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RESEARCH ARTICLE

BIFASCICULAR BLOCK REVEALING STEINERT'S MYOTONIC DYSTROPHY

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Abstract

Steinert's disease or myotonic dystrophy type 1 is a genetic neuromuscular disorder with autosomal dominant transmission. It leads to multisystemic damage, including a cardiac localization that is life-threatening.We report the case of a 55 year old patient, without cardiovascular risk factors, with a history of distal muscle damage since the age of 25 years, in his family history we find a brother followed for an unlabelled skeletal muscle damage.He consulted for exertional dyspnea and lipothymic discomfort. The clinical examination revealed a decrease in muscle strength in the lower limbs with amyotrophy. Walking is difficult with the help of crutches and the patient uses a wheelchair. The neurological investigation concluded in Steinert's myotonic dystrophy on the electromyogram.The patient's ECG showed a bifascicular block consisting of a left anterior hemiblock and a complete right bundle branch block. The echocardiography did not show any structural abnormalities but found a disturbance of the LV relaxation on the mitral profile.The electrophysiological exploration carried out for the measurement of infrahisical conduction revealed a long HV at 100ms indicating the implantation of a double chamber stimulation.The interest of reporting this case was to underline the importance of screening for cardiac damage of rhythmic origin. But also to inform with the help of literature data the incidence of these forms and the place of cardiac stimulation in the prevention of sudden death in steinert's myotonia.

Introduction:-

Steinert's myotonia or myotonic dystrophy type 1 is an autosomal dominant muscle disease. It leads to multisystemic involvement including ocular, respiratory, gastrointestinal and endocrine [1].

This disease is due to the expansion of the Cytosine-Thymine and Guanine triplet in the gene coding for a protein called "Dystrophia myotonica protein kinase" located on the long arm of chromosome 19q13, 3 [1,2].

A cardiac and especially a rhythmic investigation is therefore necessary in order to detect the existence of conductive or rhythmic disorders that could affect the prognosis of these patients [3].

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The description of our case was to orientate the diagnosis of steinert's myotonia in relatively young subjects in whom a conductive disorder is discovered and to emphasize the importance of screening for the disorder whatever the degree of neuromuscular alteration.

Patient and Observation:-

This is a 55 year old patient with no modifiable cardiovascular risk factors, having as family history a brother followed for an unlabelled distal muscle deficit. He presented with a progressive decrease in muscle strength in the lower limbs since the age of 25 years but had never been investigated.

He was referred to a rhythmology unit to investigate lipothymic malaise associated with NYHA stage 2 dyspnea which had been evolving for three months.

Results:-

Neurological examination revealed difficulty in walking with loss of tone and amyotrophy of the lower limb muscles. The patient used a wheelchair. Muscle strength was assessed at 4/5 and low ROTs in the lower limbs.

The cardiovascular examination was unremarkable, with no orthostatic hypotension or congestive signs.

The EKG showed a regular sinus rhythm with a HR of 75cpm, a bifascicular block consisting of a complete right bundle branch block and a left anterior hemiblock, no repolarisation disorders or cavitary hypertrophies.

