AMYOTONIA CONGENITA: THE RECORD OF A CASE WITH THE ACCOMPANYING CLINICAL FINDINGS[1]

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One other case of amyotonia congenita has been recorded in this country: by Spiller in 1905. All told only twenty-seven cases have been reported. Of these the pathological findings have been reported in two cases, Spiller's being the first.

In 1900 Oppenheim published the first paper calling attention to the clinical findings that characterize this disease of infancy and since then it has borne his name. In the *Neurologisches Centralblatt* of Nov., 1908, Rothmann discusses the disease; and in *Brain* of May, 1908, James Collier and S. A. K. Wilson write at length upon this subject and report cases. I will take the liberty of using some of their data.

Definition.—The latter quote Oppenheim as emphasizing certain essential diagnostic characteristics as follows: "A condition of extreme flaccidity of the muscles associated with an entire loss of deep reflexes, most marked at the time of birth but always showing a tendency to slow and progressive amelioration. There is great weakness but no absolute paralysis of any muscle. The limbs are most affected, the face is almost always exempt. The muscles are small and soft but there is no local muscular wasting. Contractures are prone to occur in the course of time. The faradic excitability in the muscles is lowered and strong faradic stimuli are borne without complaint. No other symptoms indicative of lesion of the nervous system occur."

The case herewith recorded corresponds, in what seem to me to be the essentials, with the above description. On the other hand there are certain points in which it so differs from the other reported cases that a cursory inspection might have placed the

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diagnosis in doubt; but these very points add to the interest of the case. They will be referred to later in detail. In addition to the clinical findings is the report on the blood picture by Dr. Donald Frick, of Los Angeles, which is extremely suggestive and gives new interest to this subject (I can find no other record of blood findings).

Etiology.—Nothing definite has been elicited as to the causative factor in this disease, though Rothmann says it is due to disturbance of the anterior horn cells in fetal life. The testimony has been mostly negative. In none of the cases save this one has there been bad heredity; infectious disease has played no part in the great majority of the cases; trauma, either prenatal or post natal, has not been a factor; frank maternal disease can likewise be dismissed. In a late paper of mine discussing the causative factor of hemiatrophy it was suggested that the "unknown prenatal toxic element" (of some writers on that subject) has had special predilection for the trophic elements of the nerves themselves—following out the theory of de Watteville ("The Nervous System and Its Diseases," Mills, page 195) who holds that "trophic changes in muscle may be independent of paralytic phenomena and may affect both muscles and nerves or muscle alone. If the lesion is confined to the trophic center the muscle atrophies but is not paralyzed and presents qualitative alterations in reaction." It is conceivable that the present condition could be explained by the prenatal action of the "unknown toxin" upon the trophic elements of the nerves themselves causing atony and atrophy of the muscles.

Symptoms.—Pregnancy and birth have been normal in practically all of the recorded cases but the first symptoms have been noted directly after birth in all but five and within the first year in all of these. Many of the children were small and thin at birth while others normal in appearance. In one case it was noted that when the child was held the pendent portions of the body and limbs hung limp like a "bundle of yarn." In our case it was thought to be dead because it hung so motionless (it was not a "blue baby"). In most of the cases there was a flaccid condition of joints and muscles so that the limbs could be swung about like flails. The first symptom to be noted in some of the cases was the inability to hold the head up or, when the time came, to crawl and walk or even, in some cases, to turn over while

lying down. As the result of the amyotonia the most bizarre attitudes could be assumed by the little ones—excessive flexion and extension of hands and feet with wide ranges of rotation of certain joints that could outstrip the feats of the contortionists. In several of the cases there came sooner or later contractions more or less marked and most often at the knee joints. In our case (Fig. 1) there is this contraction with some stiffness of the joints rather than a limpness. In all the cases the facial muscles escaped and the children could suckle. In all but the one here recorded the

FIG. 1.

mentality was good and in this one also it was good up to the sixth month or later, if the parents are to be believed. In all the cases there were two striking conditions noted in addition to the amyotonia, viz., diminished but present response to strong faradic currents and a most remarkable toleration of them. In our case currents were used that could illy be borne by a normal individual and the child seemed to enjoy them.

In most of the cases there was loss of the deep reflexes. In our case they are present; they were even exaggerated during a rise in temperature accompanying an otitis media. In none of

the cases has there been any involvement of the sphincters. Progressive improvement has been the almost universal verdict, but it is so slow that after seven years a normal condition has not been reached in some of the children. As to the distribution of the amyotonia, it is symmetrical and from the neck down. The hands and feet are generally long and narrow (Fig. 1).

