THE HEART IN MYOTONIA ATROPHICA

BY

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Myotonia atrophica is a familial complaint appearing in young adult life. Atrophy of muscles is conjoined with increased tone. Early baldness, with wasting of the temporal, facial, and sterno-mastoid muscles when the disease is fully developed, give to the patient a characteristic appearance (Fig. 1). Atrophy also takes place in the girdle and limb muscles.

Fig. 1.—Two patients (Cases 7 and 13) showing characteristic features of myotonia atrophica.

Increased tone is illustrated by the delayed relaxation of the hand-grip, by dimpling of the tongue when struck by a spatula or by a patella hammer against the lower teeth, and by the adduction jerk of the thumb when the thenar eminence is struck. Cataract (seen only by the slit-lamp during the early years of the disease) and atrophy of the testes with impotence are other characteristic findings. The condition is not uncommon, but because such obvious changes are absent during the early stages of the illness, it may go unrecognized for many years. The medical history of patients in whom I examined the heart convinced me of the need to implement the myopathic signs with others that might lead to an earlier diagnosis and spare the patients the unfair judgment passed on them by employers and doctors alike in regarding them often as nervous subjects or even malingerers. I found that signs discovered during the examination of the cardiovascular system contribute to the surer and earlier diagnosis of myotonia atrophica.

In the English literature dealing with myotonia there is little reference to its effects on the heart. Adie and Greenfield (1923) described 20 cases and mentioned that in one examined at necropsy the heart was healthy and in another the electrocardiogram was normal; this led them to say that the observation of the heart being affected in myotonia lacked confirmation. It was Maas and Zondek (1920) who first wrote about involvement of the heart,
reporting cardiac enlargement in one case which also showed sinus bradycardia and a prolonged P–R interval, and similar changes in the cardiogram of two other patients. In seven patients examined by Guillain and Rouquès (1932) the P–R period was slightly prolonged in two only. Sinus bradycardia was the only change noticed by Havier and Decourt (1933) in one case, but in another reported by Mondon and Pasquet (1939) the P–R interval was long and the T wave was inverted in lead I. Two out of three young patients with myotonia described by Carrillo (1941) had abnormal cardiograms, showing low voltage of the P wave, long P–R interval, wide and notched QRS complex, and left axis deviation. Segura and Lanari (1941), on the other hand, described eleven patients with different forms of myotonia in whom clinical, cardiographic, and radiological examination had shown no abnormality of the heart, while microscopy in one instance demonstrated a healthy myocardium.

In order to test these different opinions on the state of the heart in myotonia atrophica, thirteen cases were collected for special examination. Ten were males and three females. Their ages varied from 23 to 45 and the average age was 35 years. Each had been diagnosed by a neurologist as a typical case of myotonia atrophica.

Cardiac symptoms. The most prominent symptoms were always those identified with the myopathy. Two patients (Cases 12 and 13), in whom the heart was enlarged, complained of breathlessness but neither showed signs of heart failure. Two attacks of unconsciousness in one patient (Case 12) had the features of Stokes-Adams attacks although they were not directly observed. None complained of heart pain.

The pulse. In only two cases was the pulse slow and in one of these the sinus bradycardia alternated with heart block. The force of the pulse was often noticeably small and this change was so common (8 out of 13) as to suggest that it is a characteristic sign in myotonia atrophica.

The blood pressure. The blood pressure was not raised in any of the 13 cases. It was highest (160/70) in Case 13 during periods of 2 : 1 heart block. In five it was low and in three of these the systolic pressure was 95 or under (see Table I). In one case administration of desoxycorticosterone acetate raised the blood pressure and this effect is shown in Fig. 2. It is not maintained that this is a specific effect in myotonia atrophica and there was no change.

