Congenital Heart Defects in a Patient with 10q22.3-q23.2 Microdeletion Syndrome

F.Cairello 1, C.Zanzola 2, V.Saracco 2, G.Annoni 3, C.Pace Napoleone 3, S.A.Magrassi 1

1 Pediatric and Pediatric Emergency Unit, Pediatric Cardiology Service, The Children Hospital, AO U SS Antonio e Biagio e C. Arrigo, Alessandria, Italy
2 Division of Pediatrics, Department of Health Sciences, University of Piemonte Orientale, Novara, Italy
3 Pediatric and Congenital Cardiology and Cardiac Surgery Unit. Regina Margherita Children's Hospital

Background

The 10q22.3-q23.2 microdeletion syndrome is a rare partial monosomy with autosomal inheritance, characterized by variable facial dysmorphisms, psychomotor developmental delay, behavioral abnormalities, and congenital heart defects.

Case Report

We report the case of a female patient born at 35+5 weeks' gestation via cesarean section due to premature rupture of membranes. She was referred for cardiological evaluation at two years of age following the detection of a systolic murmur. Initial echocardiography identified an ostium secundum-type atrial septal defect (ASD), mild pulmonary stenosis with a dysplastic pulmonary valve, and pulmonary artery branch anomalies with slight flow acceleration in the left branch. Physical examination revealed dysmorphic features including low-set ears, hypertelorism, and mild micrognathia. Given the combination of congenital cardiac anomalies and facial dysmorphisms, genetic testing was performed, confirming a 7.2 Mb deletion at chromosome 10q22.3-q23.2. This genetic abnormality was associated with neuromotor developmental delay and multiple congenital malformations, consistent with a microdeletion syndrome. During follow-up, the patient developed progressive mitral regurgitation, which, combined with the large ASD, resulted in worsening right-sided cardiac overload.

Although clinically asymptomatic, recent evaluation revealed increased intensity of the cardiac murmur and development of a precordial bulge. Echocardiography demonstrated significant dilation of the inferior vena cava and hepatic veins, along with marked right heart enlargement secondary to exacerbated mitral regurgitation and shunting across the ASD. Due to the complex hemodynamic status, the case was reviewed at a tertiary center. Preoperative transesophageal echocardiography confirmed severe mitral regurgitation caused by a cleft in the anterior mitral leaflet. Balloon occlusion of the ASD induced a rise in pulmonary wedge pressure from 12 to 20 mmHg, indicating the need for surgical intervention. The patient underwent mitral valve repair and ASD closure under cardiopulmonary bypass. Postoperatively, mild residual mitral regurgitation persisted to avoid iatrogenic mitral stenosis. Recovery was uneventful, and follow-up echocardiography confirmed closure of the ASD without residual shunting, mild right atrial enlargement, and stable mild-to-moderate mitral regurgitation without stenosis

Conclusions. The 10q22.3-q23.2 microdeletion syndrome is a rare genetic disorder with variable clinical presentations, including congenital heart defects. While atrial septal defects and valvular anomalies occur in ~35% of cases, the combination of a large ASD and cleft mitral valve with severe regurgitation is uncommon. This case, consistent with prior reports, suggests a link between deleted genes and cardiac development. Genetic testing is essential in evaluating congenital cardiac anomalies. Early diagnosis and regular follow-up enable timely management of cardiac dysfunction, emphasizing the need for multidisciplinary care to improve long-term outcomes.

AOU AL