Case report - Congenital

Cervical aortic arch and Kommerell’s diverticulum associated with the anomalous subaortic left brachiophecalic vein in a patient with chromosome 22q11.2 deletion

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Abstract

We report the surgical case of an eight-year-old girl who had a very rare combination of ventricular septal defect and abnormal aortic arch anatomy: right cervical aortic arch, left descending aorta and Kommerell’s diverticulum from which the left subclavian artery arose with anomalous subaortic left brachiophecalic vein. She was confirmed the chromosomal 22q11.2 deletion.

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1. Introduction

Chromosomal 22q11.2 deletion is frequently associated with aortic arch anomalies. We report a case of an eight-year-old girl who had the chromosomal 22q11.2 deletion and ventricular septal defect (VSD), cervical aortic arch, Kommerell’s diverticulum and anomalous subaortic left brachiophecalic vein.

2. Case report

An eight-year-old girl was referred to us for cardiac surgery of VSD. An echocardiogram showed moderate volume-overload in the left atrium and the left ventricle with mild mitral regurgitation. Cardiac catheterization revealed pulmonary to systemic blood flow ratio of 1.6–1.7, mean pulmonary arterial pressure of 37 mmHg and pulmonary vascular resistance of 5.6 Wood unit. Aortogram also demonstrated the cervical right aortic arch (RAA) and the left descending aorta with Kommerell’s diverticulum from which the left subclavian artery (SCA) originated (Fig. 1a). In addition, an abnormal course of the left brachiophecalic vein beneath the aortic arch was also detected (Fig. 1b). She had no symptoms of compression of the respiratory tract and esophagus. The VSD was closed surgically. The postoperative course was uneventful. The aortic arch including the Kommerell’s diverticulum and the brachiophecalic vein were unchanged at the operation. A genetic test confirmed the presence of a chromosome 22q11.2 deletion.

3. Discussion

Kommerell first reported the left aortic arch (LAA) with persistence of a remnant of the right dorsal aorta which appeared as a diverticulum from which an aberrant right SCA originated [1]. Thus far, over 100 cases of Kommerell’s diverticulum have been reported. Approximately 70% of the patients with Kommerell’s diverticulum have the RAA and right descending aorta with the left aberrant SCA, the first form [2]. Over 20% of patients with Kommerell’s diverticulum have an LAA and a left descending aorta with the right aberrant SCA, the second form. The third form is a mirror image of normal branching pattern with the RAA and right descending aorta (6%). The fourth form is a normal branching pattern with the LAA (1%). A right cervical aortic arch and the left descending aorta with Kommerell’s diverticulum from which left SCA arises is extremely rare. Three papers reported similar cases with this rare combination [3–5]. Kommerell’s diverticulum sometime causes dysphagia lusoria due to esophageal compression and respiratory dysfunction or asthma due to trachea compression [6]. However, these three patients did not have symptoms related to the compression of the trachea or esophagus. Presumably because the right cervical aortic arch and the left-sided Kommerell’s diverticulum or the left ductus ligation does not align at the same level, the organs in the mediastinum may not be compressed, even though there is a vascular ring formation.

Cervical aortic arch is also a rare anomaly and it extends high into the upper mediastinum over the level of clavicles. This anomaly is considered to be due to the persistence of the second or third branchial arch with the regression of
the fourth branchial arch [7]. The aortic arch is elongated and leads to a hairpin-curved shape.

An abnormal course of subaortic left brachiocephalic vein was rare but was reported to be frequently associated with tetralogy of Fallot and RAA [8]. Furthermore, the close relationship between this anomaly and the cervical aortic arch was also reported [9]. In a normal fetus, at the eighth week of the fetal development, the ventral precardinal anastomosis which becomes the left brachiocephalic vein later, develops between both the bilateral precardinal veins. Rarely, this normally expected process does not advance due to the occurrence of an abnormal aortic arch, such as the cervical aortic arch and the RAA. Then, a dorsal precardinal anastomosis may grow into the anomalous subaortic left brachiocephalic vein.

Chromosome 22q11.2 deletion is frequently associated with congenital heart disease. It was reported that approximately two-thirds of the patients with 22q11.2 deletion had anomalies of aortic arch and aberrant branching arteries [10]. One patient of the former three cases with similar abnormal aortic arch anomalies was confirmed with the deletion of chromosomal 22q11.2 [3]. Hence, these complicated combinations of aortic arch anomalies may be related with 22q11.2 deletion.

References