Left ventricular non compaction and septal defects

Non compaction du ventricule gauche et défects septaux

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RÉSUMÉ

Nous rapportons le cas d'une patiente âgée de 23 ans présentant une association, non décrite à ce jour (au meilleur de notre connaissance), d'une non compaction du ventricule gauche à des défects septaux aux 2 étages atrial et ventriculaire. L'enquête génétique familiale a conclu à une mutation probablement sporadique du gène E101K.

Mots clés : Cardiopathie congénitale, communication inter-auriculaire, communication inter-ventriculaire, non compaction du ventricule gauche, maladie génétique

SUMMARY

We report the case of a 23-year-old woman with a not yet described (to the best of our knowledge) association of left ventricle non-compaction with both atrial and ventricular defects. Family genetic survey concluded to, a probably sporadic, E101K gene mutation.

Keywords : Congenital heart disease, atrial septal defect, ventricular septal defect, left ventricular non compaction, genetic disease

AIM

Left ventricular non-compaction (LVNC) appears as one phenotype of multiple genotypes and mechanisms. Among the LVNC wide range presentations, we report a scarcely described genetic form associated to septal defects.

CASE

Our patient was a 23-year-old female. She was referred to our cardiology department following surgical repair of a large ostium secundum atrial septal defect (ASD) performed by the mean of bovine pericardial patch. Before surgery, she had complained from dyspnea and cough. Echocardiography had shown a large ASD with an important left to right shunt. Surgery was mandated by insufficient posterior rim (3 mm). There were no surgical complications and the post-operative immediate outcome was favorable.

The patient was asymptomatic. On examination, blood pressure was 110/60 mmHg, heart rate was 68 bpm. We found however a holosystolic cardiac murmur, maximal in the inferior parasternal border. There were no signs of heart failure and no cyanosis nor clubbing.

Electrocardiogram showed a sinus rhythm, a normal PR internal (120 ms) and a right bundle branch block.

Transthoracic echocardiography did not reveal any residual interatrial shunt but showed an apical LVNC (Figure 1A, 1B) and a restrictive 4 mm ventricular septal defect (VSD) (Figure 1C,1D,1E). Left ventricular global systolic and diastolic functions were normal (left ventricular ejection fraction 59%, global longitudinal strain -19%). Magnetic resonance imaging confirmed LV apical non compaction as well as a mid-septal VSD (3-4 mm) (Figure 2). A 24h-ECG Holter monitoring did not show any supraventricular nor ventricular rhythm disorder. A Family screening was scheduled, the 4 relatives (brother, mother, father, and aunt) that were screened by echocardiography did not have any cardiac abnormality. Genetic testing performed only in the index patient identified E101K mutation in the alpha-cardiac actin gene.

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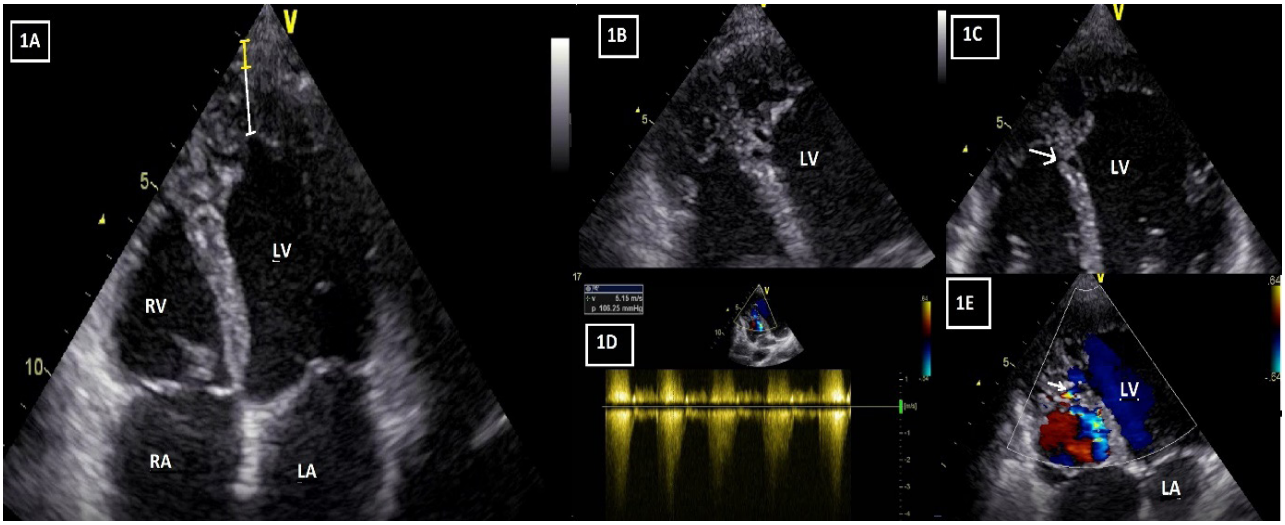

