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Image Article

Clinical Case of Rare Acute and Chronic Cardio-Pulmonary Involvement in Neurofibromatosis Type-1 (Von Recklinghausen's Disease)

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Introduction

Von Recklinghausen's disease (Neurofibromatosis Type 1) represents a complex clinical syndrome. Not every feature is completely disclosured. Few reports in the last decade showed growing cardiac involvement, but not totally understood. Cardiac abnormalities have potential long-term hemodynamic consequences, for these reasons close cardiological assessment are needed for these patients. Anecdotic cases for pulmonary involvement are described, but there may be an unrecognized association between von Recklinghausen disease and pulmonary hypertension above all about the way of clinical presentation. In the comprehensive clinical classification of pulmonary hypertension this systemic disorder was put in group 5 (pulmonary hypertension) with unclear and/or multifactorial mechanisms [1]. In this case we describe a patient with pulmonary hypertension, diagnosed in the clinical setting of neurofibromatosis type 1, presented with severe pericardial effusion. The contemporary presence of NF1, pulmonary hypertension and pericardial effusion in the same clinical setting gives the possibility to discuss about potential physio pathological mechanisms not deeply clear.

Clinical Presentation

A 58-year-old woman with von Recklinghausen's disease (Neurofibromatosis Type 1) was referred to our Intensive Care Unit for progressive dyspnea at rest, diaphoresis and hypotension. Objective examination showed reduced intensity of cardiac tones, with diffuse reduction of pulmonary murmurs without signs of pulmonary edema. ECG findings revealed sinus tachycardia, 115 bpm, with low voltage of QRS (Figure 1).

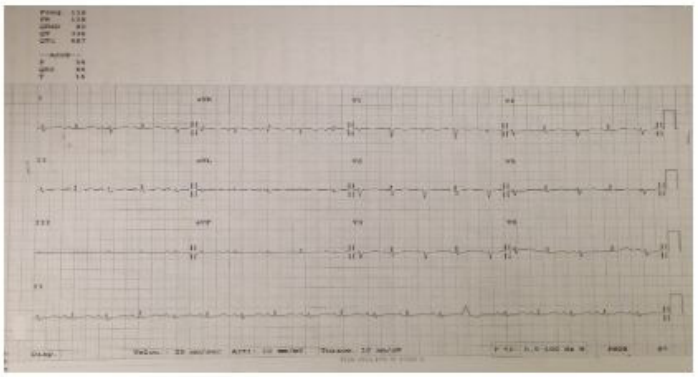

Figure 1: ECG at the moment of hospitalization.

Echocardiography showed cardiac tamponade needed to emergency pericardiocentesis (Figure 2).

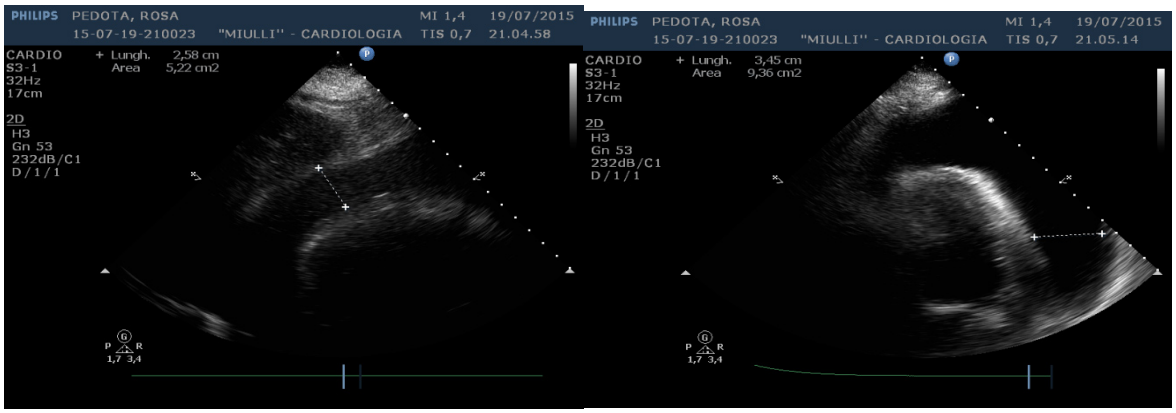

Figure 2: Echocardiogram: severe pericardial effusion ("Swimming Heart").

