Truncus Arteriosus Associated with Trisomy 18
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SUMMARY. The spectrum of cardiac anomalies in trisomy 18 typically includes septal defects and polyvalvular disease and only rarely complex malformations. We report the first case of trisomy 18 with truncus arteriosus type II.

KEY WORDS: Trisomy 18 — Truncus arteriosus

The association of trisomy 18 with congenital heart diseases is well established. Ventricular septal defects and congenital polyvalvular disease have been associated most commonly with trisomy 18 [1, 2]. There are also rare reports of complex congenital heart lesions with trisomy 18, including tetralogy of Fallot and double-outlet right ventricle [3, 4, 6]. We report a case of truncus arteriosus type II and trisomy 18 (48XXX, + 18), an association that has not been previously reported.

Case Report

The patient was a 1000 g, 32 weeks, small for gestational age (SGA) female delivered to a 31-year-old gravida 2 para 1, white married woman with good prenatal care. The infant delivered vaginally and had Apgar scores of 3 at 1 minute, 5 at 5 minutes and 7 at 10 minutes. The infant was resuscitated with bag, mask, and oxygen and was subsequently intubated and placed on mechanical ventilation. An umbilical venous catheter was placed, and sodium bicarbonate and intravenous antibiotics were administered. After transfer to Children’s Hospital—San Diego, physical examination revealed a 32-week, SGA white female infant with a heart rate of 134 per minute, respiratory rate 53 per minute, blood pressure of 58/28 mmHg and weight 980 g. The head circumference was 27.75 cm and the length 37.5 cm. The ears were small, low set, and posteriorly rotated. The palpebral fissures were small. Her sternum was short, and the labia were fused anteriorly. The hands displayed extreme thenar hypoplasia and flipper-like thumbs. The fingernails and toenails were hypoplastic. The distal creases were lacking on fingers 2, 3, and 4; and camptodactyly characterized fingers 2, 3, and 4. Clinodactyly of the fifth fingers was present. The dermal ridge pattern showed a whorl on the flipper thumbs and arches on digits 2 through 5. Cardiac examination demonstrated a right ventricular heave, no heart murmurs or clicks, and a loud single second heart sound. There were no diastolic murmurs, and pulses were palpable in all extremities. Neurologic examination revealed hypotonia and minimal response to stimulation. The patient was supported with intubation, mechanical ventilation, and intravenous fluids.

The electrocardiographic findings suggested situs solitus of viscera and atria, truncus arteriosus type II, a large subtruncal ventricular septal defect, normal position of left and right ventricles, a secundum-type atrial septal defect, and tricuspid valve redundancy and dysplasia. Admission laboratory studies showed a normal complete blood count and serum electrolytes. On room air an arterial blood gas assay revealed pH 7.39 and PO₂ 38 mmHg.

The clinical findings suggested a diagnosis of trisomy 18. Chromosome analysis confirmed trisomy 18 and XXX syndrome (48XXX, + 18) (Fig. 1). After counseling, the parents requested that the patient be removed from mechanical ventilation. Shortly thereafter, the patient succumbed to cardiorespiratory failure.

At autopsy the heart was found to be of normal size and weight, and the right atrium and ventricle were dilated. A truncus arteriosus arose primarily above the right ventricular outflow tract. There was a large subtruncal ventricular septal defect located between the two limbs of the septal band. The pulmonary artery branched from the posterior truncal surface and divided into the right and left pulmonary arteries. The truncal valve had three symmetric cusps, measured 0.6 cm in diameter, and was in fibrous continuity with the anterior mitral leaflet. A large secundum-type atrial septal defect was present, and there was no ductus arteriosus (Figs. 2 and 3). Pulmonary and hepatic congestion and retroperitoneal soft tissue edema evidenced biventricular failure. Other postmortem findings included facial hypoplasia, bilateral choanal atresia, micrognathia, proximal esophageal

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Fig. 1. Karyotype of patient showing trisomy 18 and XXX syndrome (48XXX, +18).

Fig. 2. Dorsal view shows the pulmonary artery (arrowhead) arising from the posterior truncus arteriosus (TA) and immediately dividing into the right and left pulmonary arteries.

Fig. 3. Opened right heart reveals a large subvalvular ventricular septal defect (arrow), truncus valve, and ostium (arrowhead) of the main pulmonary artery arising from the posterior wall of the truncus arteriosus.

Discussion

Although XXX syndrome has no associated cardiac abnormalities [5], extensive literature, including several recent reviews, describe congenital heart disease associated with trisomy 18 [1-4, 6]. Ventricular septal defects due to malalignment and hypoplasia of the conal septum comprise the most common cardiac anomaly. Other more complex defects, such as tetralogy of Fallot and double-outlet right ventricle (usually with mitral atresia), have been reported much less often. Lastly, congenital polyvalvular dysplasia is commonly reported with trisomy 18, often in association with other structural defects.

Our patient is the first case of truncus arteriosus associated with trisomy 18 reported in the literature. The type of truncus identified in our patient is probably due to the absence of the conal septum, subpulmonary infundibulum, pulmonary valve, and main pulmonary artery. The artery present is probably the aorta, which is suggested by the fact that the truncal valve is tricuspid and is normally connected with the mitral valve. With this type of truncus, both pulmonary arteries originate from the ascending aorta. This defect is anatomically and probably embryologically closely related to tetralogy of Fallot with pulmonary atresia. Thus given the fact that tetralogy of Fallot was found in 6 of 41 postmortem cases [6], it is not surprising that this type of truncus arteriosus would be found in a patient with trisomy 18.

References

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