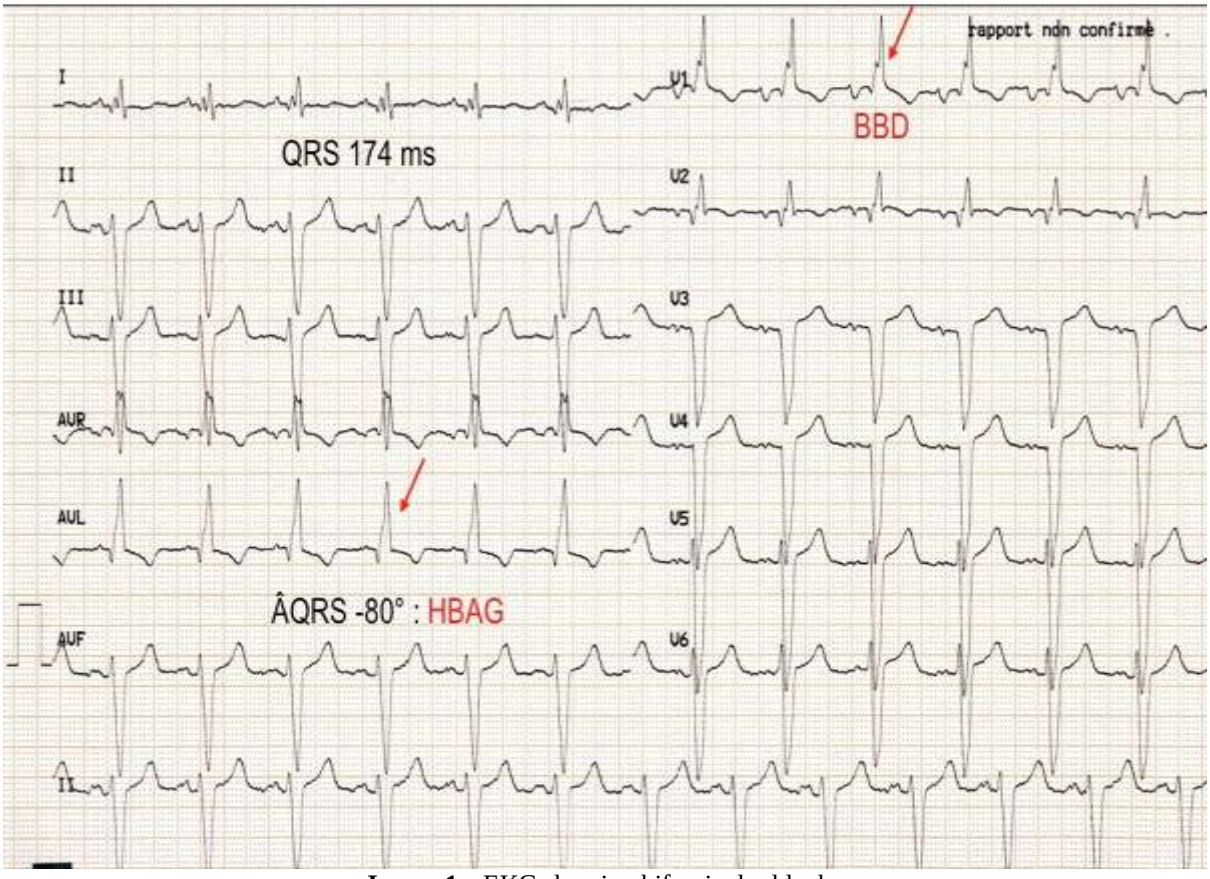

Image 1:- EKG showing bifascicular block.

The EKG holter showed the presence of numerous supraventricular extrasystoles without sustained arrhythmias. Echocardiography showed non-dilated heart chambers, preserved biventricular systolic function and a mitral profile of relaxation disorder (image 2).

