A CASE OF MYATONIA CONGENITA (OPENHEIM).

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ALTHOUGH thirteen years have elapsed since the clinical manifestations of this extraordinary disease were first described, the total number of cases on record to date is not large; consequently, we feel that this contribution may be offered without apology.

The patient is a boy of five and a half years, of healthy parents, both of whom sprang from sturdy New England stock. During the pregnancy, which terminated in an instrumental delivery at the seventh month, the child's mother felt what she assumed to be normal fetal movements. Her health was good all the while except for rather persistent and troublesome pains which she attributed to rheumatism. The birth of the patient ended the first pregnancy, and he is the only child. There have been no miscarriages.

The weight at birth was less than three pounds, and the child's head, although well-shaped, was markedly large in proportion to the body. Also, it was noticed that from the moment of birth the head tended to fall backward on the shoulders and that the neck muscles were "like jelly."

The child nursed after the first three or four days, but lay for months in a stereotyped position which strongly suggests the fetal, i.e. with arms folded and the feet so intertwined that the plantar surface of each was in apposition with the opposite tibia. At the outset the muscles generally are said to have been flabby in the extreme, but the disability of the lower extremities was greater than that of the upper and the proximal portion of the limbs was more affected than the distal. If an arm got extended and abducted the child could not voluntarily flex and adduct it, and vice versa. There was no apparent disturbance of intellect or special senses, the breathing was never hampered in any way and the bowel and bladder emptied themselves in normal fashion. Teething did not begin until the eighteenth month, but then progressed rapidly. Prior to this time the child could say "papa, mama," and a few other words.

The case was first seen by one of us in July, 1911.

At that time the child's habit was to lie upon the belly. It could neither sit nor hold the head erect. When it was placed in the sitting position and supported under the arms, the spine showed a very marked kyphoscoliosis and the head fell backward and to the right. There was more or less constant drooling; nevertheless, the intellect was in no wise below par and the patient showed the usual quota of cunning traits peculiar to children at that age. The speech was somewhat difficult of comprehension, but the vocabulary was normal. The head was noticeably large but well proportioned, the fontanelles were closed, the eyes clear and alert, the facial expression mobile. The pupils were equal, regular in outline and prompt in their response to light and distance. The facial and ocular muscles, tongue and

soft palate were normal, likewise deglutition. The tracery of the temporal veins in the somewhat pallid skin was rather striking. The respiratory movement was not restricted; the heart area and sounds were normal, the pulse in no way remarkable. All the tendon reflexes were absolutely unobtainable. The epigastric, belly, cremasteric and plantar reflexes were present but sluggish, in fact, cutaneous sensibility in general seemed rather below normal. There was no rosary and no epiphyseal enlargements.

The most striking feature of the case was the motor impotency of the child and the muscular conditions which underlay it. Everywhere the muscle-bellies seemed thin and flat, but, nevertheless, did not for a moment give the impression of a true atrophy of spinal or neuritic origin. The thinness was most marked throughout the right half of the neck posteriorly and the right side of the back. This accounted for the falling of the head backward and to the right and for the kyphoscoliosis respectively.

Very striking was the poverty of muscle-fibre—and we may add subcutaneous fat—in the arms. The chest and belly muscles seemed fairly adequate. Those of the thigh and leg were even more normal in contour and consistency than the last named, but utterly incapable of contracting voluntarily with any degree of vigor. It is to be regretted that no neuro-muscular electrical tests were made at this time.

The patient was ordered five grains of calcium lactate suspended in milk three times daily, and directly following the visit was taken to the seashore where it had the advantage of out-door life and sea-bathing. The progress made was most satisfactory. At the end of six weeks the child could sit up alone, although the head still fell backward and to the right and the kyphoscoliosis was extreme. At this time, also, the knee- and ankle-jerks were present and the cutaneous reflexes fairly prompt.

