Seven Cases of a Unique Familial Illness:
Gait Disorder, Clubfoot, Generalized Areflexia, and Clumsiness of the Hands

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We have observed seven individuals with a familial illness characterized by unique and previously undescribed symptoms. Despite certain similarities, this illness does not belong to any of the previously classified familial maladies. The clubfoot and deep tendon reflex changes are similar to those of Friedreich's ataxia and, perhaps, Charcot-Marie-Tooth disease (although amyotrophy is uncommon in our patients).

In this report we shall examine this rare clinical illness and discuss some of its nosologic and pathogenetic features.

Mrs. Berthe Pli., twenty-five years old, and her two children were evaluated at the Paul-Brousse Hospital in September, 1925, because of a gait disorder. Mrs. Pli.'s mother, maternal grandfather, several brothers and sisters, and other relatives had had a similar disorder. Almost all of the seventeen children of the patient's twice-married paternal grandfather were also affected. In addition, the descendents of four of the female children were known and are included in the following table. One of these children is the mother of our three adult patients as well as the grandmother of four children.


Figure 1. Partial family tree of the Bon family. Affected individuals – ●; healthy individuals – O. The sex is indicated by an arrow or a cross.
Muscle Strength: was equal and satisfactory in the upper extremities except, perhaps, for mild weakness of the fingers of the left hand. All movements were well performed. In threading a needle she had to steady one palm against the other; while sewing, she held the needle with all of her fingers, and tremulously pushed it into the material. This was accomplished with some difficulty. In order to peel a potato, she grasped it and the knife in her hand. While drinking she had a mild unusual tremor, which disappeared at rest. There was weakness of the right toe extensors and the left anterior compartment muscles; thigh extension was diminished bilaterally; flexion was normal, and all other leg muscles were of equal bulk and of normal strength. Head and trunk flexion and extension were weak—flexion more so. Lateral neck movements were intact.

Reflexes: including the abdominal reflexes were absent. The left plantar response was inconstantly extensor, the right flexor; but unequivocal extension of either toe was not observed.

Cerebellar testing: including finger-to-nose, rapid alternating movements, (marionettes), hell-to-knee, and heel-to-buttock maneuvers were intact.

Tone: was somewhat decreased. Postural reflexes were intact; but the left anterior tibial reflex was equivocal.

Facial exam: revealed mild asymmetry and enophthalmos. The right palpebral fissure was narrowed. The lids were lowered but not really ptotic. The left side of the face moved more than the right when the patient laughed. The cranial nerves were otherwise unremarkable. The masseters contracted equally well, the patient could purse her lips to blow or whistle with ease; the corneal reflexes were present but diminished; the palatal reflex was normal, but the pharyngeal reflex was absent.

Sensation: Subjective sensation was normal. There was no dysesthesia. Objective sensation was normal (touch, pin, heat, cold, position, stereognosis), and, except for atrophy of the thenar eminences (most marked on the right) there were no trophic abnormalities.

Electrical exam (Miss de Brancas): revealed slight faradic and galvanic hypoexcitability of the small muscles of the arms and legs as well as the muscles surrounding the popliteal fossa that are innervated by the sciatic nerve.

Ophthalmologic exam (Dr. Bollack): revealed normal pupils, normal ocular motility without nystagmus, and normal fundi. The visual fields were normal; visual acuity: Right 7/10–0.10, Left 5/10–0.10.
Hearing, speech, and writing were normal, and there were no sphincter disturbances. She had had enuresis until the age of twelve and thereafter continued to be incontinent when she laughed.

The serum Wassermann reaction was positive.

Summary: This twenty-five-year-old woman presented with “giving way” of her legs, awkwardness of hands, generalized areflexia, club-feet, and a mildly unsteady steppage gait. Except for mild intention tremor of her hands there were no cerebellar findings. The toes were flexor, and certain muscle groups were minimally weak. There were no sensory disturbances and no nystagmus; but the responses to electrical stimuli were abnormal.

