

UNILATERAL ELEVATION OF THE DIAPHRAGM IN DYSTROPHIA MYOTONICA

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The purpose of this publication is to place on record three instances of unilateral elevation of the diaphragm which occurred among a group of 25 patients with muscular dystrophy investigated in New Zealand. Two of these patients had fully developed dystrophia myotonica. The third had a myopathy which clinically was thought to be dystrophia myotonica on account of the distribution of the muscle wasting, electrical myotonia, skull changes, and auricular fibrillation. She died of an acute respiratory infection, and at necropsy affected muscles showed wasting and myopathic changes, and an extensive, round-celled infiltration which was an unusual feature.

CASE REPORTS

CASE 1.—G.L., aged 52 years, a labourer, on admission to hospital stated that his vision had been deteriorating for nine years and that for six years he had had difficulty in walking and a tendency to fall. He had found it difficult to relax his grasp for several years. There was no family history of cataract or of disabilities similar to his own.

On examination he was seen to be emaciated and had bilateral ptosis. He walked with a stick and with a shuffling gait. There was frontal baldness. He had a posterior polar cataract in the left lens. He had a myopathic wasted facies and a small neck with marked wasting of the sternomastoid and other anterior cervical muscles. The forearms and the lower extremities were wasted. There was difficulty in relaxing the grasp in both hands, and mechanical myotonia was present in the tongue and in the thenar and hypothenar muscles of both hands. The upper and lower extremities were weak in all muscle groups. All deep reflexes were absent; the superficial reflexes were normal. In the chest, there was dullness at the right base, impaired air entry and resonance, and fremitus at the same area. The trachea was not displaced. The cardiovascular system was normal. The testicles were very small and soft.

The blood count was normal. The Wassermann and Kahn reactions were negative. An electrocardiograph was normal, apart from occasional auricular extrasystoles and slight elevation of ST2 and ST3. There was thickening of the calvarium and hyper-

ostosis of the frontal and parietal bones. The pituitary fossa was normal.

Radiography of the chest on December 9, 1943, revealed elevation of the right side of the diaphragm which was 8 cm. higher than the left. The right lateral chest film confirmed this abnormality. A radiograph on October 16, 1947, showed the same elevation of the right diaphragm. On March 24, 1948, the right hemidiaphragm was markedly elevated with a very slight respiratory excursion visible on screening. Dr. Klein reported that as far as it was possible to say there was no atelectasis in the right lung, but a thick adhesive band could be seen in the basal area posteriorly. There were also some pleural adhesions at the left base, but otherwise no abnormality of the lungs or heart could be detected. He was admitted to a geriatric hospital in September, 1948, where he developed bronchopneumonia, congestive heart failure, and died.

Necropsy Findings.—The muscles about the neck were wasted and the sternomastoid muscles were completely replaced by fibrous tissue. The testicles were atrophic. There was a small cyst in the anterior lobe of the pituitary gland. There was bilateral bronchopneumonia of the lower lobes. Histological examination of muscles revealed very small and some normal muscle fibres with nuclei arranged in chains up the muscle fibres; unfortunately, the diaphragmatic muscle was not examined. There was a marked increase in the fibrous tissue between the muscle fibres. No cause for the elevation of the right diaphragm was discovered.

CASE 2.—E.W., an unmarried woman aged 40, was treated in 1938 at the Palmerston North Hospital for a partial colonic obstruction. The descending and transverse portions of the colon were removed; the resulting colostomy was closed in February, 1939. In 1951 she was treated at the Wanganui Hospital on account of further subacute obstruction due to faecal impaction, which was relieved by enemata.

On the paternal side of the family there were three members with cataract. There was no other member of the family with myotonia as far as the patient was aware. Three siblings were reported as well.

On examination the patient was a slightly built, poorly nourished woman with a myopathic facies. She had bilateral lenticular opacities and mechanical

myotonia of the tongue. There was wasting of the forearms and reduced power in all muscle groups. There was well-marked myotonia of the grip and mechanical myotonia of the thenar and hypothenar eminences. There was generalized wasting of the legs with weakness and loss of all deep reflexes. The cardiovascular system was normal. There was impaired movement, dullness, and reduced breath sounds over the base of the left lung posteriorly. Radiographs of the chest taken in 1944 and in 1951 showed marked elevation of the diaphragm on the left side. The curve of the diaphragm was not interrupted. The heart was slightly displaced to the right side. No report on fluoroscopy was available. Radiography of the skull showed great thickening of the calvarium, hyperostosis frontalis, and a very small pituitary fossa.

