Congestive cardiomyopathy in a family of Becker's X-linked muscular dystrophy

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Summary
The dissociation of gross congestive cardiomyopathy and muscle disease in different members of a family affected with Becker's benign X-linked muscular dystrophy is described. The possibility that cardiomyopathy and muscular dystrophy may be the expressions of the same mutant gene has been suggested.

Introduction
On the assumption that hypertrophic obstructive cardiomyopathy (HOCM) may be the cardiac manifestation of a general myopathy, Hootsmans and Meerschwan (1971) investigated twenty-three patients with HOCM and found electromyographic evidence of skeletal myopathy in fifteen. In support of these observations an interesting family is now reported.

Materials and methods
Five members of a family affected with Becker's benign X-linked muscular dystrophy were the subjects of this study. The details of the pedigree are shown in Fig. 1.

Fig. 1. Pedigree of the affected family. Age □ No. ○. Unaffected male, female; ■ Cardiomyopathy with hypertrophied calf muscle; □ affected male; ○ carrier female; □, ○, dead male, female.

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All the patients were examined clinically and the individual muscles were graded according to the MRC specifications (1943). The tests performed included determination of serum creatine phosphokinase (CPK) levels (Hughes, 1962), electrocardiography, chest X-ray, muscle biopsy and electromyography.

Electromyographic studies were done with a two-channel Medicor Electromyograph. Continuous recordings were obtained on a running photographic film during minimal and maximal volition at a sweep speed of 1 msec/mm and amplitude of 100 μV/mm. Both the patients and the controls were examined by the same concentric needle electrode (Buchthal, Guld and Rosenfalck, 1954). The four quadrant method was used for sampling of the muscles. The room temperature was kept constant at 27°C so that the effect of varying temperature might not alter the duration of motor unit potential (MUP) and the percentage of the polyphasic waves (Buchthal and Pinelli, 1951). The mean of twenty different action potentials from each sample was utilized for final evaluation of duration and amplitude of MUP. The potentials having five or more deflections across the base line were labelled polyphasic and were not included in the determination of mean duration and amplitude of MUP. For accuracy of calculation the film was projected on the screen giving a twelve-fold enlargement. The proximal muscles were selected for electromyography because clinically the weakness was most manifest in these muscles.

Results
The data on skeletal muscle and cardiac involvement are summarized in Tables 1 and 2.

One of the patients (III-4) presented with cardiomyopathy (Figs. 2 and 3) and slight hypertrophy of the right calf muscle with a difference of 2.3 cm in girth. Biopsy from the hypertrophied calf muscle showed variation of fibre size within the fascicles, thin fibres with prominent sarcolemmal nuclei and
Cardiomyopathy in Becker's muscular dystrophy

TABLE 1. Skeletal muscle involvement in affected members

<table>
<thead>
<tr>
<th>Pedigree no.</th>
<th>Age (years)</th>
<th>Age at onset</th>
<th>Muscle weakness*</th>
<th>Calf hypertrophy</th>
<th>Disability</th>
<th>Serum CPK† (μmol/ml/hr)</th>
<th>Dystrophic change in muscle biopsy</th>
</tr>
</thead>
<tbody>
<tr>
<td>I-1‡</td>
<td>67</td>
<td>?30</td>
<td>Face</td>
<td>0</td>
<td>0</td>
<td>+</td>
<td>?</td>
</tr>
<tr>
<td>III-4</td>
<td>26</td>
<td>24</td>
<td>Shoulder</td>
<td>0</td>
<td>0</td>
<td>+</td>
<td>+</td>
</tr>
<tr>
<td>III-5</td>
<td>23</td>
<td>9</td>
<td>Pelvic</td>
<td>0</td>
<td>0</td>
<td>+</td>
<td>+</td>
</tr>
<tr>
<td>III-8</td>
<td>16</td>
<td>8</td>
<td></td>
<td>0</td>
<td>+</td>
<td>+</td>
<td>+</td>
</tr>
<tr>
<td>III-10</td>
<td>12</td>
<td>10</td>
<td></td>
<td>0</td>
<td>+</td>
<td>+</td>
<td>+</td>
</tr>
<tr>
<td>III-15</td>
<td>24</td>
<td>9</td>
<td></td>
<td>0</td>
<td>0</td>
<td>+</td>
<td>+</td>
</tr>
</tbody>
</table>

* Graded from normal (0) to very weak (+ + + +).
† Normal CPK value in serum: 0.39–3.8 μmol/ml/hr.
‡ Patient deceased, information obtained from relatives.

TABLE 2. Cardiac involvement in affected members

<table>
<thead>
<tr>
<th>Pedigree no.</th>
<th>Clinical</th>
<th>X-ray</th>
<th>ECG</th>
</tr>
</thead>
<tbody>
<tr>
<td>I-1</td>
<td>Data not available</td>
<td></td>
<td></td>
</tr>
<tr>
<td>III-4</td>
<td>Extra systole, heart failure</td>
<td>Cardiomegaly</td>
<td>Ventricular extrasystole. Left ventricular preponderance. Non-specific T wave changes</td>
</tr>
<tr>
<td>III-5</td>
<td>Normal</td>
<td>Normal</td>
<td>Inverted T waves in V₆₄</td>
</tr>
<tr>
<td>III-8</td>
<td>Extrasystole</td>
<td>Normal</td>
<td>Ventricular extrasystole Prominent R waves in V₆₂</td>
</tr>
<tr>
<td>III-10</td>
<td>Normal</td>
<td>Normal</td>
<td>Prominent R waves in V₇</td>
</tr>
<tr>
<td>III-15</td>
<td>Normal</td>
<td>Not done</td>
<td>Not done</td>
</tr>
</tbody>
</table>

Fig. 2. Chest X-ray of patient No. III-4 showing cardiomegaly.

