

Patent Ductus Arteriosus and Pulmonary Hypertension in a Child with 49,XXXXY Syndrome

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Introduction (or Basis or Objectives): 49, XXXXY syndrome is a rare sex chromosome aneuploidy disorder. It was first reported in 1960 by Fraccaro et al. and has an approximate incidence of 1 in 85000 male births. 49, XXXXY syndrome have emphasized the "classic triad" of mental retardation, radioulnar synostosis and hypogonadism.

Case Report: A 5-month-old boy was admitted for poor feeding, heart failure and convulsion. Patient was treated by oxygen with hood, dopamine, and dobutamine for heart failure, antibiotics for suspected bronchopneumonia and phenytoin for repeated convulsions. He was born as a 19 year old mother's first child at term via caesarean section, with birth weight of 2250 gr, body length of 43 cm, head circumference 32,5 cm. He had dysmorphic facies including hypertelorism, short palpebral fissures, low nasal bridge, broad nose, dysplastic ears, cleft palate, micrognathia, short neck, micropenis, unilateral cryptorchidism. Cardiac auscultation reveals a third degree heart murmur on lower left sternal border. His thyroid function tests were normal. Cranial MRI showed both lateral dilated ventricles. His karyotype analyses was 49,XXXXY (Fig).

Echocardiography revealed a large patent ductus arteriosus (PDA). Angiography demonstrated PDA, measuring 2.5 mm at its narrowest diameter, and shunting left to right. Hemodynamic parameters were as follows: right atrium mean 11 mmHg, pulmonary artery mean pressure (MPA) 57 mmHg, aorta mean 63 mmHg. 5x 4 mm Amplatzer Ductal Occluder-I was successfully deployed through retrograde way. The MPA mean pressure decreased slightly to 40 mmHg immediately after the intervention.

Conclusions: The first case reported by Fraccaro et al. was a child with PDA. The prevalence of congenital heart disease among patients with the 49,XXXXY syndrome is 14%. PDA is the most common congenital heart defect in 49,XXXXY. Although our case was 5 month-old and had a medium PDA, severe pulmonary hypertension was established. There was not a risk factor for primary pulmonary hypertension. In fact, unless there is a very large PDA, pulmonary hypertension will not develop in a non-syndromic 5 month-infant. We think that, pulmonary hypertension develops faster in 49,XXXXY syndrome than PDA with normal population. Consequently, PDA closure should be made in early period of life for these patients.

