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The Heart in Muscular Dystrophy: an Electrocardiographic and Ultrasound Study of 20 Patients

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Summary. Twenty patients with different types of muscular dystrophy (MD) were included in a cross-sectional study by means of electrocardiography and ultrasound cardiography. A manifest cardiomyopathy was detected in 8 patients; a latent cardiomyopathy was found in 4. A hypertrophic cardiomyopathy was especially frequent in facioscapulohumeral MD, a congestive cardiomyopathy in Becker-Kiener MD. The ECG showed a reduction in the QT interval and frequent block formers in the X-chromosomal inherited forms and the trunc-girdle form. Bradycardia and a prolonged QT interval were frequent in myotonic dystrophy and facioscapulohumeral MD. Signs of cardiac infarction in the ECG were most frequent in the trunc-girdle forms. A high cardiac output per minute in conjuction with increased left ventricular volume was frequent in Becker-Kiener and Landouzy MD. A left ventricular dysfunction with reduced ejection was characteristic of myotonic dystrophy and trunc-girdle MD. A mitral valve prolapse was more frequent with increasing severity of the muscle disease and was particularly frequent in myotonic dystrophic and Landouzy MD. The cardiac output per minute and the stroke volume were significantly lower ($P \leq 0.03$) where a mitral valve prolapse was present.

Key words: Muscular dystrophy – Cardiomyopathy – Myotonic dystrophy – Heart disease

Introduction

In addition to intercurrent infections and thromboembolic complications, the patient with progressive muscular dystrophy (MD) is in danger from involvement of the heart [1–4]. Acute left ventricular failure in cardiomyopathy and life-threatening arrhythmias are feared complications of cardiac disease. For the various forms of progressive MD, qualitative and quantitative differences of cardiac involvement in the dystrophic process have been described [5–19]. In a cross-sectional study, we investigated 20 patients with various forms of progressive MD using electrocardiography and ultrasound cardiography in order to analyse forms of cardiac involvement and individual risk.

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Patients and Methods

In a cross-sectional study, 20 patients with progressive MD were investigated. The patients were assigned to various forms of MD according to the mode of heredity, distribution pattern and age at onset of the disease (Table 1). In all patients, the diagnosis was confirmed by electromyography and biopsy. At the time of the cardiological diagnosis, the patients were subjected to clinical examination, and the extent of the dysfunctions was determined. Three groups were formed based upon severity:

group 1: Mild pareses – the patient is independent
group 2: Moderate pareses – the patient needs help, but he is ambulatory (with wheelchair)
group 3: Severe pareses – the patient is severely disabled, he is dependent upon the help of others

The electrocardiographic diagnosis was made using a 12-channel resting ECG lead with standard limb and chest leads.

The ultrasound cardiographic diagnosis was carried out both in two-dimensional and in unidimensional form; the diameter of the left ventricle, the thickness of the posterior wall, ventricular sep-

Table 1. Types and severity groups of muscular dystrophy ($n = 20$)

	n	Duration of disease (years)	Severity group		
			1	2	3
Trunc-girdle type	9	22.3	6	–	3
Facioscapulohumeral type	4	23.3	2	1	1
Pelvic girdle	4				
Becker-Kiener type (3)		31.3	1	2	–
Duchenne type (1)		23.0	–	–	1
Curschmann-Steinert myotonic dystrophy	3	22.5	2	2	–
Total	20	26.1	11	5	4

Age: Mean 38.4 years (22–57 years); Sex: 14 men, 6 women; Duration of diseases: mean 26.1 years (range 7–36 years)

tum and the left atrium were analysed by standard methods of measurement. The function of the mitral valve was evaluated by sector scan, and any mitral valve prolapse present was classified as a holosystolic, late systolic or mesosystolic prolapse. In addition, the level of creatinine kinase (CK) and its isoenzymes was determined.