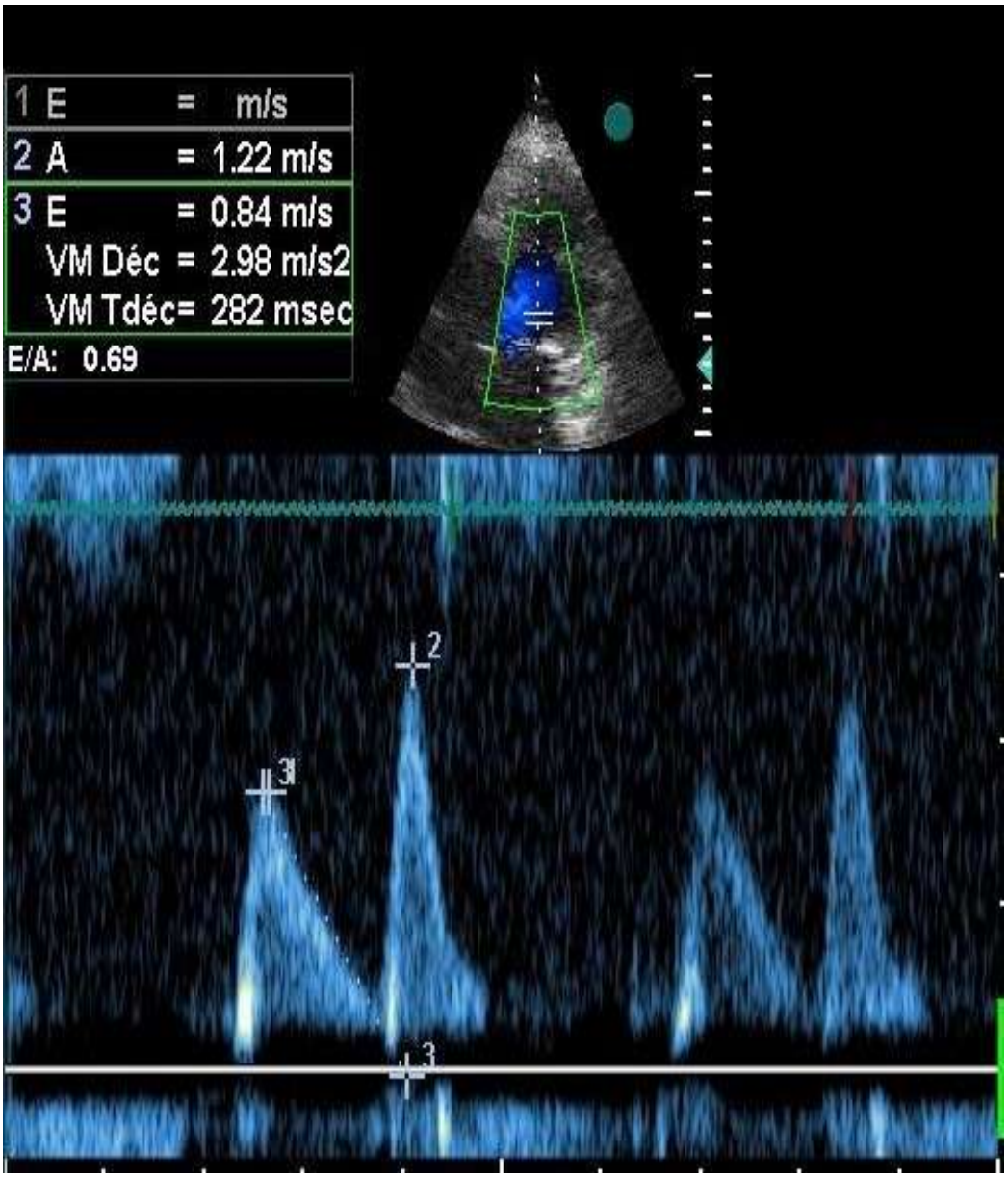

Image 2:- Left ventricular relaxation disorder in mitral profile.

The patient was referred to neurology to investigate the etiology of the muscle deficit. The electromyogram showed a myogenic pattern with predominantly distal involvement. The MDRS neurological score was evaluated at 4-5. The clinical and paraclinical neurological examination, supported by the patient's family history, was strongly suggestive of Steinert's myotonic dystrophy. The genetic study is still in progress.

The electrophysiological exploration indicated for the measurement of conduction below the his showed a prolonged interval at 100ms.

A double chamber pacemaker was implanted without incident (image 3).

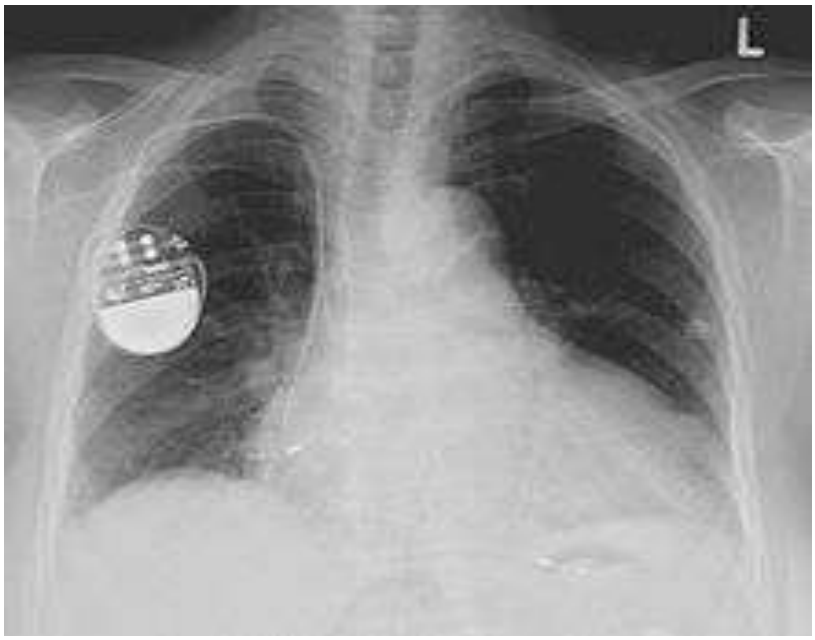

Image 3:- Chest X-ray after implantation of a dual chamber pacemaker.