CASE 3.—E.H., on admission aged 62 years, a housewife, when aged 55 fell and fractured the right patella. The fall she attributed to weakness of her legs and a tendency to falling which she had noticed for a year or two. Following this injury the right knee was ankylosed. When aged 57 she noticed increasing weakness of the back muscles so that she was unable to sit up from the supine position without first turning on her side. For six months her arms had been weaker and she had noticed difficulty in holding her head up. When she rode in a tram car the head tended to flop forward when the tram started with a jerk. For two years she had had auricular fibrillation which was well controlled with "digoxin." There was no family history of cataract. One son complained of "cramp" in the upper arm on effort, but routine examination revealed no definite evidence of dystrophia myotonica.

On examination she was well nourished. She had frontal baldness. There were equatorial lenticular opacities in both eyes. There was a patch of old choroidal atrophy below each disc. The sternomastoid muscles were both wasted and very weak, so that she could not elevate her head against gravity when in the lying position. There was no definite local wasting of the upper extremities. The tone was normal, but the power in all muscle groups was reduced. There was an arthrodosis of the right knee joint with marked wasting of the right thigh, and to a lesser extent the right calf. The left lower extremity was hypotonic, and there was wasting of the quadriceps muscle. The power was reduced. She was unable to lift her shoulders off the pillow when in the prone position. The left knee jerk was reduced; the right knee was ankylosed. Other deep reflexes were present and superficial reflexes were normal. There was no loss of sensory appreciation. There was dullness at the right lung base with a reduced resonance and fremitus. The heart was irregular from auricular fibrillation, but this was well controlled.

No myotonia was apparent in the grip or other muscles, and no mechanical myotonia in the thenar or hypothenar muscles, in the tongue, or in the small muscles of the feet. Electrical myotonia was observed in the wasted sternomastoid muscles which responded

to galvanism and faradism but took an abnormal time to relax completely after each contraction.

The cerebrospinal fluid pressure was 120 mm. There was a slight increase of globulin, but otherwise the contents were normal. The Wassermann reaction was negative. Radiography of the chest showed considerable elevation of the right diaphragm. On screening, the diaphragm was found to move poorly on the right side, its excursion being 1 cm. as opposed to 4 cm. on the left side. Radiography of the skull showed marked thickening of the whole calvarium and well-marked hyperostosis of the frontal and parietal bones. The pituitary fossa was normal to small. The falx cerebri was calcified.

On April 24, 1950, she was admitted on account of acute respiratory distress which followed an upper respiratory infection three days before admission. She was breathless, and had a weak, ineffectual cough. Over both bases there was dullness to percussion, and rales were heard throughout the lung fields. There was no consolidation. She was given intramuscular penicillin, the chest signs cleared, and she was discharged. A few weeks later she developed a similar attack in her own home and died.

Necropsy Report.—Muscles of the left forearm showed an intense, widespread cellular infiltration, diffuse and focal, mainly by lymphocytes and fibroblasts. There was great variation in size in the individual muscle fibres, and there were many small fibres. The sternomastoid muscles showed intense infiltration irregularly distributed. In the advanced areas of infiltration muscle fibres were narrow and atrophic. Some fibres were virtually destroyed. In these areas especially there was fibroblastic proliferation. There was very little evidence of migration of muscle nuclei into the substance of the sarcoplasm. The peroneal muscle showed similar atrophic changes, with some well-marked chain formation of nuclei. The diaphragm showed intense cellular infiltration and the muscle fibres were atrophic, with variation in size and an increase of interstitial tissues.

DISCUSSION

Robinson, Mosberg, and Lowe (1950) recorded observations on diaphragmatic movement in some neurological disorders. Among a large group of disorders investigated they studied six cases of myopathy—the type of myopathy not being specified. In all of these there was marked wasting of the shoulder girdles, limb, and trunk muscles, yet no marked abnormality of diaphragmatic movement was noted in any. Sekiya (1940) stated that in "myopathic muscle atrophy the diaphragm is involved late," and Hagemann (1937) recorded free diaphragmatic movement in a child with a very severe muscular dystrophy. Robinson, Mosberg, and Lowe also reported that a woman with myotonia congenita showed no respiratory abnormality, and no satisfactory record of delayed

relaxation of the diaphragm to command was obtained during forced breathing. Thomasen (1948), in his discussion on the localization of the muscular lesions in dystrophia myotonica, states that dystrophy in the abdominal and diaphragmatic muscles adds to the abnormality of the patient's carriage. Difficulties in evacuation and respiration are unknown, but coughing may be of a paralytic nature. This latter statement is not amplified in any way.

