The Electrocardiogram in Duchenne Progressive Muscular Dystrophy

By Cecylia Słucka, M.D.

SUMMARY
Interpretation of the electrocardiogram in the Duchenne form of progressive muscular dystrophy (PMD) is still debated in the literature. The group studied consisted of 106 patients with the Duchenne type of PMD, aged 3 to 29 years. The duration of the disease varied from a few months to 15 years. One hundred one patients had abnormal electrocardiograms. These were of four types: type a (64 cases) showed tall R waves in V1 and deep Q waves in V4-6; type b (24 cases), RSr' and deep Q waves in V3-6; and type c (13 cases), polyphasic R waves in V1.

The view that the electrocardiographic abnormalities in Duchenne dystrophy are a manifestation of the dystrophic process in the myocardium is discussed. The authors' material, however, failed to support this view but did support the view that the electrocardiographic abnormalities have an heredofamilial character.

Additional Indexing Words:
Familial right bundle-branch block
ECG pattern of children

INTERPRETATION of the electrocardiogram in the Duchenne form of progressive muscular dystrophy (PMD) is still debated in the literature. Numerous reports, especially early ones, describe a variety of changes—electrical axis deviations, conduction defects, arrhythmias, and other abnormalities.

These reports, however, usually do not distinguish between the various forms of PMD. In more recent work where this distinction is clear, the electrocardiographic patterns of the Duchenne form are similar.

The important features are a tall R wave in lead V1 with the resulting abnormal R/S ratio in this lead, and a deep Q wave, most commonly in leads V5 and V6 and often seen in the limb leads. The complex RSr' in lead V1 and the features of an incomplete right bundle-branch block have also been frequently reported.

Frequent sinus tachycardia, a shortened P-R interval, and a tall R wave in lead V5 are other findings that have been emphasized.

This electrocardiographic pattern has by now become accepted as typical of Duchenne dystrophy and is very useful in differentiating it from the other forms of PMD. While its quality of being typical is no longer under debate, its interpretation still is.

As is known, the Duchenne form of PMD is an hereditary, recessive sex-linked disease that in principle affects only boys. It begins in the first years of life and usually does not run beyond the thirtieth year because of early fatal and sudden termination, usually in the second decade, and often in the course of pneumonia. Severe circulatory failure has been described as another cause of death. It usually develops only a short time before death, without any symptoms having been in evidence a few months earlier.
All reports based on a large number of patients with the Duchenne form emphasize manifestations of heart failure as rare.\textsuperscript{18,21,24,25} Whether the patients died of heart failure, pneumonia, or other causes, autopsy revealed dystrophic lesions in the myocardium resembling those in skeletal muscles\textsuperscript{8, 15, 18, 19, 21}, namely, atrophy of muscle fibers and hypertrophy of connective tissue, occasionally very extensive, without signs of either inflammation or changes in the coronary vessels.

**Group Studied**

Our material comprised 106 patients with the Duchenne type of PMD. The diagnosis was based on the case history, neurological examination, biochemical analyses, electromyography, and histological findings. The ages of the patients ranged from 3 to 29 years (table 1).

Among the patients were 10 groups of siblings: five brothers, four brothers, three brothers, and seven pairs of brothers.

The duration of the disease was a few months to over 15 years (table 2).

The degree of immobilization varied: fairly ambulant, 69 patients; poorly ambulant, 14 patients; and immobilized, 23 patients.

Chest deformities were seen in 34 patients. Estimates of heart size were unreliable owing to changes in the chest. Physical and roentgenological examination did not indicate cardiomegaly. Valvular disease was not diagnosed in any of the patients, and only one showed symptoms of heart failure. One, an 18-year-old patient with no previous symptoms of circulatory insufficiency, died after collapse and pulmonary edema which had ensued in double pneumonia. At necropsy both ventricles were enlarged, the left much more so, with gray patches of connective tissue visible to the naked eye in its side and back wall. Microscopy revealed striking atrophy of the left ventricular muscle with growth of connective tissue, similar changes in the right ventricle, and none in the interventricular septum.

