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Case Report

Interrupted aortic arch without differential cyanosis due to aberrant subclavian artery

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Abstract

Differential cyanosis indicates a lower percutaneous oxygen saturation (SpO_2) level in the lower extremities than in the upper extremities and is generally observed in patients with interrupted aortic arch (IAA). We report a case of the absence of differential cyanosis in a neonate with IAA. A male neonate was born at 38 weeks of gestation. Despite routine care, his SpO_2 level dropped to 90% at 1 h after birth. On admission, there was no evidence of differential cyanosis. Systolic murmur was noted 12 h after birth, leading to suspicion of IAA on echocardiography; IAA type B was confirmed by computed tomography. IAA type B occurs between the left carotid artery and left subclavian artery. In our patient, the levels of SpO_2 were different between the ear and lower extremities without signs of differential cyanosis because his right subclavian artery branched from the aorta distal to the interruption and left subclavian artery. In conclusion, despite the absence of differential cyanosis and lack of detailed echocardiographic evaluation, careful observation is mandatory. SpO_2 measurement in the ear is important to rule out IAA.

Key words: Interrupted aortic arch, percutaneous oxygen saturation, 22q11.2 deletion, differential cyanosis, aberrant subclavian artery

Introduction

Differential cyanosis is characterized by the appearance of cyanosis in the lower extremities with a pink right upper extremity[1], indicating a higher percutaneous oxygen saturation (SpO_2) level than the lower extremities. Interrupted aortic arch (IAA) is a disorder that disrupts the aortic arch between the ascending and descending aorta. IAA is a congenital heart disease (CHD) with differential cyanosis because the descending aorta blood flow depends on the pulmonary artery blood flow via the patent ductus arteriosus[2]. On newborn screening for critical assessment of assessment, the American Academy of Pediatrics recommends monitoring the difference in the SpO_2 levels between the right hand and either foot[3,4]. Herein, we report the case of a neonate without differential cyanosis despite the presence of IAA.

Case presentation

A male neonate (the first child) with no family history of heart disease was born at 38 weeks and 1 day of gestation via emergency cesarean delivery due to fetal distress. The baby cried right after birth and with 75% SpO_2 level at 4 min after birth, indicating peripheral cyanosis and hence requiring a continuous positive airway pressure mask. His SpO_2 level improved to 88% and 92% at 5 and 10 min after birth, respectively. Despite routine care, his SpO_2 level dropped to 90% at 1 h after birth, and he was admitted to the neonatal intensive care unit (NICU) with the following physical examination results: weight, 2,538 g (−0.86 standard deviation [SD]); height, 48.0 cm (−0.09 SD); head circumference, 33.5 cm (+0.34 SD); body temperature, 36.9°C; heart rate, 120 beats/minute; respiratory

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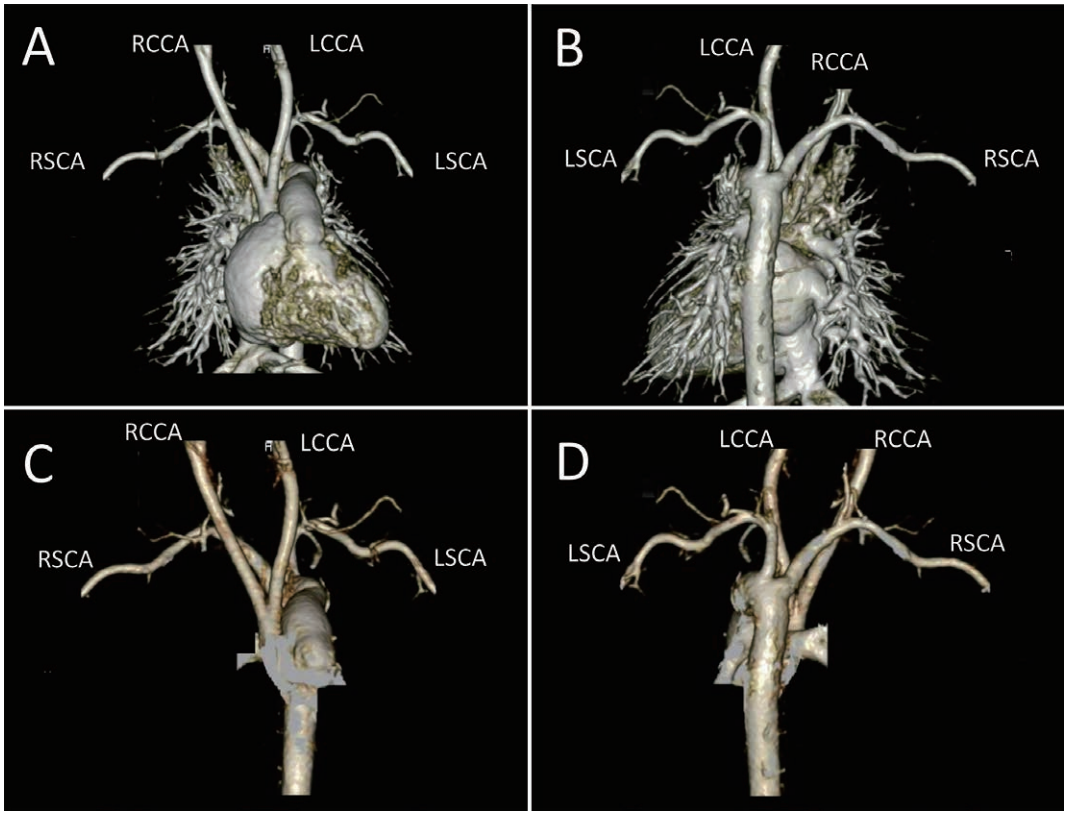

