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A very rare patient with states for HbH disease and congenital heart disease

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Abstract

We report an HbH disease patient with interventrical septum defect. DNA analysis showed that the patient had Hb Constant Spring mutation and -&endash;SEA deletion. This case illustrates typically associated clinical features of HbH disease combined with congenital heart disease.

Text

Hemoglobin H (HbH) disease, the most complicated genetic form of a-thalassemia, is not uncommonly seen in south China and South East Asia, [1] and congenital heart disease (CHD) is one of worldwide congenital anomalies with the genetic cause of each specific lesion heterogeneous.[2] However, the clinical features of HbH disease combined with CHD are poorly discussed. Here we report such a case.

The proband, an 8-year-old boy with normal growth but slight cyanosis from a Chinese family in Xinxing county of Guangdong Province of south China, was in hospital for septum repair because he had been diagnosed as interventrical septum defect at birth. Physical examination showed the boy's heart rate was 110/min, with enlarged heart and precordial region tremor. Hematological data were obtained with a cell counter (Model Cell-DYN-3500, ADBOTP, USA) and hemoglobin electrophoresis with REP system (Helena Laboratory, Beaumont, USA). Red cell indices and hemoglobin electrophoresis data were as following: Hb 83.1g/L, HCT 0.273, MCV 72.8fl, MCH 22.1pg, MCHC 304g/L, RDW 27.5%, HbA2 3.9%,HbA 73.5%, HbF 1.7%, and HbH 20.9%. Methods of DNA analysis to determine the a globin gene defects were GAP-PCR and PCR-reverse dot blot.[3,4] Analysis result illustrated that, the genotypes of the proband, his father and mother were aCSa/--SEA , aa/--SEA and aCSa/aa, respectively. b-thalassemia mutants were typed by PCR-reverse dot blot method as described somewhere.[5] The proband and his parents were excluded 18 types of b-thalassemia seen in south China. X-ray and Color Doppler Ultrasound showed that the proband was affected interventricular septum, aortic valve lower diaphram, right ventrical outflow tract stenosis, regurgitation of pulmonary valve, aortic valve, and tricuspid valve. ECG indicated that the proband had dystrophy of two ventrical and arrhythmia. B-Ultrasound imaging displayed that moderate hepatomegaly and spenomegaly existed in the proband. Thus, the diagnosis outcome for this patient should have been the complex states for HbH disease and congenital heart disease. The defect of the interventrical septum was repaired and about 300 mL whole blood was transfused during and after operation. Stable physical condition was observed and the boy was left the hospital safely. Malformation of a-thalassemia patients, especially in homozygous a0-thalssemia ones, have been reported in a few cases, and bone marrow transplantation in b-thalassemia individuals with congenital heart disease was intensively studied.[6,7] It is unclear whether these combinations are random event or not. Heart care is important in common HbH disease and qualitative and quantitative magnetic resonance imaging is useful for screening the complication of heart failure in HbH disease because of iron overload.[1,8] In this case,

the syndrome and symptom were enhanced due to combined effect and follow-up care is necessary for this kind of patient after operation.

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