Remarks

ON

SYRINGOMYELIA (SACRO-LUMBAR TYPE),
OCCURRING IN A BROTHER AND
SISTER.

BY

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THE occurrence of syringomyelia in a familial form seems to be extremely rare. That anomalies of development of the spinal cord lie at the origin of syringomyelia in most cases is established by the accumulated pathological data. Moreover, other abnormalities of the antenatal development of the nervous system are not infrequently found in association with syringomyelia, such as hydromyelus, spina bifida, meningocele, doubling of the central canal, and hydrocephalus. It is not surprising, therefore, that occasionally developmental defects of this kind should occur in more than one member of the same family.

With regard to the sacro-lumbar type of the disease—an uncommon form—the one present in the cases described below, Schlesinger¹ notes that it is not to be sharply distinguished from spina bifida, that it often arises out of it, and is observed at an early age.

The only cases which at all resemble our own which we have been able to find in the literature are three reported by Bruns² under the title of family symmetrical gangrene and arthropathy, or familial syringomyelia. Four children out of a family of five born of healthy parents were affected in the same way. The disease began in each of them at about the age of 17, with symmetrical ulcers on the balls of both great toes, which perforated into the joint. Other ulcers formed on the feet from time to time, suppuration extended into the joints with formation of fistulae and discharge of small portions of bone. Deformities and loss of some of the toes occurred, varying in the different cases. Finally, great deformity of the feet resulted. In addition, there were present muscular atrophy, loss of the tendon (including knee-jerks), and skin reflexes, and deficiency or loss of sensation to touch, pain, and temperature. The absence of much pain was shown by the fact that in spite of the extensive disease of the feet the patient was able to walk about. The cases differ from ours in that there does not appear to have been dissociation of sensation and in the loss of the knee-jerks; in other respects, and in the absence of disturbance of the bladder or rectum, they correspond. Bruns refers to cases reported by Bramann at the Berlin Surgical Congress in 1889, of which we have only been able to obtain access to a short abstract. These were three brothers, aged 13, 11, and 7 years respectively. Their parents and sisters were healthy. The patients were badly nourished and anaemic,

and suffered from symmetrical gangrene of the fingers and toes, together with ulcers of the feet. The symmetrical characters of the lesions and the sensory disturbances pointed to syringomyelia.

It is probable that a more extensive search through the literature would discover other cases of the same kind published under different titles, for it may be remarked that syringomyelia probably plays a larger part in the production of chronic joint diseases than is generally held, and that in such cases the disease of the bones and joints is so conspicuous that attention is wholly concentrated upon them, and accordingly the observer fails to look for the sensory and other manifestations of syringomyelia, which, being present in comparatively slight degree, pass unrecognized. This point is brought out by Borchard,³ who remarks upon the want of parallelism between the grade of the joint affections and the severity of the remaining nervous symptoms.

In the second of the cases described below the nature of the disease was so unusual that when the patient appeared with a large chronic ulcer on the dorsum of her foot a family likeness was at once recognized from the breadth of the foot and the out-turned great toes before it was known that the patients were brother and sister.

CASE I.

L. F., a man, aged 23, was admitted into the General Hospital, Bristol, on September 9th, 1908, for deformity of the legs.

The family history was unimportant; father, history unknown; mother, healthy. In addition to the sister described below there are three sisters alive and in good health; one sister died, cause unknown.

Past History.

He had always been healthy except for the affection of the feet. This dates from infancy, and he has never been able to stand upright since he can remember. He has often suffered from "sores on the feet," and it was for such a

Fig. 1. Case I.—Skiagram of right foot.

condition that he first came under care as an out-patient. He then had a large chronic ulcer over the dorsum and outer side of the right foot, several inches in diameter. How these ulcers originated he is unable to say, but he thinks they are due to the rubbing of the boot. They are always painless.

Condition on Admission.