LA: Left atrium, LV: Left ventricle, RA: Right atrium, RV: Right ventricle

Figure 1. Transthoracic echocardiography 1A: Bidimensional apical 4 chamber view showing the apical left ventricular trabeculations. The white line indicates the thickness of the trabeculated non-compact myocardium and the yellow line the thickness of compact myocardium. 1B: Apical 4 chamber view zoomed on the apical region showing apical left ventricular trabeculations. 1C: Apical 4 chamber view focused on the apical region showing the ventricular septal defect. 1D: Continuous wave Doppler showing the left to right ventricular systolic flow with an inter-ventricular gradient of 101 mmHg. 1E: Color Doppler 4 chamber view showing the left to right ventricular shunt (arrow).

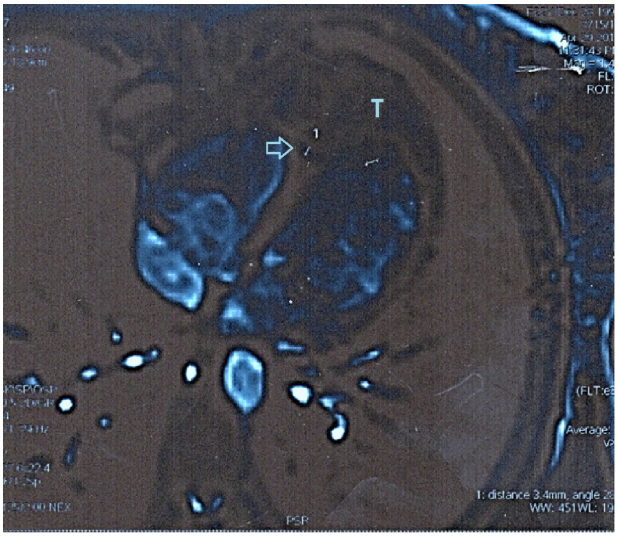

Figure 2. Cardiac magnetic resonance (T2) 4 chamber view showing the trabeculated apical region of the left ventricle (T) and the ventricular septal defect (arrow)

COMMENTARIES

LVNC encompasses a wide range of mechanisms and manifestations. It was classified as a “non-classified” or a “genetic” cardiomyopathy (1). It can be a congenital or acquired either transient of permanent condition. It can be isolated or combined to hypertrophic, dilated, restrictive, or right ventricular arrhythmogenic cardiomyopathies features (1). It was associated to sporadic or familial congenital heart abnormalities like Ebstein disease, patent ductus arteriosus and septal defects. The E101K mutation of the sarcomeric protein alpha cardiac actin gene, was the first mutation associated to both dilated cardiomyopathy and apical hypertrophic cardiomyopathy. It was then also associated to LVNC (2). When it is familial, it has an autosomal dominant transmission. Septal defects either atrial septal defects or ventricular septal defects were frequently associated to E101K mutation, but to the best of our knowledge, there was no reported case with both atrial and ventricular septal defects. The three serious and potentially fatal complications of LVNC are

thromboembolic events, ventricular arrhythmias and heart failure. The mutation E101K was first considered as benign but some cases of sudden death and severe heart failure were reported. In our patient we did not find any LV systolic or diastolic dysfunction nor rhythm abnormalities. Even if we did not screen all family members, the 4 screened relatives did not have any cardiac abnormality. A sporadic mutation was not excluded.

In conclusion: LVNC can be regarded as an anatomical description of a various genetic or acquired diseases rather than a defined cardiomyopathy disease. In patients who present for any common or complex congenital heart disease, it is very important to lead a comprehensive morphological and functional cardiovascular examination. Associated conditions like LVNC can modify the patient prognosis and management.

REFERENCES

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