In all cases tracings were recorded from 12 standard leads with a Siemens Cardirex instrument. The electrocardiograms of children were judged by Nadas' and Ziegler's standards.\textsuperscript{27,28}

**Electrocardiographic Findings**

The patterns were pathological in 101 cases and normal in five. Essentially, the changes concerned the QRS complex. Rhythm abnormalities other than sinus tachycardia were not observed, nor were conduction defects other than a shortening of the P-R interval. The most common findings were an overly tall R wave and an abnormal R/S ratio in lead V\textsubscript{1}, or an altered shape of R in the same lead, namely, the RSR' pattern, or a polyphasic R wave in V\textsubscript{1}, occasionally resembling the letter M (table 3).

These changes were usually accompanied by deep Q waves not exceeding 3 mm in width, most commonly in leads V\textsubscript{5} and V\textsubscript{6}, more rarely in the limb leads.

Changes in the T wave and electric axis deviations were rare. A tall R wave was also seen in lead V\textsubscript{5}.

In 16 patients deep Q waves were seen in leads V\textsubscript{5} and V\textsubscript{6} and in limb leads or V\textsubscript{L}.

The 101 patients with abnormal electrocardiograms were divided into four groups.

### Table 1

<table>
<thead>
<tr>
<th>Age of Patients</th>
<th>No. of patients</th>
</tr>
</thead>
<tbody>
<tr>
<td>0-2 yr</td>
<td></td>
</tr>
<tr>
<td>3-5 yr</td>
<td>7</td>
</tr>
<tr>
<td>6-10 yr</td>
<td>49</td>
</tr>
<tr>
<td>11-15 yr</td>
<td>38</td>
</tr>
<tr>
<td>16-19 yr</td>
<td>9</td>
</tr>
<tr>
<td>20-29 yr</td>
<td>3</td>
</tr>
<tr>
<td>Total</td>
<td>106</td>
</tr>
</tbody>
</table>

### Table 2

<table>
<thead>
<tr>
<th>Duration of the Disease</th>
<th>No. of patients</th>
</tr>
</thead>
<tbody>
<tr>
<td>0-2 yr</td>
<td>9</td>
</tr>
<tr>
<td>3-5 yr</td>
<td>28</td>
</tr>
<tr>
<td>6-10 yr</td>
<td>47</td>
</tr>
<tr>
<td>11-15 yr</td>
<td>18</td>
</tr>
<tr>
<td>16 or more</td>
<td>4</td>
</tr>
<tr>
<td>Total</td>
<td>106</td>
</tr>
</tbody>
</table>

_Circulation, Volume XXXVIII, November 1968_
according to the type of electrocardiographic patterns (fig. 1). Group a was composed of those whose electrocardiograms exhibited the type a pattern with tall R waves and an abnormal R/S ratio in lead VI; This abnormality usually was associated with deep Q waves, commonly in V₅-₆, and less so also in limb leads. Group b was made up of those with electrocardiograms with the RSr' complex in V₁. These electrocardiograms often also showed tall R waves in V₁ and deep Q waves. The electrocardiograms in group c showed polyphasic R waves in lead V₁; the tracings were reminiscent of incomplete and
Table 3

Electrocardiographic Data

<table>
<thead>
<tr>
<th>Abnormality</th>
<th>Cases</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sinus tachycardia (over 90/min)</td>
<td>81</td>
</tr>
<tr>
<td>Shortened P-R interval</td>
<td></td>
</tr>
<tr>
<td>0.1-0.12 sec</td>
<td>63</td>
</tr>
<tr>
<td>0.13-0.16 sec</td>
<td>43</td>
</tr>
<tr>
<td>Tall R wave in V₁</td>
<td></td>
</tr>
<tr>
<td>(45 cases* + 34 cases†)</td>
<td>79</td>
</tr>
<tr>
<td>Abnormal R/S ratio in V₁</td>
<td>75</td>
</tr>
<tr>
<td>Complex Rs'r</td>
<td>24</td>
</tr>
<tr>
<td>Polyphasic R wave in V₁</td>
<td>13</td>
</tr>
<tr>
<td>Tall R* wave in V₅</td>
<td>33</td>
</tr>
<tr>
<td>Deep Q wave in V₃₆</td>
<td>51</td>
</tr>
<tr>
<td>In I</td>
<td>6</td>
</tr>
<tr>
<td>In III</td>
<td>5</td>
</tr>
<tr>
<td>In V₆</td>
<td>10</td>
</tr>
<tr>
<td>Altered T wave</td>
<td>4</td>
</tr>
<tr>
<td>Left axis deviation</td>
<td>1</td>
</tr>
</tbody>
</table>

*Above mean normals for children.
†Above maximum normals for children.

usually atypical right bundle-branch block (table 4). Group d consisted of normal electrocardiograms.