He was then referred to neurology for further follow-up. The neurological condition did not worsen after 1 year.

Discussion:-

Myotonic dystrophy type 1 (DM1) or Steinert's disease is an autosomal dominant multisystem disorder affecting 1/8000 individuals [4].

Cardiac involvement is the most serious complication and can sometimes be asymptomatic, resulting in sudden death secondary due to a conductive or rhythmic disorder [5,6].

In order to detect these cardiac forms at an early stage, it is essential to carry out a cardiac investigation in patients with this genetic disease.

Conductive disorders are the main cardiac disorder found in Steinert's myotonia [7].

First-degree AVB appears to be the most frequent conductive abnormality found on a resting EKG in these patients. It is followed by intraventricular conductive disorders and then by bifascicular blocks (table 1).

Studies ECG abnormalities		Hawley et al. (1999)	Lazarus et al. (1999)	Babuty et al. (1999)	Antonini et al. (2000)	Finsterer et al. (2001)	S.Chebel et al. (2005)
1st degree AVB		51%	49%	41%	58%	24%	64%
Bundle block branch		27%	31%	41%	51%	19%	32%
Bifascicular block		25%	-	37%	37%	-	26%
Repolarisation disorder		5,4%	7,2%	7%	21%	14%	3%
Arrhythmia		-	-	-	-	10%	3%

Table 1:- ECG data from different series in the literature.

Rhythm disturbances on resting EKG or Holter recordings are uncommon and predominantly atrial and ventricular extrasystoles [8].

Our patient presented with lipothymic discomfort and exertional dyspnea with bifascicular block on EKG.

Cardiac functional symptomatology as the main sign of consultation has been found in several series on DM1 in 16 to 30% of cases. Asymptomatic forms are therefore more prevalent and should attract the attention of the clinical practitioner [9, 10].

The duration and severity of the dystrophic condition was significantly correlated in S. Chebel's study with the frequency of the cardiac disease (figure 1).

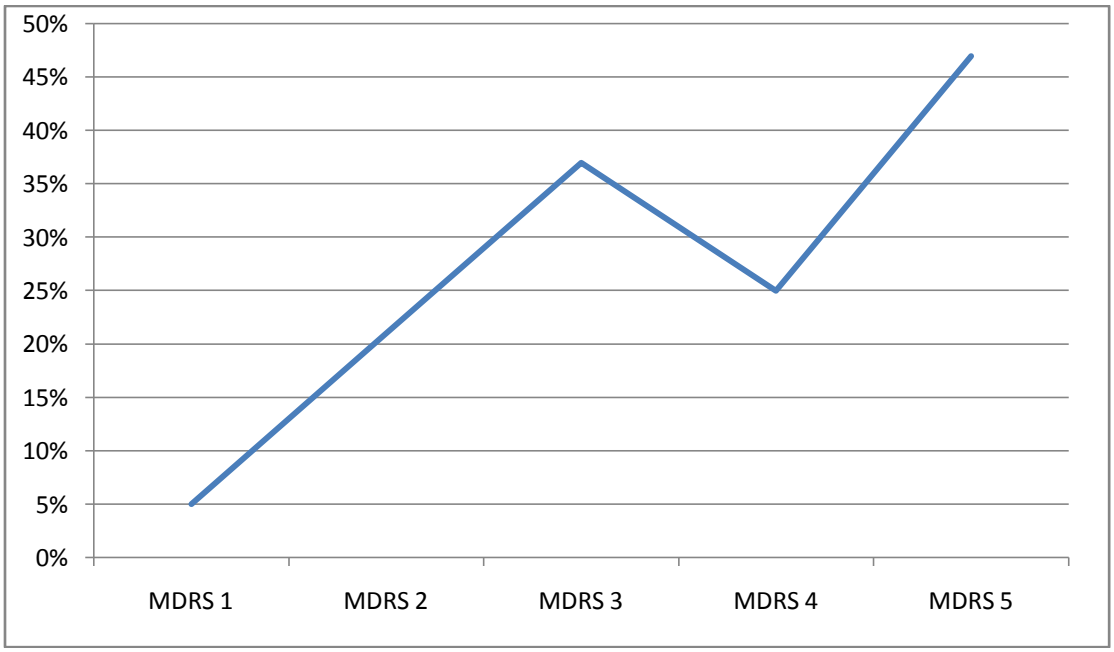

Figure 1:- Distribution of patients with cardiac involvement according to the MDRS score in the serie of S.Chebel et al.