Lucien Rouquès (1931) describes one patient with dystrophia myotonica as follows:

"A l'examen radioscopique, l'hémi-diaphragme droit est normal, le gauche est déformé, sa moitié interne est aplatie; le cul-de-sac est presque effacé, non par adhérences, mais par le muscle qui arrive au contact des côtes: la motilité de l'hémi-diaphragme gauche est nettement diminuée, surtout dans sa moitié externe. Aucune lésion su- et sous-diaphragmatique n'étant visible, il est possible que l'aspect du muscle relève d'une certaine amyotrophie."

In the three cases recorded above two undoubtedly had dystrophia myotonica. The third case was diagnosed clinically as dystrophia myotonica. No myotonia was apparent on active contraction or on mechanical stimulation, but it was apparent on electrical stimulation. This, viewed in the light of the distribution of the muscle wasting, the skull changes, and the auricular fibrillation, lends strong support to the diagnosis of dystrophia myotonica. At necropsy there was an extensive round-celled infiltration. This is an unusual finding in dystrophia myotonica. Wohlfart (1951), however, reports that lymphocytic infiltration of muscle may occur in dystrophia myotonica but is very rare. On this evidence we believe that the diagnosis of dystrophia myotonica is established in this case.

All three cases had a unilateral elevation of the diaphragm—two on the right side and one on the left. The terminology used in discussions of elevation of the diaphragm is confusing, and most authors deplore the term eventration. In general, it is used to describe an unusually high position of one half of the diaphragm due not simply to a displacement but to a congenital aplasia or an acquired atrophy of the muscle fibres of half of the diaphragm. The lesion is said to be almost invariably on the left side. On the other hand, diaphragmatic paralysis due to a spinal cord lesion, a phrenic nerve palsy, from compression, avulsion, polyneuritis, poliomyelitis, or other lesion of the nervous system, is reputed to be followed by a rapid replacement of muscle by fibrous tissue, making it difficult to distinguish from the appearance of eventration of congenital origin.

The elevation of the diaphragm in our patients could be due to an eventration, a lesion of anterior horn cells, a phrenic nerve lesion, or a local lesion in the muscle itself.

Eventration is almost always left sided and is considered to be a congenital defect. Two out of three of our patients had right-sided lesions. It is true that congenital lesions are common in dystrophia myotonica in the so-called dystrophic generation, and especially in the offspring of the dystrophic generation. One of us (J.E.C.) has encountered mental defect, cleft palate, harelip, club feet, and spastic quadriplegia. This might be said to support the suggestion of a congenital lesion in our cases.

There was no evidence of poliomyelitis in our patients and none of recent polyneuritis. No signs were detected clinically or at necropsy in two of the patients of a compression lesion or a rupture of the phrenic nerve.

The possibility of a local myotonic reaction within the affected muscle has to be considered. In another patient with dystrophia myotonica with marked myotonia—not one of the three patients here reported—one of us (J.E.C.) was unable to observe on fluoroscopy delayed relaxation of the diaphragm to demand before or after the administration of 2.5 mg. of neostigmine ("prostigmin"), which increases myotonia. As far as we are aware myotonia has never been clearly demonstrated in the diaphragm. Also in our three patients the diaphragm was in a position of relaxation, not contraction, as would be expected if this was the result of a myotonic reaction. In Case 2 the radiographic appearance was the same in 1944 and in 1951, which would be unlikely if this was due to myotonia.

Another possibility is that the affected portion of the diaphragm has become atrophic, as do muscles elsewhere in the body, and has become replaced by fibrous tissue. If this were so, the asymmetrical lesion would be unusual in a disease which is usually symmetrical as far as the myopathy and myotonia is concerned.

The nature of the lesion is therefore obscure, and awaits solution. It is suggested that, as an idiopathic unilateral elevation of the diaphragm has been discovered three times among 25 patients, it may be statistically significant and may come to be recognized as one of the variable features of the disease dystrophia myotonica.

SUMMARY

Three patients out of 25 with dystrophia myotonica had unilateral elevation of the diaphragm.

Reference is made to a fourth case from the literature.

The aetiology of this lesion is obscure.

It is considered that unilateral elevation of the diaphragm may come to be recognized as one of the variable features of dystrophia myotonica.

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