Three of the group a patients had broad Q waves, exceeding 3 mm. T waves were altered in four cases, and left axis deviation was seen in one. In group b, abnormal T waves were recorded in one patient. In group c, the tracings were suggestive of incomplete atypical right bundle-branch block, showing polyphasic R waves in lead V₁. The duration of the QRS complex was 0.08 sec in nine subjects, and its shape was often not quite typical. The other electrocardiographic abnormalities, such as tachycardia and a shortened P-R interval, were present in all three groups.

Table 4

Cases in Electrocardiographic Groups a, b, and c

<table>
<thead>
<tr>
<th>Group</th>
<th>Tall R in V₁</th>
<th>Abnormal R/S in V₃</th>
<th>Deep Q</th>
<th>Rs'r in V₁</th>
<th>Polyphasic R in V₁</th>
</tr>
</thead>
<tbody>
<tr>
<td>a</td>
<td>41</td>
<td>41</td>
<td>41</td>
<td>—</td>
<td>—</td>
</tr>
<tr>
<td>(64 cases)</td>
<td>19</td>
<td>19</td>
<td>19</td>
<td>—</td>
<td>—</td>
</tr>
<tr>
<td>b</td>
<td>11</td>
<td>11</td>
<td>11</td>
<td>4</td>
<td>4</td>
</tr>
<tr>
<td>(24 cases)</td>
<td>—</td>
<td>—</td>
<td>—</td>
<td>9</td>
<td>—</td>
</tr>
<tr>
<td>c</td>
<td>—</td>
<td>—</td>
<td>4</td>
<td>4</td>
<td>4</td>
</tr>
<tr>
<td>(13 cases)</td>
<td>—</td>
<td>—</td>
<td>—</td>
<td>—</td>
<td>9</td>
</tr>
</tbody>
</table>

Siblings often had very similar electrocardiograms. In a male sibship of five, the electrocardiograms of four were of type b (Rs'r in V₁) and much alike; the same was seen in two other male sibships of two. In a male sibship of four, the electrocardiograms of all were those of type a and similar.

Discussion

The electrocardiographic abnormalities in Duchenne dystrophy have thus far been regarded as a manifestation of the dystrophic process in the myocardium. The following basis is offered for this view: the patients usually do not survive beyond the age of 30 years, and autopsy brings to light dystrophic lesions of the myocardium, which are occasionally far advanced. This view, however, fails to explain two fundamental facts in this form of muscular dystrophy: (1) the absence of the symptoms of heart failure and (2) the uniform, typical electrocardiographic pattern.

The first fact, absence of symptoms of heart failure, has not infrequently been under consideration. The predominant view is that the depressed motor activity of the patients protects them against the onset of heart failure. Cardiac catheterization produced in some of these patients findings suggestive of occult cardiac failure. This explanation is indeed persuasive, but failed to be confirmed in our group of 106 patients, many of whom were ambulant. Frank symptoms of heart failure should have been expected at least in some of these patients, notably in those who had had the condition for over 10 years. The view that the universally seen electrocardiographic pattern is determined by dystrophic lesions in the myocardium seems even less acceptable. None of the known myocardial pathologic processes, whether of vascular or other origin, gives rise to a comparably uniform electrocardiographic pattern. For instance, scleroderma of the heart produces a variety of changes such as arrhythmias, conduction defects, and deformation of any of...
the electrocardiographic waves. Dystrophic changes resulting in myocardial atrophy should in the first place depress the electrocardiographic waves, especially the R wave, and change the shape of the T wave, and this is not in evidence in the pattern already described. Only the abnormal Q wave might be interpreted as evidence of a dystrophic process in the myocardium which, by leading to replacement of muscle by connective tissue, reduces the potentials and thus gives rise to an infarction-like pattern.

A pattern of this kind has been described in scleroderma of the heart, where development of the myocardial connective tissue gives rise to a pattern analogous to that of myocardial infarction: reduction of potentials and an abnormal Q wave.

