin; ALT: Alternative lengthening of telomeres; AST: Aspartate aminotransferase; WBC: White blood cell; ANC: Absolute neutrophilic count; PLT: Platelet.

> Transfusion dependent patients and those who do not tolerate anemia; and (2) Patients with massive splenomegaly at risk of spleen rupture. And the suitable time for splenectomy is in patients after 5 years old. While European Hematology Association suggested that splenectomy should be operated when the patient with severe PKD is 6 years old^[8]. According to a survey, splenectomy has led to an increase in the baseline hemoglobin by 10-30 g/L^[9]. While a retrospective international, multicenter registry study involving 144 patients showed that transfusion-dependency and moderate anemia persisted despite splenectomy in more than half of the patients, suggesting that surgery is less effective in PKD than in hereditary spherocytosis^[10].

> Apart from the effectiveness of splenectomy, complications of the surgery should also be considered. The two major issues are fateful sepsis caused by encapsulated organisms and thromboembolic disease due to increased platelets^[11].

> Our patient did not undergo splenectomy due to the following reasons: (1) He was so young that he was susceptible to infection; and (2) The level of baseline hemoglobin (57 g/L) is extremely low, indicating that he might need regular transfusion as usual post-splenectomy or undergo allo-HSCT.

Above all, splenectomy is an alternative option for moderate transfusion-dependent



WJCC | https://www.wjgnet.com

| Table 3 Similarity and difference between pyruvate kinase deficiency and thalassemia | | | |
|--|---|---|--|
| | PKD | Thalassemia | |
| Similarity | (1) Both are hemolytic anemia caused by gene abnormality; (2) Both can lead to severe hyperbilirubinemia in neonate period; and (3) Severe cases of both need transfusion regularly | | |
| Difference | | | |
| MCV | Normal | Smaller than the normal | |
| MCH | Normal | Smaller than the normal | |
| MACH | Normal | Smaller than the normal | |
| Hemoglobin electrophoresis | Normal | There are different abnormal bands according to different types | |
| Type of gene abnormality | Mutation of the <i>PKLR</i> gene which codes the enzyme of pyruvate kinase in red blood cells | Mutation of the gene which codes the globin chains | |

PKD: Pyruvate kinase deficiency; MCV: Mean corpuscular volume; MCH: Mean corpuscular hemoglobin; MCHC: Mean corpuscular hemoglobin concentration

> PKD, which might make patients get rid of transfusion post-splenectomy. But extremely low level of baseline hemoglobin might be a contraindication of splenectomy.

Would patients with PKD benefit from allo-HSCT?

Allo-HSCT is a controversial management for patients with PKD. Some researchers do not recommend allo-HSCT as a usual approach because of the unclear clinical criteria for transplantation^[2]. In contrast, a study reported 16 patients with PKD who underwent transplantation^[3]. The overall 3-year survival after transplantation in all 16 PKD patients was 65%. A significantly better survival was observed in patients who underwent transplantation before the age of 10. There was a distinction between Asian and European patients, with a higher survival rate in Asian patients due to nonsplenectomy and lower ferritin levels pre-transplantation.

Our patient received unrelated identical peripheral blood stem cells when he was 3 years old, and he did not develop acute and chronic GvHD during the 1-year followup. This might be due to his young age, regular transfusion, iron chelation, and nonsplenectomy, which were consistent with the existing information.

All in all, severe transfusion-dependent PKD might be an indication for allo-HSCT, which should be carried out as early as possible. If there are no identical siblings available, matched unrelated donors might also be an alternative option.

CONCLUSION

The patient in this case benefitted from copying the model of management of thalassemia, but whether hematologist should copy this or not requires more research.