Results

1. Electrocardiographic Findings

In the 20 patients, 13 (65%) showed left preponderance or were indifferent types; 4 cases were vertical types, and there was one case of over-rotated right preponderance, one of over-rotated left preponderance and one sagittal type.

Sixteen (80%) patients showed a sinus rhythm and 4 (20%) exhibited a respiration-independent sinus arrhythmia (chiefly in facioscapulohumeral MD). The heart rate, which was determined both by electrocardiography and by ultrasound cardiography, was normal for 11 patients (55%).

Tachycardia with rates above 100/min occurred in every second patient with X-chromosomal inherited MD ($n = 2$) and in 33% of the trunc-girdle forms ($n = 3$). This finding was present in 25% ($n = 5$) of the total group. Bradycardia (rate < 60/min) occurred in 2 cases, 1 with Curschmann-Steinert myotonic dystrophic (33%) and 1 with facioscapulohumeral MD (25%). The rate of a tachycardia increased with increasing severity of the muscular disorder (mean heart rate – group 1: 85.7; group 2: 75.6; group 3: 96.5).

While the PQ interval was normal in all patients, QRS-changes in the form of block formers (QRS > 0.15) occurred in 7 (35%) cases, especially in facioscapulohumeral MD ($n = 2$; 50%) and in trunc-girdle type ($n = 3$; 33%). Seven patients with normal ST segment and T wave showed a longer (Curschmann-Steinert, facioscapulohumeral form – $n = 2$ each) or shorter (X-chromatosomally inherited MD – $n = 2$; 50%) rate-correlated QT interval ($0.39 \times \sqrt{\text{heart rate}} \pm 0.4$). Signs of cardiac infarction in the EEG were most frequent for the trunc-girdle forms.

The heart rate determined by ultrasound cardiography showed a significant correlation with the duration of the muscular disorder ($r = 0.477$; $P = 0.039$).

2. Ultrasound Cardiographic Findings

An increased diameter of the left ventricle was found particularly in low-severity groups 1 and 2 (35%), especially for Becker-Kiener MD (2 of the 3 patients). On the other hand, severity group 3 showed a reduced ventricular volume in 3 of 4 cases (75%).

The mean stroke volume calculated from the difference between the end diastolic volume and the end systolic volume of the left ventricle was found to be pathologically increased in 2 Becker-Kiener patients (67%) with an average value 102 ml (normal: 55–95 ml). A reduced stroke volume was found especially in patients with trunc-girdle type ($n = 4$; 44.4%) and Curschmann-Steinert myotonic dystrophy ($n = 1$; 33%). Altogether, pathological stroke volumes were found in 9 cases (45%) of the total group.

Table 2. Cardiomyopathies in muscular dystrophy (MD; $n = 20$)

Manifest cardiamyopathy in 8 patients (40%) of the total group (Classification according to Goodwin [15])	
Assymmetrically hypertrophic cardiomyopathy (in 4; 20%)	
facioscapulohumeral MD	$n = 2$
myotonic dystrophy	$n = 1$
trunc-girdle type	$n = 3$
Congestive (dilative) cardiomyopathy (in 2; 10%)	
Becker-Kiener MD	$n = 2$
Hypertrophic congestive cardiomyopathy (in 2; 10%)	
trunc-girdle type	$n = 2$
Latent dilative cardiomyopathy (in 4; 20%) Classification according to Nigro [22]	
facioscapulohumeral MD	$n = 1$
trunc-girdle type	$n = 2$
Duchenne MD	$n = 1$

Both a reduced stroke volume and a lower cardiac output per minute were observed with greater frequency as the severity of the muscular disorder increased. A pathological increase in the cardiac output per minute was found mainly in severity groups 1 ($n = 8$; 73%) and 2 ($n = 3$; 60%), while only 1 patient (25%) in group 3 showed an increased cardiac output per minute. Only 5 patients (25%) of the total group had a cardiac output per minute in the normal range, although in 2 (10%) a reduced stroke volume was corrected by reflex tachycardia.