**Fig. 2.**—Blood pressure chart in Case 12 to show effect of treatment by, desoxycorticosterone acetate (D.O.C.A.). Continuous line represents systolic and discontinuous line diastolic pressure. Arrows indicate intramuscular injection of 10 mg. of D.O.C.A.
### TABLE 1

Cardiovascular Changes in 13 Cases of Myotonia Atrophica

<table>
<thead>
<tr>
<th>Case No.</th>
<th>Age</th>
<th>Pulse Rate</th>
<th>Pulse Character</th>
<th>Blood Pressure</th>
<th>First Heart Sound</th>
<th>Voltage of P Wave</th>
<th>P-R Interval (sec.)</th>
<th>Notching of QRS</th>
<th>Other Changes</th>
<th>Size of heart at cardioscopy</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>23</td>
<td>70</td>
<td>Normal</td>
<td>120/85</td>
<td>Normal</td>
<td>Low</td>
<td>0.20</td>
<td>Absent</td>
<td>Deep Q in III.</td>
<td>Normal</td>
</tr>
<tr>
<td>2</td>
<td>29</td>
<td>90</td>
<td>Small</td>
<td>120/95</td>
<td>Splitting</td>
<td>Normal</td>
<td>0.20</td>
<td>Present</td>
<td>Left axis deviation</td>
<td>Appears small</td>
</tr>
<tr>
<td>3</td>
<td>31</td>
<td>70</td>
<td>Normal</td>
<td>115/80</td>
<td>Distant</td>
<td>Low</td>
<td>0.20</td>
<td>Present</td>
<td>Left axis deviation</td>
<td>Normal</td>
</tr>
<tr>
<td>4</td>
<td>31</td>
<td>100</td>
<td>Small</td>
<td>125/100</td>
<td>Splitting</td>
<td>Normal</td>
<td>0.21</td>
<td>&quot;</td>
<td>Variation in electrical potential</td>
<td>&quot;</td>
</tr>
<tr>
<td>5</td>
<td>45</td>
<td>70</td>
<td>Normal</td>
<td>135/85</td>
<td>&quot;</td>
<td>&quot;</td>
<td>0.21</td>
<td>&quot;</td>
<td>Left axis deviation</td>
<td>&quot;</td>
</tr>
<tr>
<td>6</td>
<td>36</td>
<td>85</td>
<td>Normal</td>
<td>140/85</td>
<td>Normal</td>
<td>&quot;</td>
<td>0.22</td>
<td>&quot;</td>
<td>T flat in I, S deep in I and II</td>
<td>Appears small</td>
</tr>
<tr>
<td>7</td>
<td>42</td>
<td>85</td>
<td>Normal</td>
<td>90/80</td>
<td>&quot;</td>
<td>Normal</td>
<td>0.22</td>
<td>&quot;</td>
<td>T low in I, Left axis deviation</td>
<td>&quot;</td>
</tr>
<tr>
<td>8</td>
<td>31</td>
<td>75</td>
<td>&quot;</td>
<td>110/85</td>
<td>Normal</td>
<td>&quot;</td>
<td>0.24</td>
<td>&quot;</td>
<td>Variation in electrical potential</td>
<td>&quot;</td>
</tr>
<tr>
<td>9</td>
<td>31</td>
<td>82</td>
<td>&quot;</td>
<td>95/80</td>
<td>Splitting</td>
<td>&quot;</td>
<td>0.25</td>
<td>&quot;</td>
<td>&quot;</td>
<td>Slight enlargement</td>
</tr>
<tr>
<td>10</td>
<td>32</td>
<td>85</td>
<td>&quot;</td>
<td>120/75</td>
<td>Low</td>
<td>&quot;</td>
<td>0.26</td>
<td>Absent</td>
<td>&quot;</td>
<td>&quot;</td>
</tr>
<tr>
<td>11</td>
<td>41</td>
<td>50</td>
<td>Normal</td>
<td>125/95</td>
<td>Distant</td>
<td>&quot;</td>
<td>0.30</td>
<td>Present</td>
<td>Atypical bundle branch block</td>
<td>Moderate enlargement</td>
</tr>
<tr>
<td>12</td>
<td>41</td>
<td>72</td>
<td>Normal</td>
<td>85/75</td>
<td>4th heart sound</td>
<td>&quot;</td>
<td>0.31</td>
<td>&quot;</td>
<td>Left axis deviation</td>
<td>&quot;</td>
</tr>
<tr>
<td>13</td>
<td>44</td>
<td>37-48</td>
<td>Normal</td>
<td>160/70</td>
<td>Systolic murmure</td>
<td>&quot; or 2 : 1 block</td>
<td>0.31 or 2 : 1 block</td>
<td>&quot;</td>
<td>Deep Q and S in I</td>
<td>&quot;</td>
</tr>
</tbody>
</table>
in the myopathic signs although the patient felt much better during the time the blood pressure was raised by the adrenal therapy.

