A sixteen year old girl presented with history of hemoptysis of one week duration. She had history of dyspnea on exertion and frequent respiratory infections in childhood. Her father and two siblings had history of neurofibroma. On examination she was short stunted with dysmorphic features like low set ears, broad nasal root and ocular hypertelorism. She had multiple neurofibromas, café au lait spots and multiple lentigines. There was differential clubbing and cyanosis of toes and polycythemia (Figure 1). Cardiovascular system examination revealed heaving apical impulse, mild cardiomegaly, left parasternal heave, pulmonary artery pulsations and a loud and palpable second heart sound. A clinical diagnosis of patent ductus arteriosus (PDA) with severe pulmonary artery hypertension (PAH) and hypertrophy of both ventricles with no significant LV or RV obstruction (Figure 3). In addition there was a large patent ductus arteriosus with right to left shunting and severe PAH. So a final cardiac diagnosis of PDA Eisenmenger syndrome with biventricular hypertrophic cardiomyopathy was made. She also had lentigines, ocular hypertelorism, ECG abnormalities and growth retardation satisfying the criteria for LEOPARD syndrome.

The acronym LEOPARD was coined by Gorlin et al in 1971 as a mnemonic of the major features of this disorder viz. multiple lentigines, ECG abnormalities, ocular hypertelorism, pulmonic stenosis, abnormal genitalia, retardation of growth, and sensorineural deafness. Voron et al have suggested criteria for diagnosis of LEOPARD syndrome. Clinical diagnosis may be suspected in the presence of multiple lentigines and two cardinal features. In the absence of lentigines, three features in the patient and the presence of an affected close relative are diagnostic. Limongelli et al reviewed cardiac manifestations in 26 patients with LEOPARD syndrome. ECG abnormalities occurred in 73% of the patients. Left ventricular hypertrophy occurred in 73% and right ventricular hypertrophy in 30%. Previous reports suggested that pulmonary valve stenosis is the most common defect (40%). According to recent reports pulmonary stenosis occurs in only 10-20% of patients. Currently hypertrophic cardiomyopathy (HCM) is described as the most frequent anomaly (in up to 80%). Mutations in PTPN11 gene were detected in 88% of patients with multiple lentigines LEOPARD syndrome and most of them have hypertrophic cardiomyopathy. This case report describes a very rare occurrence of PDA Eisenmenger syndrome along with biventricular hypertrophic cardiomyopathy in a patient with LEOPARD syndrome. This patient has coexistence of two major structural cardiac anomalies with most of the morphological abnormalities of LEOPARD syndrome.
Fig. 1: (a) hypertelorism, multiple lentigines, (b) café au lait spots, (c) differential clubbing and cyanosis of toes

Fig. 2: Chest X-ray PA view shows mild cardiomegaly with dilated main pulmonary artery and peripheral pruning

Fig. 3: 2-D echo showing (a) asymmetrical septal hypertrophy (b and c) biventricular hypertrophy and (d) patent ductus arteriosus

References