During the winter of 1912 codliver oil was steadily given and the dietary and hygienic conditions carefully attended to. The acquisition of muscular strength was constant. Before the end of the winter the child could sit in a chair and cross his legs and, by holding on to his mother's hand, could take a few steps. He gained noticeably in weight and drooled only when very tired.

In March, 1913, the patient developed chicken-pox which was followed by diphtheria. He was ill five weeks and lost ground considerably in consequence, but fortunately escaped a post-diphtheritic paralysis. After this illness he was taken to the sea-shore where he gained to such an extent that in September he could use his hands fairly well, walk about the room unassisted and make his way over the stairs.

At the present time, last of October, 1913, the boy is well-nourished and has good color. He is bright and alert mentally and his articulation is fairly clear. He drools only when very tired. Height 42¾ in., weight 38½ pounds. The head, which still tends to fall backward and to the right, shows the following measurements: circumference 53 cm., ear to ear over sinciput 34 cm., glabella toinion 33 cm. The gait is extraordinary and difficult to describe. Before starting to cross a room the boy stands in an attitude which suggests that of a sprinter on his mark. One arm is thrust directly in front of him, the other behind. The gait is a mixture of ataxia and steppage and, withal, there is in it a suggestion of the peculiar vibratile quality observed in that of multiple sclerosis. The head waggles concomitantly. Once started the patient seems unable to stop until

some obstacle is encountered. Station in the Romberg position is impossible; with the eyes open and the feet apart there is no special swaying, but the knees are markedly hyperextended and the right foot pronated. The pupils are middle-wide, equal, regular in outline and normal to tests of light and distance. The facial and ocular muscles, tongue and soft palate are normal. Fundi healthy. The heart and lungs are normal. Chest circumference 52 cm. The spine shows a lesser degree of kyphoscoliosis. It is the child's wont to keep the fingers flexed in the palm with the thumb underneath, but there is absolutely no spasm or contracture. The finer coördinative movements of fingers and hands are quite impossible. For example, the boy cannot deftly pick up even the plug of a small Faradic battery and insert it in its orifice. In the attempt to perform the latter part of this feat the hand gyrates about the orifice in circles of varying circumference, but the goal is never attained. Meantime the head waggles concomitantly, precisely as it does when walking is attempted. Nevertheless, with solid foods the patient feeds himself fairly well. The musculature of body and limbs has developed fairly satisfactorily both in volume and strength, but the local disproportions noted above are still in evidence. Curiously enough, neuro-muscular response to even mild Faradic stimulation is everywhere equally prompt. The deep and superficial reflexes are all normal.

It requires no special diagnostic acumen to differentiate a typical case of myatonia congenita, like the above, from other conditions which, in a limited number of features, resemble it. The peculiar character of the muscle defect, the fact that this defect is present in its maximum intensity at or shortly after birth, and the further fact that the morbid process is regressive all combine to distinguish this process from whatever underlies all other myopathies and spinal atrophies, and to place upon its symptom-complex the cachet of a genuine clinical entity.

It will be remembered that Oppenheim¹, entirely without the support of pathologic data, believed, at the time he first described the disease in question, that an incomplete genesis of the muscular system was the essential underlying morbid factor, although he did not absolve the anterior horn cells from all connection with this retardation of muscular development. As a matter of fact, actual pathologic investigations, although few in number, seem, in the main, to bear out the Oppenheim theory. We shall refer, however, without prejudice, only to the article of Archangelsky² and Abrikossoff, because their case seems to us clinically typical and, at the same time, we are strongly impressed by the completeness and apparent exactitude of their pathologic findings.

These observers report the case of a female infant who came under the observation of one of them (Archangelsky) at the age of three and one-half months. The child's parents were disturbed by its slow gain in weight (only one pound in three months) and its inability to move the limbs. The child looked anemic; the nutritional state was satisfactory (!); the vertebral column of normal con-

figuration. The thorax was deformed to a marked degree—the sternum protruded sharply, and the chest walls were so sunken that on inspiration the middle portions of the ribs of one side appeared to rub against those of the other. The borders of the lowest ribs were prominent. There was no rosary. The belly was big and flabby, and in contrast to the narrow thorax appeared really enormous. The respiration was clearly diaphragmatic. On inspiration the belly increased in size to a marked degree. The respirations were seventy to the minute and were associated with strong retractions in the region of the insertion-points of the diaphragm.