After critical phase management, echocardiographic features presented right ventricular dilatation and reduced right ventricular function. Acute decompensated right heart failure, probably above an unrecognized chronic dysfunction, was treated by optimizing fluid balance, right ventricular and circulatory support through inotropes and vasopressors agents such as dobutamine (till 5γ/kg/min) and noradrenaline (0.2-1.0μg/kg/min). Stabilized clinical setting invasive hemodynamic evaluation was consistent with pre-capillary pulmonary hypertension with preserved cardiac output (MPAP 59 mmHg, PVR 8.81 WU) (Table 1). Secondary potential causes of pulmonary hypertension were discussed in accordance with the updated diagnostic approach for the definition and diagnosis of pulmonary hypertension. Even if actually off label therapy was started to give a chance to the patient (Sequential drug combination therapy: tadalafil 40 mgr. die and after a month macitentan 10 mgr. die per so, titrated in the clinical follow-up over following 2 months). The patient was followed during successive months through clinical evaluation and after three months of therapy (Tadalafil+Macitentan) she was undergoing to another cardiac catheterism resulting in a significant improvement of hemodynamic parameters (Table 1). Cath-lab findings showed persistent high cardiac output and during the follow up assessment differential causes were ruled out: value of hemoglobin were stable in range (to exclude a chronic anemia); clinical and radiological examinations did not reveal any neuroendocrine tumor; no arterial-venous shunts were found. At 1-year follow-up the patient presented clinically stable, without pericardial effusion at echocardiographic check; and with stable hemodynamic parameters. (Table 1)

		Basal	3 Month Control	12 Month Control
WHO-FC		III-IV	III	II-III
Echocardiography	**TVR (m/sec)**	4,3	2,9	3,2
	TAPSE (mm)	12	16	16
	Pericardial Effusion (Post Pericardiocenteis)	Mild	Mild	None
Chateterization	**MPAP (mmHg)**	59	44	32
	RAP (mmHg)	13	4	1
	PAWP (mmHg)	12	15	4
	CO (l/min)	5.33	8.6	8,1
	CI (l/min/m²)	3.15	4.91	4,96
	PVR	8.81 WU 704.82 dyne×sec/cm^5	3.3 WU 264 dyne×sec/cm^5	3.4 WU 272 dyne×sec/cm^5
	PVRI	14.91 WU×m²; 1.192.9dyne×sec*m^2/cm^5	5.8 WU×m^2; 464dyne×sec*m^2/cm^5	5.6 WU×m^2; 448dyne×sec*m^2/cm^5

WHO-FC: World Health Organization Functional Class; **TVR**: Peak Tricuspid Regurgitation Velocity; **TAPSE**: Tricuspid Annular Plane Systolic Excursion; **MPAP**: Mean Pulmonary Artery Pressure; **RAP**: Right Atrial Pressure; **PAWP**: Pulmonary Capillary Wedge Pressure; **CO**: Cardiac Output; **CI**: Cardiac Index; **PVR**: Pulmonary Vascular Resistance; **PVRI**: Pulmonary Vascular Resistance Index

Table 1: Stabilized clinical setting invasive hemodynamic evaluation was consistent with pre-capillary pulmonary hypertension with preserved cardiac

output (mPAP 59 mmHg, PVR 8.81 WU).

Discussion

Von Recklinghausen's disease is an autosomal dominant disease due to mutations in the tumor suppressor gene NF1 located on chromosome 17, with an incidence of 1 in 3500 individuals [2]. Some reports in the literature have shown cardiac involvement in NF1, including valvar pulmonary stenosis, branch peripheral pulmonary stenosis, atrial and ventricular septal defects, coarctation of the aorta (Thoracic and Abdominal), and hypertrophic cardiomyopathy. Recent study analyzed 65 young patients with NF1 finding 11 cases of cardiac involvement, above all valvar defect (mitral regurgitation, aortic and tricuspid regurgitation in one case) atrial and ventricular septal defects, pulmonary valvar stenosis [3]. Pulmonary hypertension is rarely reported, above all with acute onset and cardiac tamponade in right ventricular failure. Mechanisms involved in the pathophysiology are various, and not deeply detected. Regard recent comprehensive clinical classification of pulmonary hypertension, NF1 was included in V group pulmonary hypertension with unclear and/or multifactorial mechanisms [1]. Pulmonary hypertension may be due to the plexiform lesions in pulmonary arterioles, and in this case, it is possible to consider also the aspect of group 1, above all regarding the genetic involvement; in fact, in the same family, different brothers or sisters could present different cardio-pulmonary pathological features, such as various polymorphism impact in disease phenotypic. Another physio pathological pathway may be parenchymal lung lesions that reduces the vascular bed, typical features of group 3. The complex mechanism is object of further studies and the cardio-pulmonary involvement could appear suddenly, so through this case of idiopathic precapillary pulmonary hypertension in a patient with Von Recklinghausen's disease we report the recommendations regarding routine surveillance for cardiovascular disease in these patients. Early screening for pulmonary arterial hypertension in combination with modern pulmonary arterial hypertension-directed therapies may improve outcomes.

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