Fig. 1. A 3D computed tomography angiography reveals that the right subclavian artery branches from the descending aorta distal to the interrupted aortic arch.
(A), (C) : right view ; (B), (D) : posterior view
RCCA, right common carotid artery ; *LCCA*, left common carotid artery ; *RSCA*, right subclavian artery ; *LSCA*, left subclavian artery

rate, 58 cycles/minute ; blood pressure, 55 / 33 mmHg ; right hand SpO_2 (room air), 90%; left-foot SpO_2, 91% ; clear lungs bilaterally via auscultation ; no cardiac murmur ; no abdominal dilatation ; and no peripheral coldness but peripheral cyanosis. Our patient presented with telecanthus, flatten nose root, wide palpebral fissure, and microstomia as external abnormalities. Blood test results were within normal limits, and chest X-ray revealed no cardiac dilatation, pulmonary congestion, nor pleural effusion. Postoperative fluorescence in situ hybridization revealed 22q11.2 deletion.

Due to low SpO_2 level, oxygen was supplemented using a headbox (fraction of inspired oxygen : 0.3) for 6 h. His SpO_2 level was maintained at 95% on room air. However, the SpO_2 level gradually decreased to 92% on room air, and a grade 3 / 6 systolic murmur emerged at 12 h after birth. Meanwhile, echocardiography revealed a ventricular septal defect (VSD) and an indefinable architecture of the aortic arch. IAA was suspected ; thus, he was transferred to an affiliated cardiovascular center. The aortic annulus was small (4.2 mm, 62.5% of the normal aortic annulus), and the ascending aorta was slightly hypoplastic (4.6 mm).

On day 4 after birth, contrast-enhanced computed tomography (CT) confirmed IAA type B, in which the aorta interruption occurs between the left carotid artery and left subclavian artery, as well as a bicuspid aortic valve, a malalignment type perimembranous VSD of 8 mm in diameter, and the absence of thymus. In three-dimensional CT angiography, the right and left carotid arteries branched from the aortic arch, and the right and left subclavian arteries branched from the aorta distal to the IAA (Figure 1). The right subclavian artery was behind the esophagus and trachea, without sign of compression. His SpO_2 levels were 94%, 88%, and 88% on the right ear, right upper extremity, and right lower extremity, respectively (Figure 2), with no differential cyanosis. On day 7 after birth, aortic arch reconstruction, reimplantation of the right subclavian artery, and patch closure of the VSD were successfully performed. The patient's SpO_2 levels remained within the normal limit after surgery even without oxygen supplementation. During follow-up,

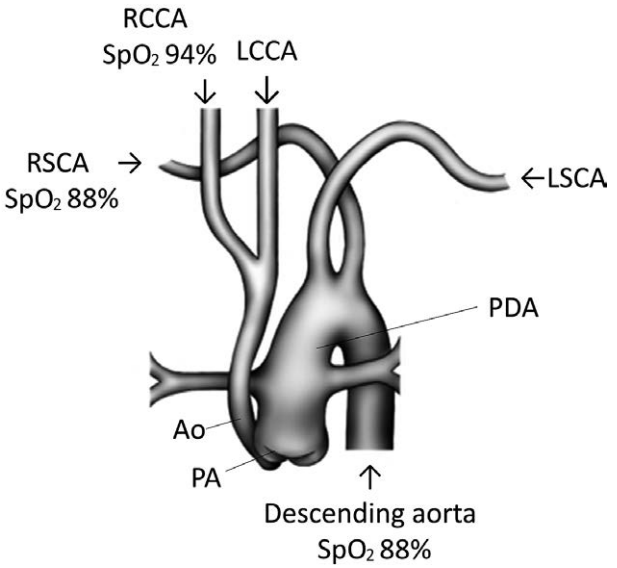

Fig. 2. Schema of interrupted aortic arch of the patient. The percentage indicates the percutaneous oxygen saturation from each vessel.
RCCA, right common carotid artery; *LCCA*, left common carotid artery; *RSCA*, right subclavian artery; *LSCA*, left subclavian artery; *Ao*, aorta; *PDA*, patent ductus arteriosus; *PA*, pulmonary artery

mild aortic stenosis was detected, and the patient was asymptomatic. Once the stenosis become significant, a mechanical valve can be placed.