The most striking feature of the case was the almost absolute immobility of the child. When taken from its bed and placed upon a pillow it assumed a whining expression, while the entire body, extremities and head remained entirely motionless.

The characteristic position of the child was that of a person crucified. The legs were extended, the arms abducted and somewhat flexed at the elbows, the hands resting upon their ulnar borders, with the little fingers bent in such a way that they almost came in contact with the outer side of the corresponding fifth metacarpal bones.

With the child raised in the vertical position the head fell limply and indifferently either forward or backward. Passive motion of the limbs could be carried to almost any extent without apparent pain. Voluntary contraction was limited to one or two muscles and was feeble and restricted in extent. For example, sharp pricking of hands and feet caused but slight movement of the distal portion of these extremities. The child seemed alert to what was going on about it and the excursion of the eyeballs was not limited. The pupils were equal and prompt to light. There was no nystagmus, and the muscles of the face, tongue, soft palate and pharynx were normal. The hearing seemed undisturbed, and the voice was clear but weak. Internal organs healthy. The tendon reflexes were entirely absent, likewise the plantar. The exact status of general sensibility was difficult to determine. While the other muscles were scarcely palpable; both biceps felt like thin, somewhat tense, strings.

The family history was negative, and the child's mother stated that the pregnancy was normal, although throughout the entire second half of it fetal movements were but feebly felt. The birth itself took place at the regular time and was normal in every way. No instruments were used. The weight of the apparently normally-developed newborn was 8½ pounds (3½ kg.). Its exact condition during the first month of life was difficult to determine, although the consensus of opinion of those concerned with its care was to the effect that during this period not even the slight movements of the fingers observed in the second month were present. At the beginning of the second month an electrical examination disclosed a complete lack of response of the muscles everywhere on neck, trunk, and extremities to even a very strong Faradic current.

Toward the end of the third month of life the child began to lose rapidly in weight and the loss continued up to the time of its death, which came suddenly, about the middle of the fourth month, from acute pneumonia. Archangelsky was astonished to observe that in spite of the marked and progressive loss of weight, voluntary motion steadily increased. Only the week before its death the child, lying supine, could move its head sideways to some extent, and could, when supported in a vertical posi-

tion, maintain the head erect for several moments. It was also able to raise the hands to the mouth and, with the knees bent, to abduct and adduct the thighs.

The necropsy, made sixteen hours after death, may well serve as a model for all future investigators. Not an organ, not a shred of tissue that might throw a ray of light on the pathology of myatonia congenita was neglected. Macroscopically this searching investigation revealed only the following abnormalities: (1) Varying degrees of hindered development, pallor and flabbiness of all the voluntary muscles; (2) Pial edema throughout brain and cord (3) Moderate venous engorgement and edema throughout the tissues of cerebrum, cerebellum and cord; (4) Unusual thinness of ventral roots throughout the cord, but most marked in the lumbar region; (5) Catarrhal broncho-pneumonia and (6) Ovarian cysts of varying sizes. The anterior fontanelle was wide open, and all signs of rachitis were lacking.

