Fast Progressing His-Purkinje Conduction Disturbances în a Myotonic Dystrophy Patient

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ABSTRACT

Myotonic dystrophy leads to multiple systemic complications and the age of death is earlier in myotonic dystrophy patients than in the general population. These patients have a high frequency of sudden death related to respiratory failure, cardiac arrhythmias and in particular to cardiac conduction disturbances. Prophylactic pacemaker implantation should be considered in asymptomatic myotonic dystrophy patients, which in the early stages of disease present minor conduction disturbances in 12-leads ECG. Even if the rate of progression of conduction abnormalities is usually slow, fast progression has been often observed thus making the clinical course of individual patients rather unpredictable.

Keywords: myotonic dystrophy, Steinert’s disease, conduction disturbances, electrophysiological study, pacemaker implantation.

INTRODUCTION

Myotonic dystrophy (DM), the most common dystrophy in adults, is an autosomal dominant disease with two genetically distinct forms: type 1 (DM1), the classic form – described by Steinert – occurs when DMPK gene on chromosome 19 contains an abnormally expanded section, and type 2 (DM2) – identified by Ricker – caused by an abnormally expanded section in ZNF9 gene on chromosome 3. In general, DM2 has a better overall prognosis than DM1.

Cardiac involvement in the classical form is characterized by supraventricular and ventricular arrhythmias, myocardial dysfunction and ische-
mic heart disease and conduction system abnormalities. The His-Purkinje system is most frequently involved, but any part of the conduction system may be affected (1).

Progressive evidence of heart block such as lengthening PR interval or the development of bundle branch block in the presence of clinical symptoms (syncope, presyncope) are found to be the best predictors when considering pacemaker implantation. Asymptomatic AV and His-Purkinje system conduction delays, in particular in the presence of a prolonged HV interval, represent one of the major therapeutic challenges in DM1. Indication for prophylactic pacing in the absence of clinically relevant bradyarrhythmias is an open issue, as data on progression to complete atrioventricular block are conflicting (2).

CASE REPORT

A 49-year-old female, known with classic DM1 diagnosed through genetic study (Table 1) is admitted for new occurred bundle branch block (RBBB). Electrocardiography: sinus rhythm, 75/minute, PR 185 ms, RBBB (Figure 1A). Trans-thoracic echocardiography revealed rather small cavities and normal ejection fraction. Given the high risk of sudden cardiac death in this particular category of patients, we decided an early invasive approach through an electrophysiological study (EPS). An intermittent second degree block and a left anterior fascicular block were recorded (Figure 1B) and the HV interval was prolonged – 81 ms (Figure 1C). In conclusion, we are faced with a diffuse disease of the cardiac conduction system which affects the His-Purkinje system. Classical indication being met, a dual chamber pacemaker was implanted, with a favorable postoperative evolution.

DISCUSSION

Asymptomatic AV conduction delay, in particular in the presence of a prolonged HV interval, represents one of the major therapeutic challenges in DM1, as data on the rate of progression to complete atrioventricular block are conflicting. In presence of prolonged HV interval more than or equal to 70 ms, His-Purkinje system disease is clear and one can discuss the implantation of a prophylactic pacemaker, even in the absence of symptoms (5, 6).

According to the guidelines permanent pacing in acquired atrioventricular block in adults is class I indication in advanced second-degree AV block at any level, associated with myotonic muscular dystrophy, Class IIa in asymptomatic second-degree AV block at intra- or infra-His levels found at electrophysiological study and asymptomatic type II second-degree AV block with a narrow QRS; when type II second-degree
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AV block occurs with a wide QRS, including isolated right bundle-branch block, pacing becomes a class I recommendation. Myotonic muscular dystrophy with any degree of AV block (including first-degree AV block), with or without symptoms, because there may be unpredictable progression of AV conduction disease, is a Class IIb indication for Pacing (7, 8).

Indications for EPS in DM1 patients are still debated (9), but an early invasive approach brought, in our case, a clear indication for pacemaker implantation.

**CONCLUSION**

Considering the unclear rate of cardiac disease progression and the risk of sudden death, an attitude in favor of invasive procedures is preferred, especially in some subsets of DM patients: disturbances and/or symptoms suggestive of arrhythmias, family history, non-invasive findings suggestive of intra- or infrahisian AV conduction disturbances (with or without symptoms), sinus node dysfunction (with or without symptoms), ventricular arrhythmias (with or without symptoms). The early invasive approach was clearly in favor of pacemaker implantation despite the fact that the patient was asymptomatic and presented only a right bundle branch block and border PR interval on ECG. So, an EPS for a minimal conduction disturbance, in the DM1 patients, can uncover a severe diffuse disease of cardiac conduction system.

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### Table 4

<table>
<thead>
<tr>
<th>Phenotype</th>
<th>Clinical Signs</th>
<th>CTG Repeat Size</th>
<th>Age of Onset</th>
<th>Average Age of Death</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Mutable normal (premutation)</strong></td>
<td>None</td>
<td>35-49</td>
<td>-</td>
<td>-</td>
</tr>
<tr>
<td><strong>Mild</strong></td>
<td>Cataracts</td>
<td>50–150</td>
<td>20-70 years</td>
<td>60 years to normal life span</td>
</tr>
<tr>
<td><strong>Classic</strong></td>
<td>Weakness</td>
<td>-100-1000</td>
<td>10-30 years</td>
<td>48-55 years</td>
</tr>
<tr>
<td><strong>Congenital</strong></td>
<td>Infantile hypotonia</td>
<td>&gt;1000</td>
<td>Birth to 10 years</td>
<td>45 years</td>
</tr>
<tr>
<td></td>
<td>Respiratory deficits</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Intellectual disability</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Classic signs present in adults</td>
<td></td>
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<td></td>
</tr>
</tbody>
</table>

*DM1 is caused by expansion of a CTG trinucleotide repeat in the non-coding region of DMPK. CTG repeat length exceeding 34 repeats is abnormal. Our Patient had over 300. The correlation of phenotype and CTG repeat length places the patient in classical Type DM, which involves cardiac disturbances (3, 4).

**REFERENCES**

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