[Muscular Dystrophies and Atrophies]

MEDICAL GROUND ROUNDS
Parkland Memorial Hospital
January 16, 1958

The situation with muscle is at present, similar to that of the holy elephant which had ninety-nine names, the real one being the hundredth, known only to the elephant himself.
Szent-Györgyi

I Case [redacted] - Chronic progressive ophtalmoplegia

The patient is a 79 year old WF who was admitted to [redacted] with bilateral ptosis and opthalmoplegia. She had first noted ptosis four months PTA and then developed diplopia two months later.

Examination revealed a WDWN white female in no apparent distress. Her abnormal physical findings consisted of left and right trochlear nerve palsy, and bilateral partial oculomotor palsy. She had no other neurological abnormalities. Skull x-ray and chest x-ray were negative. Na, K, CO_2, Cl, BUN, Ca, P and alkaline phosphatase were normal. She had a normal blood count. No parasites were found. Repeated Tensilon and prostigmin tests gave minimal nonspecific responses. Muscular biopsy was not performed.

The patient's status has remained unchanged.

II. Case [redacted] Progressive muscular dystrophy-Duchenne type

This 27 year old CM has been admitted to [redacted] several times u.d. progressive muscular dystrophy plus bronchopneumonia.

The patient was first seen here in 1948. His family history was negative. He gave a history of difficulties in walking since 1940. Atrophy of thigh muscles with some hypertrophy of calf musculature developed over the following period. He had only minimal shoulder engagement when first seen. A diagnosis of muscular dystrophy was based on biopsy report: hyalin degeneration and fibrosis of skeletal muscle compatible with progressive muscular dystrophy (49-924).

When the patient was admitted again in 1956, his shoulder musculature showed considerable atrophy. Wasting had occurred in the trunk musculature. There was still good strength in the hands.

Further progression had occurred in 1957, when last admitted. He was unable to lift his head due to sternocleidomastoid atrophy. No involvement of the small muscles of the hands was noted.

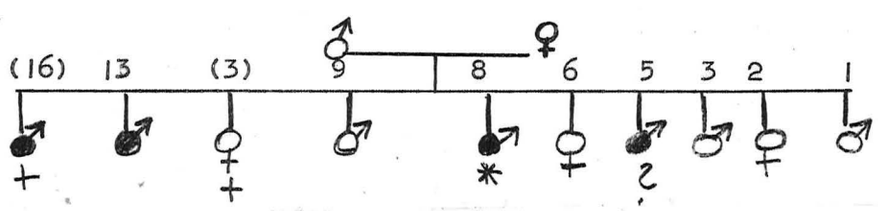
III. [redacted]

Case [redacted] - progressive muscular dystrophy - Duchenne type

This 8 year old CM developed weakness in his shoulders and legs three years ago. It was first noted that he had difficulties in getting up from a sitting position on the floor, and later that he had a waddling gait. The atrophy of the shoulder musculature developed more slowly and has still not incapacitated him.

There is no history of muscle disease in previous generations. The present family is shown below. The boy has marked atrophy of shoulders, hip, and thigh muscles. Considerable hypertrophy of the calves. No fibrillations.

Creatin in blood: 1 mg.%.



* Propositus
● Affected

Number Denotes Age
+Deceased

IV. Case [redacted] - Hereditary proximal spinal muscular atrophy

This 18 year old WM was admitted because of difficulties in walking. His parents were healthy. An older brother (32 years old) is healthy. This brother has a 4-year-old son, who at the age of six months was admitted to a pediatric clinic and treated under the diagnosis of Werdnig-Hoffman's disease, but later the child made a remarkably good recovery. Apart from this child, no other members of the family had been known to have nervous or mental diseases.

In early childhood nothing abnormal had been noticed regarding the movements of the patient. When he was learning to walk, however, his muscles appeared weak. It was difficult for him to stand and walk and keep his balance. He always found it difficult to mount steps and to run. His condition gradually became worse, especially during the last few years. During childhood he was able to raise himself from a forwardly bent posture by supporting his hands against his knees, but now he cannot do so.