In the same study, the 24 patients with cardiac involvement were also older with a mean age of 40.6 years.

These data are similar to those found in our patient who was 55 years old and had an MDRS score of 4-5 with a documented conductive disorder on the resting ECG.

Resting echocardiography did not show any structural abnormality in our patient. Some series such as Tokgozolu [11] have described the existence of mitral prolapse in 0.33 to 30% of patients with steinert's disease.

A mitral profile of relaxation disorder was found in our patient in the analysis of LV filling pressures.

The impairment of diastolic function is reported in the literature as highlighted by the series of Fragoala and Finsterer [12, 13], who linked this abnormality of left ventricular relaxation to intrinsic myocardial impairment.

The study by Lam and al [14] confirms the hypothesis of an altered ventricular myocardium by showing that the DMPK protein involved in DM1 is mainly expressed in skeletal muscle and cardiac muscle.

In the series of De Ambroggi and al. various myocardial structural abnormalities have been reported in patients with myotonia [15]. They report the presence of LV or RV hypertrophy, RV dilatation or areas of biventricular fatty infiltration [14, 15].

Histological changes in myocardial structure are variable in steinert's disease.

A myocardial fatty infiltration, myofibrillar degeneration, as well as focal myocardial lesions and mitochondrial abnormalities can be found [16].

These pathological and structural lesions may looks like arrhythmogenic heart disease of the RV and may be differentially diagnosed with DM1 [16, 17].

Cardiac MRI is valuable in this indication to look for abnormalities in LV kinetics and systolic function pointing to arrhythmogenic LV dysplasia.

RV angioscintigraphy based on analysis of the ventricular volume curve and parietal kinetics has been shown to be a great exam to decide between DAVD and DM1 with an interobserver reproducibility of 96% [17].

In the series by Guludec et al, this exam was also proposed because of its safety for systematic family screening in relatives of patients with Steinert's myotonia [18, 19].

The LV ejection fraction remains preserved for a long time in DM1, and assessment by a Global longitudinal strain or cardiac MRI performed early could eventually constitute screening tests for systolic dysfunction. But no observational study has been carried out.

Apart from structural abnormalities and impairment of diastolic function, impairment of the conduction pathways remains the most life threatening [20, 21].

The presence of conductive disorder on the surface ECG should justify electrophysiological exploration with measurement of the HV interval, as it is reported in the series of S. Chebel and Lazarus, the origin of this conductive impairment is below the his in 50% of cases.

A electrophysiological exploration was performed in our patient and allowed us to measure an HV at 100 ms indicating the implantation of a double chamber pacemaker. The procedure was uneventful.

The indication for pacemaker implantation in steinert's disease varies according to the series from 3.5 to 19% of patients [22]. The predictive factors for sudden death related to a conductive disorder on the resting ECG were described by Groh [23]. In this study, he points out that the risk of sudden death is multiplied by 3.3 in the presence of:
- Non-sinus rhythm
- PR >240ms
- QRS>120ms
- 2nd or 3rd degree BAV

Many authors emphasize the lack of prevention of sudden cardiac death in patients with pacemakers and DM1 [24].

The etiologies put forward in these sudden deaths are ventricular rhythm disorders, dominated by ventricular tachycardias. The anatomical basis for these arrhythmias is the myocardial fibrous caused by steinert's myotonia. One of the mechanisms suggested for sustained VT is branch-to-branch re-entry [25].

The indication of an ICD for primary prevention in these patients is not reported in the recommendations. On the other hand, the choice of an ICD rather than conventional cardiac pacing appears to be judicious following a resuscitated VT or VF and in the context of resynchronisation in a patient with LV systolic dysfunction [25, 26].

The long-term prognosis of steinert's myotonia is worsening with a decreased survival at 65 years compared to the general population [27].

Conclusion:-

Myotonic dystrophy type 1 causes systemic damage, the most common cause of sudden death being cardiac damage.

Asymptomatic cardiac forms, regardless of the degree of neurological impairment, are frequent.

Careful and repeated screening appears to be one of the preventive measures for these patients in order to orient them towards optimal management.

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