A thickening of the posterior wall of the left ventricle was found in 2 patients with Becker-Kiener MD (67%). A reduced posterior wall thickness was present in all 4 patients of severity group 3. In the total group, the measured values for the posterior wall of the left ventricle were outside the normal range in 40% of cases.

The stroke fraction, the fibre contraction rate and the contraction fraction were determined as contractility parameters. In contrast to the static measured values, the patients with advanced MD then had pathological symptoms more frequently.

If the various findings by ultrasound cardiography are summarized according to the classification by Goodwin ([20]; Table 2), manifest cardiomyopathy can be diagnosed in 40% of the total group. Dilative cardiomyopathy was present preferentially in the Becker-Kiener form ($n = 2$; 67%). An asymmetrically hypertrophic cardiomyopathy was found in 4 of the total group; it was most frequent in facioscapulohumeral MD ($n = 2$; 50%) and in Curschmann-Steinert myotonic dystrophy ($n = 1$; 33%). Combined dilative and hypertrophic cardiomyopathy occurred in 2 patients of the total group, both in the trunc-girdle type ($n = 2$; 22%). A further 35% of patients ($n = 7$) met the criteria of a latent stage of cardio-

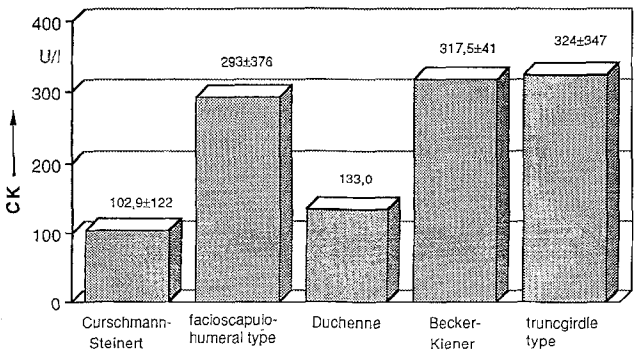

Fig. 1. Mean CK levels in the different types of muscular dystrophy ($n = 20$)

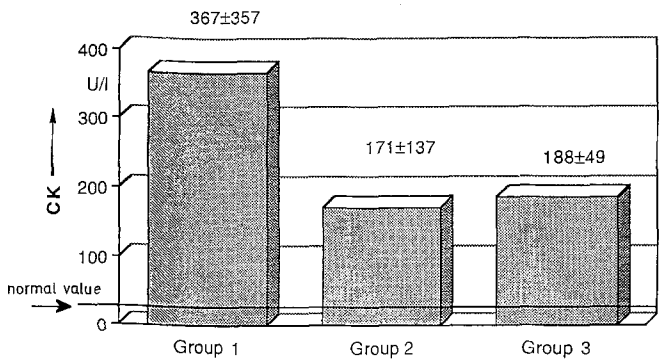

Fig. 2. Mean CK levels in the different severity groups ($n = 20$)

myopathy with readily classifiable pathological symptoms according to Nigro's criteria [21].

Nine patients (45% of the total group) exhibited a mitral valve prolapse, which was holosystolic in 6 cases and late systolic in 3. The patients affected were one patient with Duchenne MD, 3 patients with the facioscapulohumeral form, 3 patients with the trunc-girdle type and 2 patients with myotonic dystrophy. All patients of severity group 3 had a mitral valve prolapse; in severity groups 1 and 2, prolapses were observed in 27% ($n = 3$) and in 40% ($n = 2$) of cases, respectively.

Intergroup comparison of the patients with or without mitral valve prolapse showed that, where a prolapse was present, the cardiac output per minute was significantly lower ($P \leq 0.007$), the heart rate was lower ($P \leq 0.01$) and the concentration fraction was higher ($P \leq 0.0274$) (Wilcoxon U test).