Discussion

We report the case of a neonate without different cyanosis despite the presence of IAA due to the anomalous origin of the right subclavian artery.

The following two pathophysiological causes of IAA have been suggested: abnormal development of the left fourth pharyngeal arch artery or decreased blood flow to the ascending aorta. IAA has three types: type A, in which the aorta is interrupted distal to the left subclavian artery; type B, in which the aorta is interrupted between the left carotid artery and left subclavian artery; and type C, in which the aorta is interrupted between the brachiocephalic artery and left subclavian artery. IAA type B accounts for approximately 28% of the IAA cases in Japan[5,6], including the present case, and it is characterized by the blood flow in the right carotid artery is from the aorta proximal to the interruption, indicating differential cyanosis. However, differential cyanosis was not detected despite the presence of IAA type B due to a concomitant right subclavian anomaly. His right subclavian artery branched from the aorta distal to the interruption and left subclavian artery. An aberrant right subclavian artery is a congenital aortic arch anomaly in which the right subclavian artery originates from the distal interrupted aorta; this anomaly occurs in 0.5%–1.6% of the general population[7]. An aberrant right subclavian artery, usually originating as a fourth brachiocephalic branch from the upper descending thoracic aorta, is common in IAA type B but can also occur in type A[6].

The 22q11.2 deletion syndrome (i.e., Velo-cardio-facial syndrome) may manifest the pattern of malformation and abnormality, including CHD (80%), dysmorphic facial features, cleft palate, velopharyngeal incompetence, immunodeficiency secondary to thymic hypoplasia, hypoparathyroidism, and mental retardation[8]. Two-thirds of IAA cases occur in patients with 22q11.2 deletion syndrome, which manifests with IAA type B and concomitant subclavian anomaly[4,9]. VSD perimembranous malalignment type, which is observed in this case, is present in 15% of patients with 22q11.2 deletion syndrome. In our patient, Tetralogy of Fallot, though occurring in 30% of patients with 22q11.2 deletion syndrome, was ruled out because of the absence of right ventricular hypertrophy or an overriding aorta[4].

As differential cyanosis suggests CHD, measuring the SpO_2 levels in both the upper and lower extremities is recommended in Japan[3]. However, differential cyanosis was not observed in this case despite the presence of IAA type B. Because of the concomitant right subclavian anomaly, the artery supplied desaturated blood to his right hand and led to the absence of differential cyanosis. However, the levels of the right ear SpO_2 and either foot SpO_2 were different. Blood flow in the ear is supplied by the carotid artery, which branches from the aorta proximal to the aortic interruption, which also present in patients with IAA type B and concomitant anomaly, thus hiding differential cyanosis. Thus, even when detailed echocardiographic evaluation is not feasible, simultaneous SpO_2 measurements in the ear and the lower extremities is crucial for CHD screening.

The SpO_2 levels of the upper extremities can be monitored by an ear sensor, which however has a high bias rate[10]. Thus, echocardiography is the first choice for the lack of SpO_2 difference between the right hand and the lower extremities and for low SpO_2 in both extremities.

Therefore, despite the lack of SpO_2 difference between the upper and lower extremities, careful observation and detailed examination are mandatory, particularly in patients with inappropriate SpO_2 levels.

Moreover, SpO_2 measurement in the ear is important to detect complex anomalies of the aortic arch with the duct-dependent systemic circulation.

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Conflict of interest disclosure

The authors declare no conflict of interest.

Author contributions

YI : Contributed to the conception ; mainly drafted the manuscript ; critically revised the manuscript ; gave final approval and agrees to be accountable for all aspects of work ensuring integrity.

YW, MM, and TF : Contributed to the conception ; drafted the manuscript ; critically revised the manuscript ; gave final approval and agrees to be accountable for all aspects of work ensuring integrity.

HI : Supervised the conception ; drafted the manuscript ; critically revised the manuscript ; gave final approval and agrees to be accountable for all aspects of work ensuring integrity.

All authors read and approved the final manuscript.

Ethical approval

This report was approved by the Ethics Committee, Showa University School of Medicine (No. 21-058-B).

Informed consent

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

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