REFERENCES

- Miwa S, Fujii H. Molecular basis of erythroenzymopathies associated with hereditary hemolytic 1 anemia: tabulation of mutant enzymes. Am J Hematol 1996; 51: 122-132 [PMID: 8579052 DOI: 10.1002/(SICI)1096-8652(199602)51:2<122::AID-AJH5>3.0.CO;2-#]
- 2 Grace RF, Zanella A, Neufeld EJ, Morton DH, Eber S, Yaish H, Glader B. Erythrocyte pyruvate kinase deficiency: 2015 status report. Am J Hematol 2015; 90: 825-830 [PMID: 26087744 DOI: 10.1002/ajh.24088]
- van Straaten S, Bierings M, Bianchi P, Akiyoshi K, Kanno H, Serra IB, Chen J, Huang X, van Beers E, Ekwattanakit S, Güngör T, Kors WA, Smiers F, Raymakers R, Yanez L, Sevilla J, van Solinge W, Segovia JC, van Wijk R. Worldwide study of hematopoietic allogeneic stem cell transplantation in pyruvate kinase deficiency. Haematologica 2018; 103: e82-e86 [PMID: 29242305 DOI: 10.3324/haematol.2017.177857]
- Pérez-Albert P, Guillen M, Prudencio M, Gonzalez-Vicent M, Sevilla J. [Haematopoietic stem cell transplantation in pyruvate kinase deficiency: When is it indicated? An Pediatr (Barc) 2018; 88: 106-107 [PMID: 28389197 DOI: 10.1016/j.anpedi.2017.02.016]



- 5 Akiyoshi K, Sekiguchi K, Okamoto T, Suenobu S, Izumi T. Cord blood transplantation in a young child with pyruvate kinase deficiency. Pediatr Int 2016; 58: 634-636 [PMID: 27460399 DOI: 10.1111/ped.12889]
- 6 Quintana-Bustamante O, Fañanas-Baquero S, Orman I, Torres R, Duchateau P, Poirot L, Gouble A, Bueren JA, Segovia JC. Gene editing of PKLR gene in human hematopoietic progenitors through 5' and 3' UTR modified TALEN mRNA. PLoS One 2019; 14: e0223775 [PMID: 31618280 DOI: 10.1371/journal.pone.0223775]
- 7 Grace RF, Rose C, Layton DM, Galactéros F, Barcellini W, Morton DH, van Beers EJ, Yaish H, Ravindranath Y, Kuo KHM, Sheth S, Kwiatkowski JL, Barbier AJ, Bodie S, Silver B, Hua L, Kung C, Hawkins P, Jouvin MH, Bowden C, Glader B. Safety and Efficacy of Mitapivat in Pyruvate Kinase Deficiency. N Engl J Med 2019; 381: 933-944 [PMID: 31483964 DOI: 10.1056/NEJMoa1902678]
- Bonnet S, Guédon A, Ribeil JA, Suarez F, Tamburini J, Gaujoux S. Indications and outcome of 8 splenectomy in hematologic disease. J Visc Surg 2017; 154: 421-429 [PMID: 28757383 DOI: 10.1016/j.jviscsurg.2017.06.011]
- Zanella A, Bianchi P, Fermo E. Pyruvate kinase deficiency. Haematologica 2007; 92: 721-723 9 [PMID: 17550841 DOI: 10.3324/haematol.11469]
- Iolascon A, Andolfo I, Barcellini W, Corcione F, Garçon L, De Franceschi L, Pignata C, Graziadei G, 10 Pospisilova D, Rees DC, de Montalembert M, Rivella S, Gambale A, Russo R, Ribeiro L, Vives-Corrons J, Martinez PA, Kattamis A, Gulbis B, Cappellini MD, Roberts I, Tamary H; Working Study Group on Red Cells and Iron of the EHA. Recommendations regarding splenectomy in hereditary hemolytic anemias. Haematologica 2017; 102: 1304-1313 [PMID: 28550188 DOI: 10.3324/haematol.2016.161166]
- Weledji EP. Benefits and risks of splenectomy. Int J Surg 2014; 12: 113-119 [PMID: 24316283 DOI: 11 10.1016/j.ijsu.2013.11.017]





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