Differential Diagnosis.—Hitherto the diagnosis lay between amyotonia and the myopathies but this case of ours makes the diagnosis necessary between it and amaurotic family idiocy. In fact the presence of marked disturbance of vision and mentality in a case such as this is might easily cause that diagnosis to be the first to be thought of. In this child the history points out that she was "born as though dead" but improved later, there was normal mentality up to the sixth month and at that time sufficient cause in this instance—she was the child of an alcoholic and syphilitic father—to produce mental involvement such as she has; there is absence of marasmus; the pathognomonic condition of the eye-grounds is absent; there is no involvement of any other member of her family; she is not a Hebrew by descent. Thus it would seem that the mental defect is accounted for and is an extra condition to the amyotonia.

As to the diagnosis between amyotonia congenita and the myopathies—the former tends to progressive improvement; there may be a return of deep reflexes; there is no local muscular wasting or fibrillary spasm; the electrical reaction is not that of the myopathies; there is the characteristically increased tolerance of faradic currents but no absence of response. There is another condition to be considered—familial amyotonia. In 1902 Beevor published a case with pathological findings in which there was "absolute flaccid paralysis of all the muscles of the body except those of the face, neck and diaphragm. The intercostals were completely inactive. Faradic excitability in the affected muscles was completely lost." Thus, the clinical picture was very like amyotonia congenita. The pathological examination was made by Batten and showed a normal brain, cerebellum and medulla with normal nuclei of the cranial nerves, but with intense atrophy of the anterior horn cells throughout the spinal cord. "There was intense recent degeneration of the fibers of all the posterior columns but the posterior roots were not degenerated. There was great atrophy of all the affected muscles." In Beevor's case

there were four children of a family of eight who were similarly affected. In 1906 Sorgente published two cases, occurring in the same family, under the title of Oppenheim's disease but they evidently, according to Collier and Wilson, belonged to the familial type described by Beevor. They were twenty-seven days and five days old, respectively, at the time they were first observed and they died forty days and twenty days old, respectively. In them there was no response to faradic stimulation. In both cases death was preceded by convulsions that were general in extent.

Pathology.—Spiller published the first pathological report of a case of amyotonia congenita, or, as he expressed it, "myatonia congenita," in 1907. Later in the same year Baudouin reported his findings in the second case in which necropsy was performed. The important points in Spiller's findings are: (1) "The microscopic examination shows that there was an arrest in development of the muscle fibers, and that the central nervous system and peripheral nerves were normal." (2) The muscles have a hyaloid appearance, the fibers are small and differ directly as the muscle tone of the locality from which it was taken. (3) The thymus gland was normal but there was some involvement of the lymphatics and liver. In Baudouin's case there was a like disturbance of the muscle fibers. He noted a sclerotic condition of the thyroid. He found the anterior root of the left third lumbar pair of nerves to be four or five times less in size than its posterior root, whereas in the normal individual it should be about half the size. He also noted some changes in the quality of the anterior horn cells and in the peripheral nerve trunks that suggested to him an arrest of development of nerve fibers.

The case herewith recorded was referred to my service in the Los Angeles Children's Hospital, by Dr. Donald Frick, Physician to the Staff. His examination of the blood picture will be referred to later and is the first blood examination, in these cases, that I know of.

History.—I. L., Female, aged four years (Sept., 1908).

Family History.—Her father is a Mexican and had gonorrhea and syphilis about twenty years ago. He was for many years a drinker of alcoholics in excess; his father is alive and healthy and his mother died of tuberculosis. The patient's mother is a healthy German. She has had no miscarriages and has four other children who are healthy and normal. The patient is her third child.

Previous History.—Quickening was felt. The child was born

at full term without instruments. She was thought to be dead when born because she made no movement or cry; she was not a "blue baby." Up to six months of age her parents considered her normal. At that time she was exposed to a long, hot wagon trip in the sun. It is asserted that after that she was never strong. Soon after it she began to have crying spells and exhibit mental irritation; during the spells she would pound her head and pull her own hair. When about a year old she began to have convulsions. They were always right-sided and began with twitchings of the face and arm. During them there were noted stertorous breathing and a decided curling of the child over to the right side. These recurred quite often during a month's time. At that time, the father says, a "Spanish doctor" prescribed a "blood medicine" for her and her convulsions promptly ceased. The father seems to be clear about her history but due allowance must be given to the fact that the events related happened four years previously. She has never sat up alone, has never talked and has never fed herself. She was not breast-fed as her mother could never suckle any of her children, owing to a lack of milk supply; all the children were fed on condensed milk. Before coming to the hospital she was wont to cry and grit her teeth but since her admittance and previous to an attack of bronchial catarrh and otitis media, she behaved well and seemed contented.

