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# Complete Atrioventricular Block Revealing Von Recklinghausen Neurofibromatosis

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**Abstract** **Case Report**

This is the case of a woman aged 55 years with no particular antecedent who presented with dyspnea of progressive onset occurring with effort associated with a sensation of lipothymia without other signs, such as fever, angina pectoris, and palpitations. The initial clinical examination found bradycardia with an irregular rhythm. Twelve-lead electrocardiogram documented bradycardia with atrial fibrillation equivalent to a high grade atrio-ventricular block. Transthoracic echocardiogram reveals bi-auricular dilatation without hypertrophy of ventricular cavities with preserved systolic function without Segregation disorder neither global nor presence of a significant valvulopathy. The patient benefitted from the installation of a pacemaker with significant improvement of the symptoms.

**Keyword:** Von Recklinghause desease, high grade atrio-ventricular block, Pacemaker, thyroid hormon.



## INTRODUCTION

NF1 represents 95% of all neurofibromatosis (NF). Incidence is approximately 1/3,500 and prevalence of approximately 1/4,000. Endocrine complications are common particularly dysthyroidism and pheochromocytoma [1]. They have a higher cardiac incidence. However, there is little data regarding incidence of arrhythmias in this population.

## CASE REPORT

A 55 years old woman with cardiovascular risk factors: Menopause, Sedentarity, without Diabetes or Hypertension, had 1 week before her hospitalization a notion of emotional shock secondary to the death of her 21-year-old girl, following an undocumented pathology.

The beginning of the symptomatology dates back to 72hrs before hospitalization by the sudden installation of an episode of lipothymia (notion of falling with partial, progressive loss of consciousness) without other associated signs.

On admission, the patient was very tired, no chest pain, no dyspnea, palpitations, or other signs of appeal. The clinical examination found a patient who was conscious and stable, bradycardia at 39 bpm, TA: 134/74 mmHg, heart sounds B1 and B2 perceived without added noise.

The examination of the thyroid gland revealed a goiter of about 6 cm of diameter, polylobulated (fortuitous discovery). The dermatological examination found pale brown pigmented spots "café au lait" rounded about 1.8 cm in diameter without relief located on the dorsal region of the trunk (*fig 1,2*) . This lesion description was in favour of Von Recklinghausen's disease.

An electrocardiogram performed at the patient's bed showed an irregular rhythm, normal heart axis, and atrial fibrillation-like bradycardia equivalent to atrioventricular block III, fine QRS with giant T-wave inversion (*fig 3*).

Cardiac Doppler ultrasound showed non dilated left ventricle with cardiac output estimated at 5 l / min, LVEF (left ventricular ejection fraction) conserved at 57%. She also noted biatrial dilatation (SOG = 24 cm2 and SOD = 24 cm2), left filling pressures (E / E '= 17), good systolic function of the right ventricle with pulmonary arterial hypertension (PAPS estimated at 53 mm Hg.

The biological analysis showed a normal blood ionogram, CRP = 10 mg / l, Troponin I = 344 → 380 negative troponin kinetics (variation <10%), creatinine in Umol / L (value??). The dosage of thyroid hormones showed a TSHus (0.001 <0.05 mIU / L) and a free T4 = 25 ng / L.,

On cervical ultrasound: There was multi nodular goiter; the scintigraphy has not been realized. The diagnosis of NF complicated by dysthyroidism and complete atrioventricular block was retained.

As initial treatment le patient received DIMAZOL (neomercazole)+ SINTROM (acenocoumarol) with installation of euthyroidism and persistence of AVB and permanent conductive disorders on HOLTER EKG.

A single chamber pacemaker was inserted (*fig 4*) and her symptoms improved significantly with AVB regression (*fig 5*).