Case 2

Simone Pli., the seven-one-half-year-old daughter of Case 1, presented with a gait disorder. The disorder had improved but was still present. She was the product of a normal-term pregnancy, weighed 8 pounds at birth and cried immediately. She was breast-fed until one year of age, cut her first teeth at eight months, spoke at one-and-one-half years, but began walking at three years, at the same time as a younger brother. She had attempted to walk at two years of age but couldn’t. She fell and continued to crawl on all fours. This condition improved slowly. She reached her current level of performance at about five years of age. Although she appeared to be very intelligent and could recite a fable perfectly, her school performance was poor. She had had whooping cough at age four and roseola recently.

On examination she was a 17-kg., pockmarked, well-formed child with genu varum but without scoliosis. Her feet were bent but normal in size. She easily stood, with both feet together and without disequilibrium, but had difficulty standing on one foot. Eye closure caused her body to oscillate. She wavered when she walked, supporting herself on her heels which she dragged along the floor. She fell easily. She lay down and stood up without difficulty. We observed that the patient had difficulty with fine finger movements. The patient’s mother had not noticed this. There was no tremor, and muscle tone seemed normal; postural reflexes were intact. Her upper and lower extremities were very strong, although her thigh extendors and flexors were weak (the extensors more than the flexors). Head and trunk flexion were weak but extension was normal. Lateral head movements were also well performed. The deep tendon and upper abdominal reflexes were absent, whereas the lower abdominal reflexes were intact. Both plantar responses were inconstantly and equivocally flexor.

Finger-to-nose cerebellar testing was well performed with a mild tremor, more marked on the right. Rapid alternating hand movements such as pronation-supination of the hands on the thighs were poorly performed. These movements were difficult when performed rapidly. The heel-knee-shin test was done well but there was some difficulty, possibly due to inattention, in touching the knee. The heel-to-buttock test was well performed.

The face appeared normal and was symmetric. No cranial nerve abnormalities were observed. The facial nerve was normal; she blew well. The corneal responses were normal and the pupils equal and reactive to light. Sensation was subjectively and objectively normal, and no trophic abnormalities were noted. There were no sphincter disturbances, but the patient was enuretic and suffered from incontinence. Electrical examination revealed changes similar to those of her mother.

Ophthalmologic examination (Dr. Bollack) revealed normal pupils and motility with small horizontal nystagmoid jerks on right lateral gaze. The fundus was normal and acuity 2/10 bilaterally.

Speech and hearing were normal. She wrote slowly and tremulously with the pen grasped in her hand.

The blood Wassermann test was faintly positive.

Summary: This was a seven-and-one-half-year-old patient with a gait disturbance dating from the time she began walking at three years of age. During the ensuing years the gait difficulty had improved. The patient had a clubfoot, absent deep tendon reflexes, and a mild intention tremor; but despite these signs, there were no obvious cerebellar, sensory, or atrophic disturbances.

Case 3

Raymon Pli., the two-year-old brother of the preceding case presented with a tabetic-like gait, a right clubfoot deformity, and a tendency to fall when unsupported.

The patient’s birth followed a full-term pregnancy and uncomplicated delivery. He cried immediately and weighed 9 pounds. He was breast-fed until thirteen months of age, cut his first teeth at seven months and spoke at a year. When he learned to walk at eighteen months, it was noted that his legs bent as soon as he stood on them. He had recently recovered from measles.

On examination he was a normal-looking child with rachitic bulges on the wrists and malleoli, and flaccid, perhaps atrophic, calves. The lumbar lordosis was exaggerated, and the right foot was slightly flat. His gait was unsteady and quite abnormal: he dragged his heels, and threw out his feet as a tabetic would, after which he staggered and fell. He stood with his feet spread apart, swayed, and appeared un-
The patient (Case 3) could only stand for several seconds without support if his legs were spread widely apart.

stable. He easily arose from a prone to a standing position, which he maintained unsteadily. On arising he did not rest his hands on his thighs as a dystrophic might.