The detailed description of the microscopical examination is too long to reproduce in its entirety. From the side of the nervous system the findings were, briefly, as follows: (1) A persistence of the remains of the peripheral granular layer of the cerebellum. Incidentally, this may be regarded, in part, as an individual peculiarity not infrequently encountered at the age in question; (2) An incomplete myelination of the pyramidal tracts and Gower's bundle—both structures in which the fibres are not ordinarily fully myelinated until after birth; (3) Absence of the normal nerve-cell grouping in the posterior horns (absence of Clarke's column), with presence in these parts of numerous small multipolar cells; (4) In the lateral horns substitution of many small multipolar cells for the usual large cells; (5) In the anterior horns marked reduction in the number of nerve-cells, with entire absence of large ganglion cells and presence of a few scattered small multipolar cells. Here all nerve-cells appear small, as shown by diminished size of cell-bodies, nuclei and nucleoli. No signs of degenerative changes. The number of nerve-fibres in the gray matter is reduced and the majority of them lack myelin sheaths: (6) In the lateral anterior roots numerous amyelinized axis-cylinders and empty neurilemmata, together with cylinders which are myelinated but thinner than normal; (7) In the median anterior roots nearly complete absence of myelinated fibres, with a peculiar swelling of the amyelinized; (8) In the peripheral nerves a combination of myelinated and amyelinized fibres, with empty neurilemmata. No actual degenerative changes.

In the muscular system the following findings are recorded: "All the voluntary muscles, with the exception of the diaphragmatic, facial, pharyngeal and laryngeal, are very slender in consequence of the extremely marked thinness of the majority of the within-retained muscle-fibres; there are no signs of gradual muscle atrophy, no proliferation of nuclei, interstitial connective or fat tissues."

As Archangelsky and Abrikosoff view the problem, the question to be resolved, as far as the nervous system is concerned, is whether we are dealing with absent or impaired development of the gray substance of the cord, anterior roots and peripheral nerves or with an actual degenerative process in the same. For these authors the absence of the normal nerve-cell grouping of the gray substance, the complete

absence of Clarke's columns, the great number of cells in the substantia Rolandii (suggesting embryonal conditions), further, the presence of very numerous multipolar nerve-cells irregularly distributed in the gray substance of the lateral and posterior horns—all may indicate an interference with the normal distribution and growth of the nerve-cells. On the other hand they point out the fact that the primary development of the ganglion cells of the anterior horns begins very early (before the fourth embryonal week) and hesitate to explain the absence of these cells on the ground of hindered development, because it seems to them highly probable that the hindering process would also exert a deleterious influence upon the development of the horns themselves. They recognize, moreover, the existence of atrophic processes in nerve-cells characterized by diminution in size or even by complete disappearance, without grave change in their structure. This fact complicates the solution of the problem, even though not all the changes found can be attributed to such an atrophic process.

The character of the changes in the nerve-fibres in the gray substance is regarded as of no importance in the solution of the pathologic problem, since these changes could equally well come either from hindered development or from atrophy of the nerve-cells.

Of far greater importance to the above-mentioned authors is the character of the changes in the anterior roots and the peripheral nerves. They note that in spite of the small content of myelinated fibres and of axis-cylinders deprived of their sheaths (lateral groups) and, further, in spite of the almost complete absence of the same (median groups), the mass of roots in its entirety does not seem to be correspondingly diminished but, to a certain degree, retains its normal area. They note, furthermore, that on cross sections of the roots a mesh-like structure is apparent, whose lumina and the arrangement thereof point absolutely to absent nerve fibres. And from this circumstance they conclude that, beyond all doubt, at least a part of the nerve-fibres of the anterior roots have undergone some sort of degenerative process and disappeared, leaving behind only empty nerve-sheaths.

From the above pathologic data Archangelsky and Abrikosoff conclude, finally, that two kinds of changes are present in the spinal cord; 1. A hindered development of its gray substance—as shown by the lessened dimensions of the cells of the lateral and posterior horns, the interference with the customary grouping-process of these cells, the thinness of the myelinated fibres and the axis-cylinders of the anterior roots, the absence of myelination of the majority of the nerve-fibres of the gray substance, anterior roots and peripheral nerves and the embryonal appearance of the substantia Rolandii. 2. Degenerative changes of the cells (simple atrophy)—as shown by the failure, for the most part, of the cells of the anterior horns and

the disappearance of a portion of the nerve-fibres in the anterior roots and peripheral nerves (empty neurilemmata).

The changes in the muscular system are unhesitatingly ascribed by Archangelsky and Abrikosoff to hindered development (hypoplasia) of the muscle-tissue, as evidenced by the uniform thinness of the muscle-fibres—with complete preservation of their structure—and the absence of nuclear proliferation and increase of interstitial tissue.

