ORTHOPAEDIC PROBLEMS OF HEREDITARY SENSORY NEUROPATHY

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Petrie (1953) described a patient with multiple arthropathy from congenital indifference to pain. Cram (1953) reviewed neuropathic derangement of joints associated with diabetes mellitus. We have studied the family reported by Heller and Robb (1955), who traced the disorder through six generations, with evidence of hereditary sensory neuropathy in five patients. Three other members of the family showed an incomplete form of the disease, one also having an affection of the spinal cord, probably from multiple sclerosis. Price (1913), Schultze (1917), Hicks (1922), Ogryzlo (1946) and Denny-Brown (1951) reported cases. Denny-Brown showed histologically that the fundamental pathology was in the dorsal root ganglia. There was demyelination of the afferent fibres and a paucity of cells in the ganglia. Around these, a satellite proliferation of Schwann cells could be noted, small nodules being formed. Within these cellular nodules were dendritic proliferations. This was described as the essential pathology in hereditary sensory neuropathy.

Each of the following cases shows 1) severe trophic disorders of all extremities; 2) loss of all sensibility in the extremities, in a glove-and-stocking distribution; and 3) loss of deep tendon reflexes. The peripheral pulses, sweating, temperature and other manifestations of sympathetic function were normal.

CASE REPORTS

Case 1—A man of twenty recalled that when he was about five years of age he had difficulty, in walking, from tripping over small objects. He also had difficulty in buttoning his shirt because of a lack of feeling in his fingers. At the age of nine he scratched the toes of both feet while swimming, and the scratches took a long time to heal. Later the same year he experienced a dull burning sensation in his feet, unrelated to activity. An operation of unknown nature was performed on both feet, and he was in plaster for six months. Thereafter he enjoyed good health until he reached the age of twelve, when he noticed that his nails began to crack, particularly on the second and third right fingers. Pus accumulated painlessly under the nail, and the condition progressed until the nails and a small piece of bone were removed from the second and third phalanges. The wounds healed well.

In that same year he suffered recurrent infection of the dorsum of the left foot. Later he underwent incision, drainage and sequestrectomy of the lower third of the left tibia. Radiographs at that time showed marked degenerative changes of the foot. The onset of the various febrile episodes is of interest. He first noticed a cough, with a feeling of discomfort over the lower retrosternal area. This coughing would be followed by malaise, sweating, chills and fever. His oral temperature would reach 103–104 degrees Fahrenheit. So long as the pyrexia continued, the cough persisted. This would last for several days, the patient remaining in bed. Lysis of the temperature was associated with a feeling of well-being. No marked pain other than a dull widespread ache would occur in the extremity, but an area of infection was always found on examination.

At the age of fifteen years the tibial osteomyelitis had progressed, and a below-knee amputation was carried out. The stump healed slowly, and when he began to use a prosthesis the stump became ulcerated. The records of the following years reveal ten admissions to hospital for various procedures for the infected stump and for osteomyelitis of the right foot.

For the past two years there has been a persistent sinus in the foot, necessitating excision of the third and fourth metatarsals. The fingers have become progressively more ulcerated and several sequestra have been discharged. At present there is a large ulcer on the sole of the foot.

The whole life of this patient has been governed by these recurrent episodes of infection. As each occurs, he remains at home and elevates the part until healing occurs.

On examination the head, thorax, abdomen, shoulders, elbows and hips were normal. A photograph of the patient is shown in Figure 1. The hands showed thickened skin over the palmar and dorsal surfaces of the fingers (Fig. 2), the distal phalanges of which were missing. Ulcers were scattered throughout the fingers. The skin of the left popliteal fossa was thickened and the whole stump was irregular, the skin being of variable roughness. There was a discharging sinus at the anterior surface of the stump. The right foot was in slight valgus; there was only a small range of movement at the ankle and subtalar joints. The dorsum of the foot was reddened, indurated and warm and showed the scars of previous sinuses and operations. The sole of the foot was ulcerated (Fig. 3).

Neurological examination showed normal cranial nerves, muscle groups and co-ordination. All superficial and deep reflexes were absent. The sense of touch and pain was absent below the knee and over the forearms and hands, with hypaesthesia fading to the mid-thigh level and above the elbow. Temperature sensation was absent below the lower forearms and below the knees. Position sense was absent in the toes of the right foot and diminished in both

FIG. 1

Case 1. Photograph of the patient, showing lesions on fingers and deformity of right foot and ankle.