It was impossible to test muscle strength, but all movements— including those of the feet, toes, and legs—were well performed, and muscle tone seemed absolutely normal.

The deep tendon reflexes were absent, the superior and inferior abdominal reflexes were present bilaterally, and the plantar response was equivocal on the right and not elicited on the left.

The cerebellar examination was difficult because of the child's age but appeared normal with the possible exception of finger-to-nose testing on the left. Sensation could not be adequately evaluated. There was no atrophy or sphincter disturbance, but the child had been enuretic. The eyes and fundi, examined by Dr. Bollack, were normal. Extraocular movements were normal and without nystagmus. Speech was normal.

Summary: the patient had a clear-cut gait disorder and generalized areflexia without clubfoot or dorsiflexion of the great toe. There were no obvious cerebellar difficulties in this intelligent two-year-old with normal speech and without amyotrophy.

Case 4

Miss Julia Ur., a thirty-two-year-old factory worker and sister of Case 1 presented with bilateral painful clubfoot, which had resulted in a marked gait disturbance. She had always had foot deformities and a gait disorder and recalled falling often as a child. Her past medical history was otherwise unremarkable. Her birth was uneventful, and it was not known when she began to walk. She seemed of normal intelligence and attended school through age thirteen, after which she was employed. Her first husband was tuberculosis. A normal-appearing child, the product of this marriage, died of asphyxia one hour after birth. Her second marriage to a healthy man produced no children, no miscarriages.

On examination her gait seemed normal at first, although complicated by her foot deformity (especially on the right). The foot appeared foreshortened, squared off with a marked plantar curvature. The toes appeared malpositioned. Their distal portion was on a level with the sole of the extraordinarily short foot. The patient wore a size (33) shoe.

Standing erect, the patient tended to hold her right leg abducted and externally rotated. No atrophy was observed. The calves were large. The right calf was slightly deformed. Its fleshest portion lay antero-laterally and was located immediately under the knee.

The patient spoke and wrote well, and her face and hands were normal, although she complained that exercise brought on cramps in her hands. Her facial muscles contracted well on both sides. The pharyngeal, palatal, masseter, and corneal reflexes were normal. Hearing was intact.

Visual acuity had been impaired since the age of two. Dr. Bollack's ophthalmologic examination revealed acuity OU of 2/10 + 0.75 and nor-
mal fundi (with bilateral congenitally small, inferiorly placed discs). There was no nystagmus, and ocular motility was normal.

Muscle strength was normal and equal in all four extremities. Toe movements were limited by the malposition of the toe in the foot as previously noted; only the great toe moved normally. There was generalized deep tendon areflexia, and the cutaneous abdominal reflexes were abolished. The plantar reflexes were flexor bilaterally, and the sphincters were intact. Muscle tone and bulk seemed normal; no hypertonia or hypotonia was observed during passive movement. Cerebellar testing of the upper and lower extremities was normal. Although the patient complained of cramps while walking, sensation was intact to pin, temperature, position, and stereognosis.

Blood serology was slightly positive.

**Summary:** the patient had a marked clubfoot and generalized tendon areflexia but no cerebellar, pyramidal, or sensory disturbances. In addition, there was no nystagmus, speech disorder, or atrophy. Her difficulty walking was attributed to cramps and to her foot deformity. She also suffered from cramps in the hands after exertion.

**Case 5**

**Summary:** Mr. Bonne, a turner, refused to be examined. He was thirty-eight years old and the oldest brother of the two preceding cases. He stated he was normal and gave the appearance of a remarkably robust, muscular, and athletic man.

He had done his military service “without ever missing a march.” His face was clearly abnormal—with mild bilateral exophthalmus and a flat nose (probably the result of old trauma). His feet were small and clubbed, similar to those of his most affected sister. He wore a size 39 shoe; was 1 meter, 20 cm. [sic] tall and weighed 75 kg. We were able to examine his deep tendon reflexes, which were all absent. He had been married twice. A daughter (Case 6) by his first wife, and one of his two sons by his second wife (Case 7) presented similar signs. He tended to sway when standing. This was especially true when his eyes were closed. He had no difficulties using his hands.

