THE RESULTS OF THE ELECTROPHYSIOLOGICAL INVESTIGATIONS IN MUSCLE AGENESIS

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ABSTRACT

Objective: The aim of this study, is to find out whether a myopathic involment accompanies muscle agenesis or not.

Methods: Eleven patients were gathered in this study. The ages of cases were between 17 and 22 (mean: 19.9) years. Needle electromyography was performed in all patients. The serum CK and LDH values in the serum of all the patients were studied and in two cases muscle biopsies were taken from the unaffected side.

Results: The electrophysiological findings in ten out of 11 patients with pectoralis major agenesis (six of them being very pronounced) were in accordance with myopathic involvement. In one case the histopathological examinations were in accordance with myopathic involvement, while there was not any pathological findings in the other one.

Conclusion: As a result, we have concluded that a slowly progressive and benign myopathy may accompany muscle agenesis and this can be demonstrated by our electrophysiological investigations.

Key Words : Muscle agenesis, Poland's Syndrome, Pectoralis muscle agenesis.

INTRODUCTION

It is a known fact that some muscles of the body are not present or do not develop in some people (1-3). The most frequently seen muscle ageneses are pectoralis, trapezius, serratus anterior and quadratus femoris.

It is reported that in some cases with muscle agenesis, fascio-scapulohumeral muscular dystrophy (Landouzy-Déjerine) may develop (4).

In this study, the electromyographic studies were made in 11 cases with congenital muscle agenesis or hypoplasia, and it was investigated whether myopathic involvement accompanied the agenesis.

PATIENTS AND METHODS

Eleven cases referred to our outpatient department who were diagnosed as having muscle agenesis were included in this study. General clinical and neurological examination was made in all patients to find out other abnormalities that may accompany muscle agenesis. Needle electromyography (EMG) was made and serum CK and LDH levels were assessed in all patients. In two patients a muscle biopsy was taken from the non-agenesic side.

The ages of the 11 cases were between 17-22 (mean:19.9) and all patients were male. In 8 patients right-sided, in three patients left-sided pectoralis major muscle agenesis was found; while in one patient there was a right pectoralis major agenesis plus a right trapezius muscle agenesis. In one patient with right pectoralis major muscle agenesis, there was also a hypogenesis of the arm muscles on the same side; in one patient with right pectoralis major muscle agenesis, there was a syndactyly of the 2nd and 3rd fingers on the same side, hypogenesis of the hand on the same side and mild atrophy of the facial muscles. In another patient with right pectoralis major

agenesis, there was a 2-3rd finger syndactyly and mild hypogenesis of the biceps brachii muscle on the same side, and a bilateral pes cavus deformity (Fig. 1) and also in another patient with a left pectoralis muscle agenesis, there was an atrophy of arm muscles on the same side.

The common complaint of all the patients was the presence the weakness of the chest muscles that was present for a long time. The case with a trapezius muscle agenesis had droping and weakness of the shoulder and the case with an atrophy of the arm muscles on the same side with the left pectoralis major muscle agenesis had a weakness of the abduction of the arm from the shoulder and its extension from the elbow, as well as a weakness of the facial muscles (2nd patient-Table I).

Needle EMG of the deltoid and biceps brachii muscles were made in all eleven patients. Additionally, the abductor pollicis brevis muscles were studied in 9 patients, the vastus medialis muscles in 7 patients; and extensor digitorum brevis, abductor digiti minimi, pectoralis, extensor digitorum communis, trapezius, infraspinatus and supraspinatus muscles were studied, each in one patient.

Table I: The results of electrophysiological investigations.

		NEEDLE EMG : MUAP TIMES												
n	Side	D	BB	APB	VM	P	T	ADM	EDC	EDB	SS	IS	MCV	F
1	Right	m↓	m↓				m↓				N	m↓		
2	Left	↓	↓	m↓	↓					↓				
3	Right	N	N			↓								
4	Right	m↓	N	m↓				m↓	m↓				54	24.2
5*	Left	m↓	↓	↓	↓								64	28.8
6°	Left	↓	↓	↓	m↓								57.3	29
7	Right	m↓	↓	↓									61	27.6
8	Right	↓	↓	↓	m↓								57	26.8
9	Right	↓	↓	↓	m↓								62	27.2
10	Right	↓	↓	m↓	m↓									
11	Right	↓	m↓	↓	↓								61	25.2

D: Deltoid, BB: Biceps Brachii, APB: Abductor Pollicis Brevis, VM: Vastus Medialis, P: Pectoralis T: Trapezius, ADM: Abductor Digiti Minimi, EDC: Extensor Digitorum Communis, EDB: Extensor Digitorum Brevis, SS: Supraspinatus, IS: Infraspinatus, m/sec MCV: Motor Conduction velocity (Median nerve, elbow -wrist segment,)
F: Wrist - Abductor Pollicis Brevis, m/sec.
↓: Shortened, m↓ Mildly shortened, N: Normal
(*): Dystrofic findings in biopsy
(°): Normal findings in biopsy