In contrast to this patient, the other affected members (I-1, III-5, III-8, III-10, III-15) had varying grades of skeletal muscle weakness without any evidence of cardiac involvement clinically or radiologically, although they had electrocardiographic abnormalities in the form of R and T wave changes in the right precordial leads. None of them had any significant disability. All the patients in whom muscle biopsy was done had histological evidence of muscular dystrophy in the form of variation in the fibre thickness, prominent and centrally placed sarcoclemmal nuclei, areas of necrosis and fatty infiltration. Electromyography revealed myopathic pattern in all the patients (Table 3).

Discussion

In view of the relatively late onset and benign character of the dystrophic process in the affected males and considering the fact that their maternal grandfather survived long enough to have children,
**Fig. 3.** Electrocardiogram of patient No. III-4 showing ventricular extrasystole and non-specific T wave changes.

**Fig. 4.** Longitudinal section from the calf muscle of patient No. III-4 showing myopathic changes (× 60).

**Fig. 5.** Longitudinal section from the calf muscle of patient No. III-4 showing myopathic changes (× 150).
it is felt that the family is affected with Becker's benign X-linked muscular dystrophy. Occurrence of cardiomyopathy in one member of this family, who also had evidence of latent muscular dystrophy, seems to be more than a coincidence. Similar dissociation of muscular dystrophy and cardiomyopathy in different members of a family was also observed by Hootsmans and Meerschwam (1971) in two of their twenty-three patients with HOCM. These facts point to the possibility that idiopathic cardiomyopathy may be an expression of the same mutant gene which is responsible for muscular dystrophy. In view of this, the authors strongly feel that all the patients with idiopathic cardiomyopathy and their relatives should be thoroughly investigated for the presence of primary skeletal muscle disease which may be minimal or clinically absent. It is likely to be missed unless specifically searched for.

Acknowledgments
We thank Dr V. Lalitha and Mr N. K. Banerjee of K.E.M. Hospital, Bombay, for doing the histopathological studies and CPK estimations.

Addendum
The patient (III–10) who initially had mild skeletal muscular dystrophy was seen recently in gross congestive heart failure with cardiomegaly mimicking the cardiac status of III–4. The present electrocardiogram showed left ventricular hypertrophy with non-specific T wave changes.

References


<table>
<thead>
<tr>
<th>Case no.</th>
<th>Deltoid D (μV)</th>
<th>P</th>
<th>Biceps D (μV)</th>
<th>P</th>
<th>Triceps D (μV)</th>
<th>P</th>
<th>Quadriceps D (μV)</th>
<th>P</th>
<th>Gluteus maximus D (μV)</th>
<th>P</th>
</tr>
</thead>
<tbody>
<tr>
<td>Controls</td>
<td>6.8 ± 0.97</td>
<td>&lt;10</td>
<td>7.2 ± 0.86</td>
<td>&lt;10</td>
<td>6.8 ± 0.78</td>
<td>&lt;10</td>
<td>7.0 ± 0.58</td>
<td>&lt;10</td>
<td>6.7 ± 0.62</td>
<td>&lt;10</td>
</tr>
<tr>
<td>III–4</td>
<td>5.0</td>
<td>&lt;10</td>
<td>4.2</td>
<td>&lt;10</td>
<td>4.4</td>
<td>&lt;10</td>
<td>3.4</td>
<td>&lt;10</td>
<td>3.0</td>
<td>&lt;10</td>
</tr>
<tr>
<td>III–5</td>
<td>3.6</td>
<td>&lt;15</td>
<td>4.2</td>
<td>&lt;15</td>
<td>4.4</td>
<td>&lt;15</td>
<td>2.6</td>
<td>&lt;20</td>
<td>2.0</td>
<td>&lt;20</td>
</tr>
<tr>
<td>III–8</td>
<td>5.4</td>
<td>&lt;10</td>
<td>4.5</td>
<td>&lt;10</td>
<td>5.7</td>
<td>&lt;10</td>
<td>2.9</td>
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<td>&lt;15</td>
</tr>
<tr>
<td>III–15</td>
<td>4.0</td>
<td>&lt;10</td>
<td>4.8</td>
<td>&lt;10</td>
<td>5.0</td>
<td>&lt;10</td>
<td>3.2</td>
<td>&lt;15</td>
<td>3.0</td>
<td>&lt;25</td>
</tr>
</tbody>
</table>

Full interference pattern was observed in all the muscles in all the cases.

D, Duration of motor unit potential; P, polyphasic action potential %.

Table 3: Electromyographic studies in the affected members of the family.

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