This was also how this electrocardiographic pattern was interpreted on the basis of two cases in which the deep Q waves in V5-6 were attributed to the profuse development of the connective tissue found at autopsy in the parabasal lateral wall of the left ventricle. The following facts should be opposed to this view of the origin of the deep Q waves: the Q waves of myocardial infarction should be not only deep but above all broad, whereas in dystrophic cases they are clearly narrow, not exceeding 0.03 sec.

Oddly, the abnormal Q wave is almost invariably localized in the side points. Furthermore, this pseudo-infarction wave should go together with a reduction of the amplitude of the R wave owing to a diminished number of myocardial fibers and their replacement with connective tissue.

In dystrophic tracings, however, the reverse is a frequent finding; that is, the R wave V5-6 is either normal or unduly tall. Finally, the view here discussed is contradicted by the case already described, in which patches of connective tissue visible to the naked eye were revealed at necropsy in the lower part of the wall of the left ventricle. In this case the electrocardiogram differed from those of other dystrophic patients in that the Q waves in V5-6 were not only deep but also broad (0.04 sec) with the R waves clearly reduced in height in the same leads.

It is also difficult to explain the invariably tall R waves in lead V1. They may be due to changes in the chest, but an analysis of our material has failed to demonstrate a relation between the degree of chest deformity and the electrocardiographic pattern. Nor has such a relation been found by other authors.

A tall R in lead V1 could be an expression of right ventricular overstrain or pulmonary hypertension. Autopsy, however, has revealed neither hypertrophy of the right ventricle nor a thickening of its myocardium. Catheterization has shown normal pressure in the right ventricle and pulmonary artery.

The tall R wave in lead V1 and deep Q in V5-6 are reminiscent of the pattern described in cases of septal hypertrophy, but findings at necropsy contradict such an explanation by failing to demonstrate this hypertrophy. In our case, too, the septum was normal at autopsy. Exclusively parabasal left ventricular infarctions may give rise to a similar pattern. With massive development of the connective tissue, potential defects in the posterior wall may cause tall R waves in lead V1, and this was the interpretation of the electrocardiogram in two cases. If this explanation were to be accepted, replacement of muscle with connective tissue should be assumed to occur invariably in exactly the same areas.

Another assumption would have to be that the dystrophic process involves the myocardium from the very beginning of the disease and is in all cases equally far gone.

**Table 5**

<table>
<thead>
<tr>
<th>Duration of the Disease in Groups a, b, c, and d</th>
<th>Group a</th>
<th>Group b</th>
<th>Group c</th>
<th>Group d</th>
</tr>
</thead>
<tbody>
<tr>
<td>0-2</td>
<td>6</td>
<td>3</td>
<td>—</td>
<td>—</td>
</tr>
<tr>
<td>3-5</td>
<td>16</td>
<td>6</td>
<td>4</td>
<td>2</td>
</tr>
<tr>
<td>6-10</td>
<td>31</td>
<td>9</td>
<td>4</td>
<td>3</td>
</tr>
<tr>
<td>11-15</td>
<td>10</td>
<td>4</td>
<td>4</td>
<td>—</td>
</tr>
<tr>
<td>16 or more</td>
<td>1</td>
<td>2</td>
<td>1</td>
<td>—</td>
</tr>
</tbody>
</table>

*Circulation, Volume XXXVIII, November 1968*
as is suggested by the uniform electrocardiographic pattern independent of the duration of the disease. This point is borne out by table 5, which shows that the same pattern (type a, b, or c) was seen in patients with very short as well as very long histories.

The table shows that the duration of the disease varied in all groups within a wide range—from 0 to 16 years or more.

The tall R in V1 has also been interpreted as evidence of disordered right ventricular conduction,17 but the vectorcardiogram has shown no such abnormality,21 and detailed postmortem examination of the conducting system has revealed changes in the minute vessels of the pacemaker and antroventricular node and none in the conducting system itself.22

The complex RSR' used to be ascribed earlier exclusively to hypertrophy of the interventricular crista and right ventricle.36 It is seen in mitral lesions, congenital valvular diseases, and pectus excavatum.37 But in spite of a detailed analysis of cases with this complex many have remained unexplained³⁷; it is often seen in healthy subjects,36 is especially frequent in children, and not uncommon in adolescents.37 We have never seen it related to chest deformities, and it has never exceeded 6 mm in V1, which is considered as evidence against its pathological origin.38

The question of the pattern interpreted as reflecting incomplete right bundle-branch block also calls for an analysis. The pattern is not completely typical. Some authors have described a change of pattern from tall R in V1 to that of right bundle-branch block17,18 and have concluded therefrom that the block pattern is evidence of myocardial involvement.