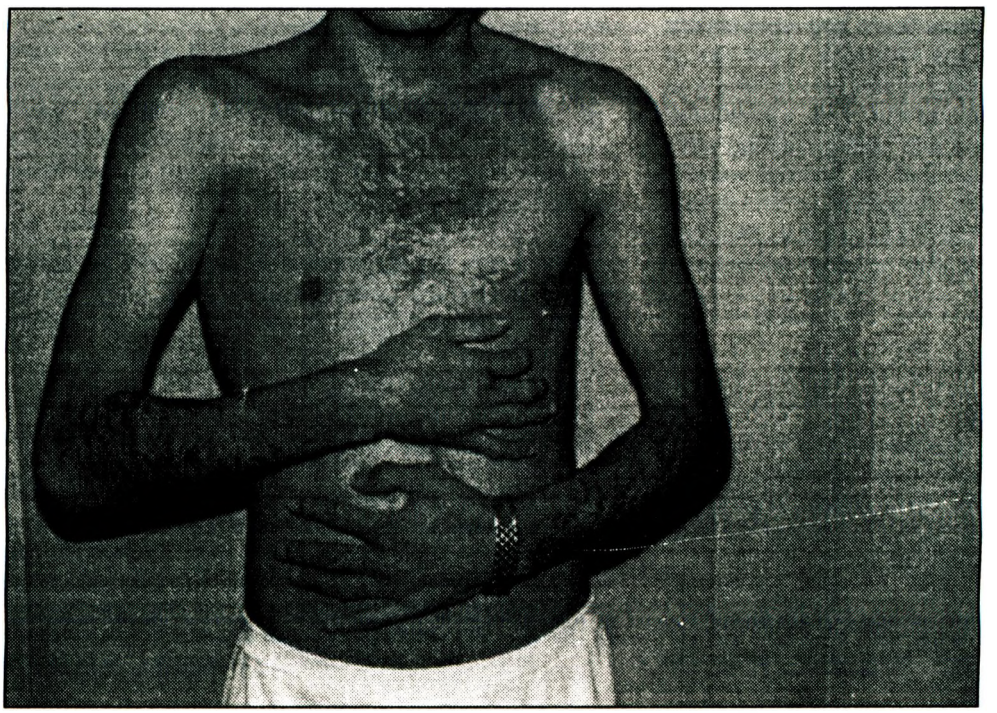

Fig. 1:
The patient with right pectoralis major agenesis, 2nd-3rd finger syndactyly (operated) hypogenesis of biceps brachii muscle.

RESULTS

In one patient with right pectoralis major muscle agenesis, there was shortening in the Motor Unit Action Potential (MUAP) duration, reduced recruitment pattern in maximal contraction and significant reduction in the amplitude at the pectoralis major muscle on the same side while all the other muscles studied had normal electrophysiological patterns. In all the other patients, the MUAP's were shortened, the polyphasic potential rates were increased, yet the distal motor latencies to the muscles were found to be normal. In seven patients the elbow-wrist motor conduction velocity was found to be normal for the median nerve. The F response values were also normal. The results of the elecrophysiological investigations are summarized in the Table I.

The CK and LDH levels were normal in all patients. Muscle biopsies were taken from the deltoid muscles of unaffected sides in two cases. The 2. cms long samples were strecthed on a 1x5x10 cms piece of wood keeping the muscle in its original length sizes. For pathologic examination, half of the sample was taken for frozen sections and the other half was put in procedure with alcohol, acetone, xsilene and paraphine for cross and longitudinal sections. Then the paraphine blocks were prepared and stained with hematoxilene,eosine and examined in light microscope. The sample which was taken for frozen sections, was put in isopropyl alcohol and frozen in liquid nitrogene and again cross and longitudinal sections were taken and stained with oil red 0 and PAS.

In histopathological examination, when and if, atrophy and disappearence of muscle fibers and replacement by fat and fibrous tissue, the variations in the size of muscle fibers (without neurogenic group atrophy) myofibril disorganization, granular or vacuolated appearence of sarcoplasm and multinucleation were found, than it was decided that the findings were in accordance with muscular dystrophy. In one of our cases, the histopathological changes were consistent with dystrophy and there was not any pathological findings in the other.

DISCUSSION

Pectoralis major muscle agenesis is one of the most frequently found congenital muscle agenesis. Usually it is not complete, and only the sternocostal portion is agenesic. It can occur singularly or as a component of Poland Syndrome (5,6).

Poland Syndrome was first described by Alfred Poland in 1841 (7). It is a rarely-seen congenital absence of the chest wall; it is usully unilateral, the partial or complete absence of pectoral muscle may accompany. In more severe cases, the partial absence of the costae or sternum, renal hypoplasia, leukemias and Mobius syndrome may also be present (7). In our patients, apart from the pectoralis major muscle agenesis, in one case the trapezius muscle was absent, in two cases syndactyly of the hand muscles and in one case bilateral pes cavus deformity was present. No other abnormalities were present.

In the rewieved classification of the Neuromuscular Diseases Research Group of the World Neurology Federation, congenital muscle agenesis is included in the diseases of the anterior horn group; yet it is also noted that since definite indication of the absence of the anterior horn cells is not present, these cases may be classified under the muscle diseases subheading. (8)

Adams et al. (4) report that in same cases with congenital muscle agenesis, Fascio-scapulohumeral type muscular dystrophy (Landouzy-Dejerine) develops in the late period. Apart from these, we could find no publications on the association of pectoral muscle agenesis and myopathy.

Of our cases, the needle EM findings of the one with left pectoralis muscle agenesis and atrophy at the left upper extremity muscles were typical of myopathic involvement. When these findings were put together with the clinical examination, it was concluded that the patient had pectoralis muscle agenesis and Landouzy-Dejerine type muscular dystrophy. The muscle enzymes of the case were normal, and muscle biopsy was not taken (2nd patient - Table I).

In all the other cases, the muscle enzymes were normal. Muscle biopsies were taken from two cases. In one case the histochemical changes were consistent with dystrophic muscle, while no abnormality could be shown with the conventional method in the other case.

As a result, it was concluded that a mild myopathy may accompany muscle agenesis and more advanced neurophysiological investigations must be done in the future.

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