FIG. 2

FIG. 3

Case 1. Figure 2 Right hand showing shortened fingers, ulceration, and thickened creases. Figure 3 Right foot, showing a large ulcer on the sole.

little fingers. Vibration sense was lost over the fingers of both hands, right toes and left stump, and diminished over the right tibial surface. All areas of sensory loss had fading borders. *Radiographic examination*—The right ankle showed narrowing of the joint space with irregularity of the anterior part of the articular surface of the tibia. Hypertrophic changes

FIG. 4

Case 1—Radiograph of right foot. The talus shows sclerotic degenerative changes with narrowing of its articular margins.

were shown at the neck of the talus (Fig. 4). The subtalar joint was almost obliterated. The talo-navicular, navicular-cuneiform, and first and second metatarso-cuneiform joints all showed a similar change. The bones were sclerotic and irregular. The right third and fourth

FIG. 5

FIG. 6

Case 1. Figure 5—Right foot, showing destruction of the third and fourth metatarso-phalangeal joints. Figure 6—Right hand showing loss of phalanges.

metatarso-phalangeal joints were destroyed and the second distal phalanx was represented by only a small fragment of bone (Fig. 5).

The hand showed narrowing of the right first metacarpo-phalangeal joint with medial and lateral osteophyte formation of the proximal phalanx (Fig. 6). The distal phalanx of the thumb was fragmented, as were those of the right and left little fingers and the left fourth

finger. The middle phalanx showed marked loss of substance in the second and third fingers and in the fourth finger of the right hand.

Case 2—This boy, the brother of the patient described in Case 1, was eighteen years old at the time of examination. His history is similar to that of his brother, progressive degenerative processes taking place at the periphery of the limbs. When he was four the mother noticed fissuring of the toe-nails, which eventually separated. At the age of nine he suffered a burn to his right hand when touching a stove. This did not cause any pain. Subsequent skin necrosis and spontaneous extrusion of bony fragments occurred from the finger tips, the wounds healing slowly. Similar incidents have recurred over the years, the most recent being three months ago. At that time a painless inflammation of the right third distal phalanx of the hand occurred, resulting in a loss of that part.

FIG. 7

Case 2—Right and left feet. The left shows marked swelling and deformity. Note trophic changes of the nails.

When he was twelve he fell and fractured the right femoral shaft. The fracture was treated by open reduction and internal fixation, but despite prolonged immobilisation in plaster union was delayed. After a further operation the femur became infected. When he was seventeen he fractured his left femoral shaft in another fall. This fracture was treated by skeletal traction and later by immobilisation in plaster.

While he was walking with crutches he developed a swelling on the left foot, with a dull ache, and this degenerative process has progressed. He also developed an infected ulcer at the right fifth metatarsal, which was later excised. At age eighteen an inflammatory process occurred at the lateral malleolus of the left ankle. After surgical drainage this wound eventually healed. This patient, like his brother, suffered malaise and fever before he would notice any inflammation of a part.

On examination, both hands showed loss of the distal phalanges. The skin was thickened, with small dry ulcerations at several sites.

There was an inch of shortening of the left lower limb. The right foot was in slight

valgus (Fig. 7), and the range of ankle and subtalar movement was impaired. The fifth metatarsal and phalanges were absent and the other toes were deformed. The left foot and ankle were markedly deformed (Fig. 7). The range of ankle movement was only 20 degrees, and of tarsal movement nil. Both malleolar regions were markedly swollen and indurated and there was a sinus over the lateral malleolus.

Neurological examination revealed normal cranial nerves, musculature and co-ordination. All abdominal and deep reflexes were absent. The plantar responses were normal. Peripheral sensory loss was complete over the hands and feet, and faded towards the shoulders and waist. *Radiographic examination*—There was slight spondylolisthesis at L.5-S.1. The uppermost third of the right femur was irregular and sclerotic—the site of previous fractures. The

FIG. 8

FIG. 9

Case 2—Radiographs of feet. On the left side (Fig. 9) the talus and the adjacent joints show marked degenerative changes.

right ankle showed only slight changes (Fig. 8), but the left ankle and foot showed severe destruction that has progressed rapidly in the past two years (Fig. 9). The ankle joint was greatly narrowed. The changes were confined to the calcaneum, talus and navicular, with their adjacent joints. Marked sclerotic changes have occurred of each of these bones with fragmentation of the navicular. The distal phalanx of the fourth toe was partly destroyed. The hands showed changes like those in Case 1.