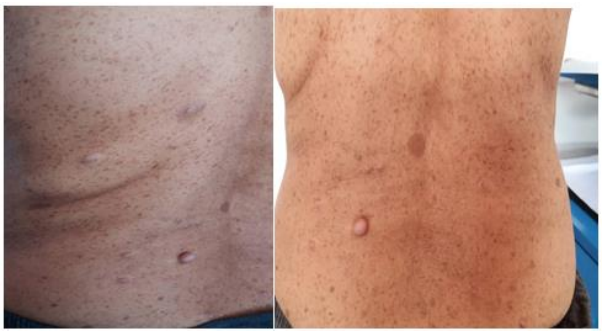

**Fig-1 ET 2: Pale brown pigmented spots "café au lait"**

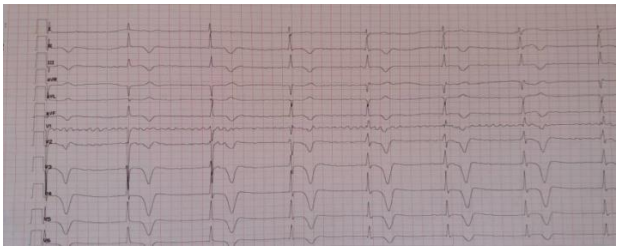

**Fig-3: ECG show complete atrioventricular block with giant T wave inversion**

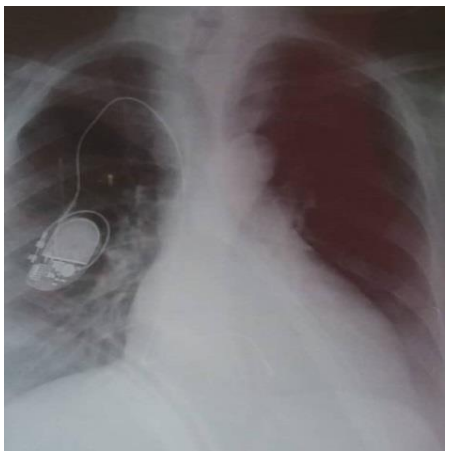

**Fig-4: x-ray show the pacemaker**

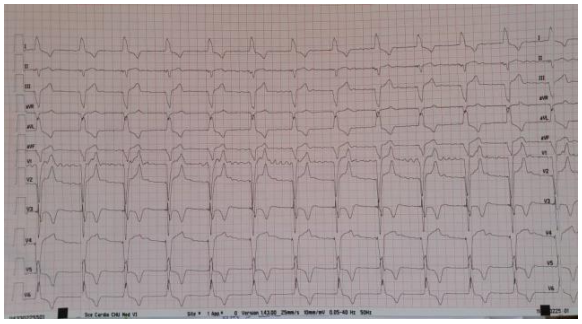

**Fig-5: ECG after pacemaker**

## DISCUSSION

Neurofibromatosis is a genetic neurocutaneous disorder characterized by the development of benign tumors of peripheral nerve cells called neurofibromas and skin lesions, i.e. café au lait spots [1]. NF1 is caused by a NF1 gene mutation on chromosome 17q11. 2[2]

There are three major clinically and genetically distinct forms of neurofibromatosis. Neurofibromatosis type 1 (NF1) or von Recklinghausen disease is the most common type of the neurofibromatosis [3]. NF1 is genetically transmitted according to an autosomal dominant pattern with an incidence of 1:3000 individuals [4].

At the time of this analysis, despite a higher incidence of anatomic cardiac abnormalities, there is little data regarding incidence of arrhythmias in this population.

Patients with NF1 are at increased risk for a variety of cardiovascular disorders, but the natural history and pathogenesis of these abnormalities are poorly understood [5].

There is very little published data on conduction system disorders in neurofibromatosis. There is an isolated report of bundle branch block in association with neurofibromatosis. It is possible that excess parasympathetic stimulation may contribute to genesis of arrhythmia in these patients [6].

Neurofibromas may be evident all over the body including the heart, thus, may contribute to the evolution of arrhythmias in NF1 patients.

This is in our opinion the third case of complete AVB on NF1 reported in the literature. A 73-year-old woman followed for NF1 and an isolated case of NF1 in a 5-year-old child who developed sinus bradycardia with second-degree atrioventricular block advanced in adolescence to an AVB [7].

Diagnosisof NF1is based on the presence of at least 2 of 7 clinical criteria described by United States National Institutes of Health (NIH) Consensus

Conference in 1987 with an update in 1997 which had been commonly utilized for standard clinical care. The distinctive features of NF1 are multiple "cafe au lait" macules, axillary and inguinal freckling, neurofibromas, and iris hamartomas [8, 9].

Present case diagnosed NF1 with two clinical criteria (cafe au-lait spots on skin and axillary freckling). In NF1, Endocrine complications are common particularly dysthyroidism and pheochromocytoma [4]. The prevalence of HT in the general population is 2% and it is more common in females. Screening for autoimmune thyroid disease and thyroid function is unnecessary. A thyroid gland examination is of the greatest value as the first step in an investigation, and thyroid hormone levels and ultrasonography should be performed as a second step in cases of suspicious findings [2].

As for many rare diseases, research has not yet found a miraculous cure, but symptomatic treatment is most often proposed. The complete resolution of the symptoms occurred after the definitive implantation of a pacemaker in all patients reported.

For our patient we did not measure the anti-TSH receptor antibody, however, in view of exophthalmia, clinically palpable goitre, biological hyperthyroidism and confirmed thyroid disorder by cervical echography, we initiated the antithyroid treatment, while suggesting the diagnosis of Graves' disease rather than hashitoxicosis. It is not clear that the relationship between NF1 and Graves' disease is a coincidence or association. On the other hand, it is known that the NF1 gene encoding neurofibromin is a tumor suppressor gene and that the genetic mutation of this gene is at the origin of several types of cancer. Hyperthyoridism has been reported in patients with McCune Albright Syndrome (MAS) [10].

## CONCLUSION

The atrioventricular blocks during hyperthyroidism are rare; their association during the NF1 is also rare, but well described in the literature. Implantation of a pacemaker remains imperative in the management with the disappearance of the AVB.

**Conflict of interests**

There are no conflicts of interests for the development of this publication.

**Ethical standards**

Informed consent was obtained from the patient's parents for the publication of this case.

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