On physical examination, no definite signs of muscular atrophy were seen, nor of pseudo-hypertrophy. No definite fasciculations. The distribution of the pareses closely resembled muscular dystrophy with general weakness of the muscles of the trunk and the proximal muscles of the extremities. The gross functional strength of the hands as well as of the lower arms, was somewhat decreased, but the strength of the lower legs and feet was practically normal. He could not raise himself from a bending posture. After bending he was able to get up if he supported himself against a table or the like. He walked with a marked waddle. The muscle reflexes were generally weakened, except the calf reflexes, which were normal. No sensory disturbances.

Moderate creatinuria: 488, 545, and 585 mg./day, respectively.

Electromyography:

a. Gastrocnemius: On insertion and at rest, numerous denervation potentials were seen. Most of the action potentials showed long duration and some of them were markedly polyphasic. There was widespread synchronization.

b. Tibialis anterior: The action potentials were prolonged and of large amplitude. They showed synchronization but there were no denervation potentials.

Muscle biopsy (tibialis anterior dx): Marked disseminated neurogenic atrophy with groups of normal sized muscle fibers and groups of muscle fibers in various stages of atrophy. Some of the normal sized muscle fibers showed a slight immigration of hypolemal nuclei disposed in longitudinal rows. Isolated fibers were in a state of waxy degeneration.

Considerable interstitial connective tissue increase with moderate fat infiltration. Muscle spindles were of normal appearance. One small intramuscular nerve trunk showed a considerable reduction of coarse nerve fibers. No interstitial cellular infiltration.

V. Case [redacted]-Dystrophia myotonica

This 44 year old WF cam originally to the outpatient clinics in 1953 with a complaint of failing vision. She was to have bilateral cataracts and a remarkable frontal baldness was noted. She also was found to have difficulties opening her fists rapidly. There was a suggestion of wasting of circumoral musculature. She had moderate diabetes.

Two years later, she had developed definite weakness of quadriceps and calf musculature. Her musculature status in 1957 was unchanged. She has a family history of frontal baldness and cataracts.

Muscular Dystrophies and Atrophies

Muscular Origin

Without myotonia

Pseudohypertrophic muscular dystrophy (Duchenne;Gowers)
The pelvic girdle atrophic type (Leyden-Möbius)
The juvenile (scapulohumeral) type (Erb)
The facioscapulohumeral type (Landouzy-Dejerine)
The distal type (Gowers)=Myopathia hereditaria distalis tarda (Welander)
The late juvenile type (Nevin)

The congenital myopathy of Turner
Ocular myopathy (Hutchinson, Fuchs, Kiloh, and Nevin)
Menopausal muscular dystrophy (Shy and McEachern)
Benign childhood myopathy (probably polymyositis) (Walton and Nattrass)

With myotonia

Myotonia congenita
Paramyotonia congenita (Eulenbur
Dystrophia myotonia (Steinert, Batten, and Gibb)

Nervous Origin

Central

Poliomyelitis
Amyotropic lateral sclerosis
Hereditary amyotropic lateral sclerosis
Hereditary proximal spinal muscular atrophy
Amyotonia congenita (Oppenheim)
Infantile muscular atrophy Werdnig-Hoffmann)
Syringomyelia
Malignancy
Parietal lobe lesion

Peripheral

Polyneuropathy
Hypertrophic intersitial neuriti
Peroneal muscular
Atrophy (Charcot-Marie-Tooth)

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General reviews:

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Biochemical Disturbances:

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Aldolase:

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Increase in phosphatase (and 5 nucleotidase). Anti-snake venom sera!)

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Different muscles differ in their metabolic needs.