**Case 6**

Andrée B., the seventeen-year-old daughter of Case 5 and half-sister of Case 7, presented with a gait disorder and awkward hand movements (especially fine movements). Her birth and childhood history were not available. It is known that she walked at twenty-two months and had a right otitis. She could not read or write, having attended school for only fifteen days. If one took into account her cultural deprivation, her
speech and intelligence could be considered normal. She had the same nasal deformity as her father (she had fallen on her nose), bilateral clubfoot, as well as very suspicious teeth [suspicious of congenital lues].

Her legs were pockmarked. There was marked genu varum. Her shoe size was 35 (quite small). Her gait had a wavering quality. When she stood she had the same difficulties as her father. This unsteadiness increased when she closed her eyes. Her hands were unequivocally weak, and hand movements were awkward. When she laced her shoes she held the lace between her thumb and the back of the proximal portion of her index finger (as do the patients with Charcot-Marie-Tooth disease). When she drank from a glass she was tremulous; she had great difficulty picking up coins from a table. Extremity tone and strength were preserved. Her deep tendon and cutaneous abdominal reflexes were absent, but sphincter function was preserved. The plantar reflex was flexor on the left and equivocal on the right. Standard cerebellar testing was normal, and the face was unremarkable except as noted above. A small corneal opacity was present on the right. The pupils were normal and equally reactive to light, and there was no nystagmus. Sensation was intact. There was thenar, hypothenar, and interosseous (especially on the left) atrophy.

Figure 7. The brother of the two adult patients (Case 5). Note his slender lower legs and foot deformities.

Figure 8. Foot, detail (Case 5). His shoe size was 39 (unusual for a man).

Figure 9. Slender legs with clubfeet (Case 6).
severe gait disorder, and he fell frequently. As result of these falls he re-fractured his right tibia, which had been broken before in a mishap with a bicyclist. His birth was normal at term. He was breast-fed, spoke at one year and walked at three years of age. He was considered to be very intelligent. He had had measles, mumps and gastrointestinal illnesses. On examination he appeared to be intelligent and quick to learn. He had normal speech and an unsteady gait. When he walked he seemed to throw his feet forward. Standing with his hands clasped together, he swayed and fell when he was pushed. The swaying increased when he closed his eyes. He could not stand on one leg and ran only with difficulty. At rest his legs gave way, as did those of other members of his family. His feet seemed of normal length. His left foot was slender and his right foot was rather flat—possibly a result of trauma. Muscle strength and tone were normal, but the deep tendon reflexes were absent. The cutaneous abdominal reflexes were preserved, and the plantar responses were equivocal on the right and flexor on the left. There were no cerebellar abnormalities, except for a mild intention tremor of the left arm. The face was unremarkable except as noted; the pupils contracted well, and there was no nystagmus. Although the child complained of foot cramps after walking, his sensory examination was normal (temperature testing was not performed).

The last three cases refused ophthalmologic, electric, and serologic examination.

Summary: this nine-year-old child presented with a disturbance of gait and station, an equivocal left clubfoot, generalized tendon areflexia, and a mild left-sided intention tremor.

Thus, we have had the opportunity to analyze seven cases of a familial illness involving twenty individuals in four generations. In order of frequency, the features of this illness are: a. Disorder of station and gait; b. Generalized areflexia; c. Clubfoot. In addition, certain members show: d. Awkward hand movements; e. Palmar muscular atrophy (rare); f. Absent cutaneous abdominal reflexes with extension of toe (suggesting an extensor plantar response); g. Sphincter weakness (enuresis and stress incontinence). We will discuss the positive and negative features of this illness in detail.