*The first heart sound.* Impurity of the first heart sound was a common finding and the sound was normal in three cases only. The association of splitting of the sound with a prolonged P–R interval in the cardiogram is shown in Table I. Once a prolonged P–R period gave rise to a triple rhythm from the addition of the fourth heart sound.

*Cardiographic changes.* A lengthened P–R period was the commonest change in the electrocardiogram. In none of the thirteen cases was the period less than 0·20 sec.; it was 0·24 or greater in five and in two patients it measured 0·30 and 0·31 respectively. In the last case, 2 : 1 heart block was more often present than sinus rhythm (Fig. 3 and 4). In two,

![Fig. 3.—Cardiogram in Case 13 during phase of sinus bradycardia. Prolonged P–R period. Small P waves. Slurring of QRS complexes.](image)

![Fig. 4.—Cardiogram in Case 13 during phase of 2 : 1 heart block.](image)

injection of atropine and tachycardia induced by glyceryl trinitrate did not affect the length of the period. Notching of the QRS complex was another common cardiographic abnormality and it was present in nine patients. The Q–S interval, however, was only once greater than 0·10 sec. when it was 0·15 (Fig. 5), giving to the tracing the appearance of left bundle branch block, but unlike such a curve, the T wave was upright in leads I and CR7. A low voltage of the P wave was the third characteristic change in the electrocardiogram of myotonia atrophica (Fig. 5 and 6); it was present in eight. In the remaining five, the P in lead I was invariably low, but as it was normal in one of the other leads it has not been counted as an
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abnormal change. Thus, a well-formed P wave in lead I is an unlikely finding in the cardiogram of a patient with myotonia atrophica. Left axis deviation (Fig. 6) was another common finding (7 out of 13). This was found in the absence of hypertension; it was present when

the blood pressure was low, although it was a feature in three of the four patients in whom the heart was found enlarged at cardiography. In two there was no enlargement and the heart appeared to be small in two other cases. Three patients showed a varying electrical potential of the QRS complexes (Fig. 7). It is not suggested that this is a change common to

myotonia for it is not infrequently seen in tracings from healthy subjects, although the incidence of this sign in this series (3 in 13) is somewhat high. Changes in the Q, S, and T waves were inconstant and they have been listed in Table I.
Neither characteristic lens opacity (present in seven), family history of the complaint (common to eight cases), age of the patient, duration of symptoms (over five years in five), nor the severity of the myopathic symptoms, appeared to determine the presence of such cardiographic irregularities.

**Size of the heart.** No obvious cardiac enlargement could be made out in any of the cases on clinical examination, although the apex beat was a little displaced outwards in two. The actual size of the heart, therefore, was estimated at cardioscopy. In five patients it was judged to be normal, and in four, smaller than was expected when age and stature received due attention (Fig. 8). In the remaining four the heart was enlarged, slight in two (Fig. 9) and moderate in two (Fig. 10 and 11). The presence of cardiac enlargement was closely
related to, if not dependent on, the length of the P–R period. Thus, when the P–R was 0.24 sec. or less, the heart was not enlarged; in the two cases showing slight enlargement the P–R interval was 0.25 and 0.26, and 0.30 and 0.31 sec. in two others in whom the heart was moderately enlarged. These last two complained of breathlessness on exertion, but there was no evidence of heart failure in either.

**SUMMARY AND CONCLUSIONS**

Examination of the heart in 13 cases of myotonia atrophica has shown that the presence of cardiovascular signs may help in the earlier diagnosis of the condition.

The pulse is often small and occasionally infrequent. The blood pressure is sometimes very low. The first heart sound in the mitral area commonly shows splitting, and sometimes triple rhythm may appear from addition of the fourth heart sound, this depending on the degree of elongation of the P–R period.

The changes that commonly characterize the electrocardiogram include elongation of the P–R period, low voltage of the P wave, slurring of the QRS complex, and left axis deviation.

The size of the heart varies so that it may be normal or may appear small, but in the presence of considerable lengthening of the P–R period, moderate enlargement takes place.

I wish to thank Dr. John Parkinson, Physician to the Cardiac Department, for his helpful criticism of this paper, and also several colleagues who kindly referred to me cases for this investigation.

**REFERENCES**