Case 3—This thirteen year old girl is the youngest sibling. When she was six she injured her right foot and suffered some loss of skin. Three weeks later she noticed a vague dull ache over the whole of the foot, which was enlarged but not reddened. After rest in plaster the inflammation subsided but the foot was never restored to normal. By the age of ten the right foot had developed a painless inflammation of the fifth toe. She remained in bed for three months, but the ulceration progressed. Later she fractured the left femur in a fall. When she was twelve the right fifth toe ulceration had progressed to the degree that osteomyelitis of the metatarsal had occurred. The distal part of the metatarsal and the phalanx were excised. A biopsy of the right lateral femoral cutaneous nerve was obtained at that time. The pathology was as reported by Denny-Brown (1951). At about that time successive fractures of various metatarsal bones occurred. Some were related to injury, whereas others were attributed to stress phenomena. Union occurred in all cases.

On examination, the present time, the patient was able to walk only with crutches. A small ulcer was present over the lateral aspect of the right cuboid bone. Part of the

right fifth metatarsal and all of the phalanges were absent (Fig. 10). An area of redness was present at the lateral aspect of the base of the left fifth toe, and several of the toes were deformed. The right index finger showed inflammatory changes of the distal phalanx with some loss of substance at the tip. Trophic nail changes were present.

Neurological examination revealed normal cranial nerves, musculature and co-ordination. All abdominal and deep reflexes were absent except the triceps jerk, which was sluggish on both sides. There was no Babinski sign. Sensation to touch and to pain was absent over the periphery of all four limbs, with fading borders towards the shoulders and thighs. Temperature sensation was present to a slight degree over the toes and fingers, but vibration sense was absent in these areas. Impaired appreciation of position sense was noted in the fingers and toes.

FIG. 10

Case 3—Right foot showing ulceration over the cuboid area. The skin shows patches of vitiligo.

Radiographic examination—The right foot showed osteolytic destruction of the fifth metatarso-phalangeal joint, the site of the skin ulcer. The distal phalanges of the fourth fingers and of the left fifth finger showed early irregularity at their extremities. There was slight spondylolisthesis at L.5–S.1.

DISCUSSION

The patients described all began to have symptoms in the first decade of life. The initial and some subsequent problems have had their origin in the damage done to the skin. Others have been due to progressive joint destruction. In spite of having other members of the family suffering from this disease, the youngest sibling could not limit her activities in order to prevent a similar sequence of events for herself. Her problem at the present time is ulceration of the lateral aspect of the right tarsus. Normal care of the feet and the use of proper shoes have not been sufficient to prevent the skin necrosis. It is probable that she too will suffer destructive changes of the foot and ankle, with perforating ulcers.

The pathological processes at work in the various joints are similar to those described

in *tabes dorsalis* and named Charcot joints. The constant abuse of the bony structure, its movements unlimited by reflex or pain activity, allows for the development of traumatic arthritis. This progresses, fragments of bone being released and joint spaces being obliterated. This is shown well in the radiographs of the feet. In Case 2 the patient's left foot is unstable, and it is not controlled well by a surgical boot and side-bar. Though arthrodesis might be attempted it seems likely that amputation will be required. But below-knee amputation through an area of hypaesthesia has its own problems, as illustrated in Case 1. The skin is of poor texture and a draining sinus persists. This area still has a moderate sensory deficit. Supracondylar amputation may have been a better choice. With the frequent occurrence of infected ulcers of the right foot, the outlook is most gloomy.

Though the cases of Tocantins and Reimann (1939) may not have been of the same etiology, the management of their problem was similar. They described a man of fifty-three who was able to continue at work despite multiple amputations. The family reported by Heller and Robb (1955) also included a man of eighty-two. He has lived his life without major surgery being performed, though he suffered a spontaneous right mid-tarsal amputation. Several small ulcers exist at his stump. One should not by-pass the best and obvious method of therapy, distasteful as it may be. Suffice to say, there is room for conservative treatment and it should be used to the full.

The management of these perforating ulcers is based upon complete rest, constant attention to the wounds, and antibiotics. Most of the problems have been of the lower extremity. Weight bearing has had to be either reduced or eliminated. Though bed rest is the desirable objective, most of these people do not have the patience to remain recumbent for many months several times each year. Crutches may be used, alone or in combination with plaster-of-Paris.

When ulcerations constantly occur over a bony prominence the question of resection of that area should be considered. However, a word of caution: not infrequently, the soft tissues are the site of a chronic inflammatory process. This leads to slow healing of the wound, as noted in Case 1.

SUMMARY

1. Three cases of hereditary sensory neuropathy are reported.
2. Neuropathic destruction of joints and chronic infected ulcers are the primary problems of management.
3. This entity must be considered in the differential diagnosis of trophic ulcerations of the extremities.

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