POSITIVE FEATURES

Disorder of station and gait: The gait disturbance seems to be due to mild ataxia as well as cramps or pains associated with the foot deformities. The ataxic gait has an inebriated, steppage, swaying quality. The patient walks with his legs thrown forward, but the motions are not as brusque, nor do they have the great lurching to-and-fro quality seen in
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<th>Berthe Pli. 25 years Case 1</th>
<th>Simone P. 7½ years Case 2</th>
<th>Raymond 2 years Case 3</th>
<th>Julia U. 32 years Case 4</th>
<th>Mr. Bonne 38 years Case 5</th>
<th>Andrée B. 17 years Case 6</th>
<th>Robert B. 9 years Case 7</th>
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cases of Friedreich’s ataxia. Also, unlike Friedreich’s ataxia, our patients become less ataxic as they grow older. Our most affected patient (Case 3) was also our youngest. Case 2 (age 7½), who showed only mild difficulty when examined, had been as disabled as her brother (Case 3) when she was younger. Their mother’s difficulty showed the same pattern of evolution.

As we shall see later, the ataxia is most apparent on standing. Paradoxically, traditional tests of coordination do not reveal any abnormalities.

The episodic leg and foot cramps and pains occur after walking or standing. As in Case 4, these symptoms alone may be severe enough to make walking difficult or hesitant. These cramps may be associated with incoordination (Case 6).

The illness may present a range of abnormalities from ataxia without foot deformity (Case 3) to painful clubfoot without ataxia (Case 4), to clubfoot without gait disturbance (Case 5 is unique in this regard). The station difficulties reflect equilibrium problems as well as a tendency for the legs to quickly collapse. The disequilibrium is not completely explained by coordination difficulties. In the uncomplicated condition, when the patient is standing, disequilibrium shows itself as small-amplitude antero-posterior or transverse oscillations of the whole body (Cases 4 and 5). The unequivocal clubfoot and concomitant neurological symptoms seen in both patients may be responsible for the unsteadiness while standing and the inability to stand on one leg. However, these deformities would not explain the presence of Romberg’s sign (increased unsteadiness with eyes closed) noted in most of our patients. It is also impossible to relate the patients’ need to stand with legs spread and their titubation to the clubfoot deformity (a deformity clearly present in Case 6 but not in Case 3). One approach to the problem is to consider our patients’ sudden lapses in leg control. These brusque, involuntary flexion movements of the legs sometimes associated with traumatic falls in our patients are also seen in tabetic. These episodic lapses seem to be of motor origin, are not provoked by pain, but may be preceded by activity (such as carrying a burden).

**Generalized tendon areflexia:** The second most constant symptom is complete absence of tendon reflexes in the upper (radial, brachial, and triceps) and lower (knee and ankle) extremities. Jendrassik’s maneuver had no effect. In some patients the idiomuscular contractions in the posterior leg seemed feeble.

**Clubfoot:** A classic, bilateral clubfoot deformity was seen in four cases (Cases 1, 4, 5, and 6). In three others it was either unilateral or atypical. The classic deformity consists of antero-posterior compression, foreshortening, and widening of the foot (Case 4), sometimes associated with extension of the first phalanx of the great toe and flexion of the second phalanx without abnormalities of the remaining toes. In addition, the plantar arch, viewed from the lateral border of the foot, is exaggerated, and the foot seems to jut out dorsally. In Case 4 the deformity was so severe that the distal phalanges of the toes were on a level with the sole of the foot. X rays show abnormal size and position of the bones of the foot without abnormalities in their form or number. In less typical examples of the clubfoot there is (sometimes unilaterally) dorsal protrusion and excessively convex plantar arching without appreciable foreshortening. In addition, certain patients have slender lower legs, in contrast to the other calf (Case 5) and thigh muscles (Case 6), together with a genu varum (Case 6).