Treatment:

28. Milhorat, A.T. Therapy in muscular dystrophy. Med. Ann. District of Columbia, 23:15-21, 1954.

Tissue Culture:

29. Geiger, R.S. and Garvin, J.S. Pattern of regeneration of muscle from progressive muscular dystrophy patients cultivated in vitro as compared to normal human skeletal muscle. J. Neuropath. Exp. Neurol., 16:532-543, 1957.
Normal skeletal muscle maintained for one year through 12 subcultures. Assembled in groups. Motile.
PMD: Muscle all has shorter latency, more heavily granulated cytoplasm, no crossstriation, and develops, after a month, hypertrophy, increased number of nuclei and granular inclusions. No motility.
Myotonia dystrophica: Muscle cell as in PMD, but with fat droplets and motility. Retaining pathologic properties in subcultures would indicate inherent abnormality, but cells from thyrotoxic myopathy also grow abnormally.
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Myotonia

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The decrease in γ-globulins is due to increased disappearance.
40. Kuhn, E. and Weicker, H. Serumproteine und Lipide bei myotonischer Dystrophie. Schweiz. Med. Wochenschr., 87:460-462, 1957.
A slight decrease in γ-globulin and increase in β-globulins was noted. Lipid pattern unchanged.
41. Landau, W.M. The essential mechanism in myotonia: An electromyographic study. Neurology, 2:369-388, 1952.
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Hereditary aspects:

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Substances and Procedures that Have Been Used in the Treatment of Muscular Dystrophy

NOTE: None of the substances listed has been shown to have any significant lasting effect on the course of the disease.

VITAMINS AND RELATED SUBSTANCES

Vitamin A
Thiamine hydrochloride
Pyridoxine hydrochloride*
Calcium pantothenate
Riboflavin
Nicotinamide
Folic acid
Vitamin B12
Para-aminobenzoid acid
Ascorbic acid*
Vitamin E*
Alpha tocopherol* (natural, synthetic)
Tocopheryl phosphate*
Delta-tocopherol*
Tocopheramine*
Alpha-tocopherylhydroquinone*
Tocopherl + inositol*
Wheat germ*
Wheat germ oil* (cold pressed and extracted)
Corn oil
Liver extracts*
Lecithin
Lyxoflavin
Stigmasterol
Ergostanyl acetate
Soybean sterols
Mixtures of all known vitamins
Dimethylaminoacetate of gluconic acid

ENZYMES AND RELATED COMPOUNDS

Coenzyme A (from liver)
Cytochrome C
Adenosinetriphosphate*(ATP)
Adenosine-3-monophosphate*
Adenosine-5-monophosphate
Procaine adenylate*

PRODUCTS OF GASTROINTESTINAL TRACT

Mucin of hog stomach*
Extracts of hog stomach linings
Extracts of pancreas, pancreatic ferments*
Extracts of duodenum

PROTEINS, AMINO ACIDS AND RELATED SUBSTANCES

Amino acetic acid (glycine)
Glutamic acid*
Arginine
Methionine*
Lysine
Cyteine*
Hydrolysate of liver protein (pancreatic digestio
Gelatine*
Creatine
Glycocyamine
Betaine
Choline
Desiccated liver
Neucleic acid
Sodium desoxyribonucleate

HORMONES AND RELATED SUBSTANCES

Testosterone* (methyltestosterone, testosteron-epropionate)
Insulin*
Epinephrine with pilocarpine*
Ephedrine*
Adrenocorticotropin (ACTH)
Adrenocortical extracts of various types*
Growth hormone of the naterior pituitary
Pituitary thyrotropic hormone
Thyroid extract*

SUGARS AND SUGAR ALCOHOLS

Inositol
Arabinose
Galactose
Lactose
Mannose*
Mannitol
Raffinose

DIETS

Meat-free
High fruit and vegetable*
High protein

REDUCING AGENTS

Methylene blue
Butylated hydroxyanisole

ANTIMICROBIAL AGENTS

Sulfadiazine
Terramycin
Aureomycin

MISCELLANEOUS

Strychnine*
Phenylbutazone
Fever therapy* (malaria)
Resection of nerves of carotid bodies*
Iontophoresis* (calcium)

* Reported to be of therapeutic value, or found, in an occasional patient, to produce mild effects.