An analysis of our material fails to support this view. We have seen no relation between the type of electrocardiographic abnormalities and the duration of the disease (table 5) or severity of the condition and immobilization.

Tall Rs in V1 and deep Qs in V5-6 are most reminiscent of a child's heart. According to Nadas,27 deep Q waves in V5-6 and in the limb leads are universal in children, and neither their depth nor their breadth have the diagnostic significance that they have in adults. Tall R waves in lead V1 are typical in the first months of life²⁹ and a frequent finding in children.²⁷,²⁸

According to normals for children,²⁷,²⁸ a shortened P-R interval and a tall R wave in V5 are also childhood features. The pattern described, therefore, can be regarded as persistence of the electrocardiographic childhood pattern rooted in genetic factors. Manning and Crop²⁰ suggested persistence of the childhood QRS complex. In this light it would be easy to find an explanation for the patterns in groups b and c. The complex RSR' and the pattern of incomplete right bundle-branch block are frequent in healthy children²⁰ and not uncommon in adolescents.²⁷ The pattern of right bundle-branch block is often seen in clinically healthy subjects without symptoms of myopathy.⁴¹,⁴⁶

There have been descriptions of familial congenital right bundle-branch block.⁴⁷,⁴⁸ The concept of a heredofamilial condition is supported by the frequently similar electrocardiographic pattern in siblings. Investigating mice from a genetically dystrophic litter, Kleinfeld and associates⁴⁹ found no abnormalities in the myocardium, whereas the lesions in the skeletal muscles were typically dystrophic.

Our previous suggestion of a heredofamilial condition was based on material comprising 47 cases of Duchenne dystrophy.⁵⁰ At that time we began studying the electrocardiograms of the mothers of patients. In a certain percentage of cases (detailed results will be reported separately) we found electrocardiograms similar to those of the dystrophic sons.

To sum up, the heredofamilial character of the electrocardiographic abnormalities is suggested by the following findings:

1. The electrocardiographic pattern is strikingly universal and uniform.

2. The type of the changes is unrelated to age, duration of the disease, and its severity.
3. Tall R waves in lead V₁ and deep Q waves in leads V₅ and V₆ bear the closest resemblance to the pattern of the childhood heart.

4. Tracings with the complex RSr' and a polyphasic R wave in lead V₁ are often obtained from healthy subjects and especially often from children.

5. There have been reports of familial occurrence of the features of the right bundle-branch block.

6. Electrocardiograms are strikingly similar among siblings.

7. In a certain proportion of cases mothers and their dystrophic sons have similar electrocardiograms.

References

12. Storstein, O., and Kinge, F. O.: Heart involve-
The Electrocardiogram in Duchenne Progressive Muscular Dystrophy
CECYLIA SLUCKA

Circulation. 1968;38:933-940
doi: 10.1161/01.CIR.38.5.933
Circulation is published by the American Heart Association, 7272 Greenville Avenue, Dallas, TX 75231
Copyright © 1968 American Heart Association, Inc. All rights reserved.
Print ISSN: 0009-7322. Online ISSN: 1524-4539

The online version of this article, along with updated information and services, is located on the World Wide Web at:
http://circ.ahajournals.org/content/38/5/933

Permissions: Requests for permissions to reproduce figures, tables, or portions of articles originally published in Circulation can be obtained via RightsLink, a service of the Copyright Clearance Center, not the Editorial Office. Once the online version of the published article for which permission is being requested is located, click Request Permissions in the middle column of the Web page under Services. Further information about this process is available in the Permissions and Rights Question and Answer document.

Reprints: Information about reprints can be found online at: http://www.lww.com/reprints

Subscriptions: Information about subscribing to Circulation is online at: http://circ.ahajournals.org/subscriptions/