A point much more difficult for these authors to make clear to their entire satisfaction is the connection between the hindered muscle development and the changes in the spinal cord. They finally conclude, however, that the paralysis and atony of the musculature spring from hindered development of all peripheral motor neurons, but chiefly from a hindered development of the muscle-tissue, which is contemporaneous with that of the cord. The disappearance of the feebly developed anterior horn ganglion cells they regard as secondary and as a result of the consecutive atrophy of these cells.

To the present writers the term—myatonia congenita—seems satisfactory from a clinical standpoint, for the reason that the muscular atony which characterizes it does certainly dominate the picture. On the other hand, the pathology ascribed to it by Oppenheim and subsequent observers impresses us as too narrow in conception.

Its exact pathogenesis must, for the present at least, remain shrouded in obscurity. We venture to suggest, however, a probable correspondence between its pathogenetic factors and those which act upon the primary embryonal tissues in such a way as to bring about spina bifida or hare-lip. Just how early in fetal life the developmental activities are arrested we do not pretend to say. Archangelsky and Abrikosoff, in their case, found muscle-fibres morphologically and structurally identical with those ordinarily observed in the third or fourth fetal month, while our own case has, so far as we know, the distinction of being the first to exhibit the typical picture of the disease at the end of the seventh.

Our reasons for considering the reigning pathologic conception of the disease too narrow are, in part, derived from the microscopic studies of others and, for the rest, from personal clinical observation. In our case, for example, the impaired articulation and the drooling show that the brain-stem, as well as the cord and peripheral nerves, is involved. Furthermore, the marked disturbance of equilibrium observed by us makes it clear that inhibition of sensory neuronal development is also to be reckoned with. This fact alone proves that the theory of hindered development of peripheral motor neurones and muscle-tissue does not cover the whole pathology of the disease. We feel that it may be that the impaired equilibration is due to the absence of Clarke's columns, and we

would urge future investigators to keep this interesting point prominently in mind.

As to prognosis, the reported cases show that the mortality is rather high—about 30%—and that pneumonia is the commonest cause of death. Our case may be classed among the most favorable, but even here complete restitution ad integrum seems to us out of the question, when we consider that the pathology embraces atrophy and disappearance of anterior horn cells and peripheral nerves.

Treatment consists, in the main, in the administration of tonics—we found codliver oil and calcium lactate most serviceable—massage and electricity. Among other important therapeutic measures, warm sea-bathing, generous diet, open-air life and simple coordinative exercises should be mentioned. On general principles, artificial support of the atonic muscles by apparatus should be avoided.

REFERENCES.

- ¹ *Monatsschrift f. Psychiat. u. Neurol.*, 1900, vol. viii, p. 232.
- ² *Archiv f. Kinderheilk.*, 1911, vol. lvi, pp. 101-120.

THE SUSPICION OF MALIGNANCY.*

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NEXT to hemorrhage there is probably no abnormal phenomenon which is to the patient himself more disquieting than the appearance of a tumor. Even with a brave though foolish attempt to disregard the unwelcome presence, and by ignoring to drive it away, there is still the almost subconscious dread that after all we may be facing that ominous, that malignant thing which we know and fear as cancer.

What comes next will depend upon the temperament of the patient; and the human soul is ever individual and never duplicate.

In general, people may be classified as timid or courageous. Yet when confronted with this dreadful spectre of malignancy it is not always the coward who shrinks from the scalpel nor the brave man who seeks the prompt aid of the surgeon. Indeed, the timid are often the first in very panic to seek operative relief, while I have known at least one noble-spirited man, who, with the full understanding of what delay would mean, put off for a year the operation which might have saved him, so that his wife, herself the victim of fatal cancer, should be spared the knowledge of her husband's peril.

The average individual, alarmed at discovering a tumor, will lose little time in consulting his doctor; and often enough his future will hang upon the tactful and conscientious quali-

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