Physical Examination.—(Previous to the onset of the otitis media.)—The child is very small for her age but is symmetrical. She is not wizened or old looking. Her skin is clear and soft. It is evident that she cannot see well or hear acutely. Her hair is copious and the teeth are well shaped and of the normal number. The palatine arch is high. There is some internal strabismus that is not constant. The head is large for the size of the body. She will not hold anything that is placed in her hands. The hands and feet are long and narrow and well shaped. There is some flaring of the lower ribs together with a lateral bulging of the upper abdomen. Her general appearance suggests rachitis.

The muscles all over the body (except the face) show marked lack of tone. They are fairly well used by the child but the joints are stiff, giving a false impression of the muscles. The muscles themselves are soft to the touch, look almost transparent, may be identified. While the child is using one set of muscles the opposing set will be lax and flaccid. She cannot lift her head or hold it up nor can she sit up unaided (Fig. 2). Walking and standing are impossible. There is considerable contracture of the hamstrings (Fig. 1).

The electrical reactions are characteristic. There is response on the part of all the muscles to the faradic current but it is qualitatively lessened. The child can take and seems to enjoy currents that would normally cause much pain. Pain sense is not lacking and she quickly draws away from a pin. Patting and rubbing seem to be especially acceptable to her.

FIG. 2. The child is not held down, but only balanced, to keep her from falling sideways.

FIG. 3. The evident distress of the child was not due to the bending of the foot but to being turned on her face, which she disliked. There were no tears.

Reflexes.—Knee jerks are present on both sides but can be obtained best only when the heels are supported in the hand of an attendant. The skin reflexes are diminished or absent. There is no abdominal reflex. (During the rise of temperature that accompanied the otitis and catarrhal disturbance the child was restless and the knee jerks were exaggerated.) There is no disturbance of function on the part of the sphincter muscles. There is no disturbance of the urinary or alimentary tracts.

Examination of the eyes and ears was made very carefully by Dr. C. H. Montgomery, of Los Angeles, and is as follows: "Lids are normal in shape and structure; they show no abnormal conditions. Lashes are normal in situation and arrangement. Conjunctivæ are somewhat pale but otherwise normal. Scleræ normal. Irides are normal in shape and pigmentation. There is no evidence of inflammatory conditions. Corneæ are perfectly clear and normal. Pupils are active to light and in accommodation. Extrinsic muscles: The right eye shows an internal strabismus of a moderate degree. No muscular paralysis evident and the range of movement is apparently normal in every direction except the one noted. There is no nystagmus.

Fundi.—The eye grounds are of normal color. The discs are paler than normal, the left more so than the right. The edges of the discs are rather sharply marked. The cupping of the disc on the right side is suggestive of an atrophic condition. The blood vessels are not numerous and the branches are less than in the average eye. The maculæ show nothing of note. The retinæ and choroids show nothing suggesting a past inflammation. The vitreous and lenses are normal. Perception of light, peripherally, is markedly below normal.

Ears.—They are normally placed and show no stigmata. The membrana tympani on the right side shows a perforation near the periphery near the bottom; from this comes a sero-purulent discharge (the examination was made at the beginning of the catarrhal and aural disturbance which was not present on admittance to the hospital).

Dr. Frick made a thorough physical examination and reported, in part, as follows: Lungs normal. Heart is normal in size. A systolic blow is heard best in the pulmonic area—probably hemic; other sounds normal. The liver and spleen are not palpable. No tumors are felt in the abdomen. The genitalia are normal. Urine negative.

Blood Examination.—Hemoglobin, 45 per cent.; erythrocytes, 2,600,000; leucocytes, 9,000. Differential count: Polymorphonuclears, 60 per cent.; lymphocytes, 30 per cent.; large mononuclears, 6 per cent.; eosinophiles, 4 per cent.; myelocytes, 2 per cent. Widal Test, negative; malarial organisms, none.

Diagnosis of Blood Condition.—A secondary anemia of infantile type.