Both the CK values and the CK-MB values were highest for the Becker-Kiener MD and the trunc-girdle type (Fig. 1) – they were pathological in all patients. In severity group 1, the enzyme values were substantially higher than in severity groups 2 and 3 (Fig. 2). There was a significant correlation between the cardiac output per minute and the CK-MB ($r = 0.698$; $P \leq 0.004$).

Discussion

For the different forms of progressive MD, various types of cardiac involvement have been described in the literature. For X-chromosomally inherited MD of the Duchenne type, congestive cardiomyopathy with development of supraventricular arrhythmias is considered typical [12, 15, 16, 22–27].

Congestive cardiomyopathy with a conduction defect is described for the X-chromosomally inherited Becker-Kiener form [2, 3, 28], and detection of a mitral valve prolapse and of a conduction defect is considered characteristic of Curschmann-Steinert myotonic dystrophy [10, 14, 16, 27, 29]. Manifest cardiac arrhythmias are mentioned in particular for Emery-Dreifuss MD [8, 15, 19].

In the present cross-sectional study, congestive cardiomyopathy was found in 67% of the patients with Becker-Kiener MD and hypertrophic cardiomyopathy predominantly in facioscapulohumeral MD and Curschmann-Steinert myotonic dystrophy. In particular, mixed forms occurred in the trunc-girdle type. The X-chromosomally inherited forms frequently showed tachycardias, and bradycardia was encountered most frequently in the Landouzy-Déjerine and Curschmann-Steinert forms. The heart rate increased with an increasing duration of the illness. Block formers of the QRS complex occurred mainly in facioscapulohumeral dystrophy and in the trunc-girdle type. An extension of the QT interval, which is dangerous when cardiac arrhythmias occur [3, 12, 16], was found in the Landouzy-Déjerine and Curschmann-Steinert forms in 75% of cases for each form.

A pathological cardiac output per minute was observed in every patient with Becker-Kiener MD and in 55% of the total group. The cardiac output per minute decreased with increasing severity of the muscular disorder. A mitral valve prolapse was found more frequently with increasing MD. This might be due to the fact that thorax deformities are responsible for the mitral valve prolapse [30, 31].

Patients in the group investigated who appear particularly at risk from cardiac arrhythmias and acute cardiac emergency situations are those with Becker-Kiener MD and those with Curschmann-Steinert myotonic dystrophy. In specific cases, these patients whose disease per se has a favourable long-term prognosis should be treated by a specific cardiological therapy (e.g. cardiac pacemaker). While treatment with the calcium antagonist diltiazem showed no convincing results in cardiomyopathy [32], coenzyme Q improved cardiac function in a double-blind study [33]. In severe cases, especially of Becker-Kiener MD, cardiac transplantation has been performed [3].

As in our study of children and young patients [16], we were unable to detect a mitral valve prolapse in patients with Becker-Kiener MD among the present group of patients. This is in agreement with statements in the literature [15, 19, 31] and may be due to the fact that there are no deformities of the thorax in this group. On the other hand, mitral valve prolapse was frequent in Curschmann-Steinert myotonic dystrophy, in facioscapulohumeral dystrophy and in the trunc-girdle type. This is in agreement with the observations made by other authors [1, 6, 11, 14, 27, 29]. Comparison of patients with or without mitral valve prolapse showed a significantly

higher cardiac output per minute in the group for which no prolapse was detected (7.11 compared with 4.51. U test, $P \leq 0.007$). The haemodynamic situation is therefore more favourable for patients with mitral valve prolapse; however, patients with mitral valve prolapse and conduction defects are more at risk from sudden cardiac arrhythmias [9, 16, 19, 31].

Our observation indicates that a very high percentage of patients with progressive MD are likely to have pathological cardiac symptoms. While certain forms of muscular dystrophy have a statistically high incidence of certain forms of cardiomyopathy, in specific cases, the constellation of symptoms may be very variable and not reliably assignable. In our opinion, what is decisive is regular monitoring of electrocardiographic and ultrasound cardiographic symptoms in sufferers from MD in order to be able to apply cardiological treatment in adequate time.

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