**Secondary Symptoms**

In addition to the above three characteristic findings there are less constant ones listed here in order of frequency:

1. **Toe extension (most often unilateral) in response to painful stimuli. In no case, however, was a clearly defined Babinski response present.**
2. **Frequent absence or diminution of the cutaneous abdominal reflexes.**
3. **Slight awkwardness of the hands during activities (sewing, peeling, buttoning, picking up coins) which involve finger movements. These activities were associated with a slight, atypical tremor in Case 1. Writing remains normal but is slow. In Case 2 it was associated with tremor and contraction of the digits. Marked weakness and movements usually associated with the Charcot-Marie-Tooth disease were present in Case 6. In several cases weakness could be demonstrated objectively in the hands or proximal lower extremities.**
4. **Atrophy and weakness of the thenar, hypothenar, and interosseus muscles were observed in one case. This combination of atrophy and weakness will be considered later when we discuss pathogenesis. The faradic and galvanic hypoexcitability of the muscles of the distal upper extremities is consistent with the theory of pathogenesis I will propose.**
5. **Sphincter dysfunction (enuresis and stress incontinence) were present in some of our patients.**

**NEGATIVE FEATURES**

The illness is defined both by positive and negative findings. Our patients do not exhibit appreciable cerebellar dysfunction (with standard
tests), objective or subjective sensory abnormalities, significant amyotrophy or scoliosis. Moreover, there is no evidence of cranial nerve dysfunction, nystagmus (with one mild exception), speech impairment, or intellectual decline.

EVOLUTION OF THE ILLNESS

The evolution of this illness is as mysterious as its symptoms. The child’s first attempts to walk are delayed and difficult. As the foot deformities are probably congenital, it is probable that the major symptoms are also present from birth. This impression is supported by the fixed or even regressive nature of the neurological deficit. We are uncertain as to whether other, more seriously affected family members had a progressive form of this illness, since we were unable to examine them. Furthermore, we did not have exact information concerning previous generations. Our oldest patient was thirty-eight years of age. His deficit was so minimal that he would have been ascended to have been included among the afflicted family members. In fact, he was remarkably robust and appeared practically normal. The other affected adults did not appear to have suffered from any progression of their disease since its onset. Some seemed to have improved!

FAMILIAL EVOLUTION

The precise mode of inheritance of this illness is unknown. The process affects and is transmitted by both sexes. There is incomplete penetrance since certain patients have unaffected children, but we have been unable to prove that the disease is transmitted by otherwise healthy individuals.

DIAGNOSIS

In view of what we have already said about the illness, how is it to be classified, and what is its possible etiology?

DIFFERENTIAL DIAGNOSIS

The specific symptoms associated with the illness include a disorder of gait, clubfoot, and absent reflexes. These symptoms in combination with the familial character suggest a theoretical link to Friedreich’s ataxia. However, the patients’ symptoms and the mode of appearance and evolution of the illness distinguish it from Friedreich’s ataxia. The latter is characterized by obvious and predominant cerebellar symptomatic, difficulty speaking, nystagmus, choreiform instability, clubfoot, and spontaneous extension of the great toe. It appears most often in late childhood or adolescence and follows a progressive course.

By contrast, cerebellar symptoms were minimal in our patients and not related to the onset of the illness. As we shall mention later, even after thorough examination we were not able to make a definite statement regarding cerebellar symptoms. Our patients did not have choreiform instability, difficulty speaking, nystagmus, or scoliosis. If one disregards the fact that our patients became symptomatic first in early childhood, do not seem to progress, and do not exhibit tonic extension, they are linked to patients with Friedreich’s ataxia by the presence of clubfoot and the absence of deep tendon reflexes.

If our cases are not typical of classic Friedreich’s ataxia, could they be atypical forms of that illness? They might be attenuated, regressive, or possibly latent forms of Friedreich’s ataxia. In truth, this hypothesis is not satisfying, nor does it resolve anything. No other published instances of forme fruste Friedreich’s ataxia appear similar to ours. In this regard one fact is bothersome: Gardner* reported one family in which the mother suffered from spastic paraplegia, intention tremor, nystagmus, clubfoot, etc.; three of the six children were affected, and three were considered “normal.” All three “normal” children had absent knee jerks, one had scoliosis, and one had scoliosis and a clubfoot. Gardner considered that this reflected the multifaceted presentations of nervous-system illness. Thus, the general features (of these illnesses) permit their separation into large groups, while the secondary features separate the illnesses within these large groups. Our cases may represent an intermediate or transitional form of familial illness linking Friedreich’s ataxia and Charcot-Marie-Tooth disease. Some of our cases seemed closer to the latter by virtue of awkward hand motions, atrophic features, electrical abnormalities, and the presence of clubfoot. In truth, this separation may be rather fanciful and should not replace the anatomo-clinical data.

Despite the lack of anatomic verification we feel that the clinical picture is so distinctive as to allow us to separate this illness from others and describe it in this report. Our ignorance of the cause and pathology of this illness leads us to several hypotheses.

HYPOTHESES CONCERNING THE NATURE OF THE ILLNESS

From an anatomo-physiologic point of view, how might we view the causal lesions in our patients? A lesion of the posterior columns is

suggested by the absence of deep tendon reflexes and the mild incoordination observed in all patients. We are aware that this is the clinical picture associated with cases of familial tabes (two brothers or sisters as reported by Londe or Crouzon). This incoordination could also be explained by a lesion of the cerebellar tracts as in Friedrich's ataxia.

The clinical picture leads only to hypotheses.

A posterior column rather than cerebellar tract lesion is suggested by the absence of dysmetria, true intention tremor, obvious cerebellar signs, and increased unsteadiness with eye closure.

However, the very mild pyramidal signs exhibited by some of our patients suggest an involvement of the lateral columns, such as is seen in Friedrich's ataxia.

Yet again, these signs may be due to lesions of the anterior horn cells. Such lesions would also explain the mild atrophic changes and above-mentioned electrical abnormalities. If one adds to these findings the involvement of the posterior columns, our cases resemble those with Charcot-Marie-Tooth disease.

The nature of the lesions in our patients eludes us. It would be easy to attribute this illness to hereditary syphilis, especially in view of the inadequate serologic examinations in our cases; but this would not explain the familial occurrence of the illness. This familial pattern is a general feature of the illness. None of our patients accepted a lumbar puncture, and only three had a serological examination. These serologies (two sisters and the child of one of them) were positive, which suggests an acquired illness rather than hereditary transmission. The criticisms are applicable to Cases 6 and 7, who had facial and dental deformities suggestive of hereditary syphilis. These two children did not have the same mother; and the father, who transmitted the familial illness, had somewhat suspect facies. As for other infections, none of our patients with the inherited illness became symptomatic after an acute infection. Moreover, we were unable to uncover any personal or genealogic fact which might explain the mysterious etiology of this malady. The problem of etiology is one which concerns all of the familial illnesses and, in a general way, reflects their hereditary nature.

CONCLUSIONS

1. We have described a unique familial illness (without pathologic verification) characterized by difficulty walking and standing, clubfoot, generalized tendon areflexia, and occasional mild clumsiness of the hands. Its clinical features permit us to separate it from the other familial illnesses.

2. We do not feel that this illness represents either an atypical form of Friedrich's ataxia or an intermediate form of other familial diseases. It must be pointed out that this last statement is theoretical and does not lead to a definite classification. The two hypotheses complicate rather than elucidate, and raise the possibility of a unified pathogenesis for all the familial illnesses.

3. Even if future anatomic studies allow us to more clearly unite the various previously described forms of familial illness, the process we have described remains unique, in need of explanation, and worthy of our report. This report describes a separate and apparently new illness with common symptoms and a familial pattern, which makes it of some importance.

Even if this illness is ultimately proven to be an aberrant or transitional form of one of the already described familial maladies, the nature of the evolution, variety, and interrelationships of the familial illnesses will still represent a remarkable area for future research. Thus, we did not think that it was inappropriate to present this long, imperfect, and provisional description.