He is a robust, broad-chested, intelligent man, with normal head, arms, and trunk. He is obliged to walk with crutches because both knees are so much bent. His natural height—which is about 5 ft.—is reduced to 4 ft. by the flexion of the knees and hips. There is marked lordosis and flexion of the hips, which are no doubt compensatory for the knee flexion. All the deep reflexes are much exaggerated, and there is marked ankle and patellar clonus on both sides. Extension plantar reflex (Babinski's sign) present on the right side. Both feet are remarkably broad and elephantine. This is partly due to the great breadth of the metatarsal arch and partly to the almost complete loss of the toes. On the left foot there is no vestige of a toe, and the heads of the metatarsal bones have also disappeared. The stump is covered by the scar tissue left after the recurrent ulceration. The right foot retains a great and second toe, both of which are shrunken and deformed, and the stumps of two other toes. He states that the toes have been lost after various attacks of ulceration.

Skiagrams of the right foot (Fig. 1) show that in the great toe the first phalanx has practically disappeared, the distal phalanx is short and broad; the second, third, and fourth toes have a proximal phalanx, that of the fourth being atrophic at the distal end; there is no other trace of any other phalanges. Of the metatarsal bones, the first is short, its distal end is

atrophied; the distal ends of all five are transparent as compared with their shafts. The tarsus is normal.

In the left foot the bones of the tarsus are remarkably transparent; their outlines are so indefinite, and the amount of destruction so great as to make it impossible to say exactly what bones remain, the condition of the whole tarsus resembling the extensive changes of an advanced Charcot's joint. The fourth and fifth metatarsal bones remain, the third is atrophic and shows a fracture; the basal stumps of the first and second are indistinctly visible, and the remnant of one phalanx only lies loosely in the soft tissues.

The hands are rather broad; no anomalies are shown by the x rays, except that the terminal phalanges of the thumbs are remarkably short and broad (Fig. 2).

Both knees are flexed; the right to 100 degrees, and the left to 110 degrees, and though they cannot be extended beyond this point they move freely in the direction of further flexion.

Each knee-joint contains a great excess of fluid, and the lower end of the femur is unnaturally prominent owing to the backward displacement of the tibia. The condition of the patella is very remarkable. It is distinctly double on both legs, a wide gap being palpable between the fragments. On the right side each is of about the size of a normal patella, but on the left side the upper fragment is decidedly smaller than the lower. The drawings (Fig. 3) from the skiagrams show these points. Over both feet and lower part of the legs there was dissociation of sensation (Fig. 4), in that heat, cold, and pain (pin-prick) were not distinguished as such, whilst a light touch (cotton-wool) and firm pressure were normally felt. A pin-prick was occasionally felt as "hot." On the sole of the left foot there was also slight anaesthesia to a light touch. Hyperidrosis or other vasomotor phenomena were not observed.

First Operation.

On September 11th Mr. Groves divided all the hamstring tendons, with the fascia at the back of both knees, by subcutaneous tenotomies, and it was possible then to extend the knees slightly. But in spite of long-continued weight extension, and the use of a hinged splint devised by Mr. Maynard, the house-surgeon, it was impossible to obtain full extension. Moreover, the more extended the knees became, the more was a position of genu valgum produced; so that, for walking unaided, the legs were as useless as ever.

Second Operation.

On December 1st the right knee was opened by a U-shaped flap, and the lower portion of the patella removed, the appearances being exactly those found in cases of fractured patella with long fibrous union. The object of this removal was to shorten the quadriceps tendon. The joint being freely opened, the cartilaginous surfaces were seen to be rather vascular and velvety in front of the femur, where articular friction had never taken place. The posterior and crucial ligaments were freely divided, and full extension then became possible. The internal condyle was separated by Ogston's incision, and the leg put up in a normal position. On the left knee an osteotomy was performed just above the condyles, and this allowed correct reposition.

After-history.

He has since made a good recovery (Fig. 5), and can walk with sticks. The lordosis and hip flexion into which he has grown by long use still remain, but are becoming less prominent as he walks about.

A few remarks may be added as to the nature of the leg deformity. The flexed knees seemed to be due to a spastic paralysis, the marked hypertonicity of the muscles being evidence of this. He had worked from boyhood as a basket-maker, squatting on his doubled-up legs. The condition of the knee-joints resembled the early stages of neuropathic arthritis, inasmuch as there was a painless synovial effusion with partial dislocation. The double patella is a very rare—if not unique—phenomenon, and admits of one or two explanations. Either that a double ossification took place in the quadriceps tendon, or else that a fracture took place after ossification was complete. Now there does not seem to be any reason why an abnormality in ossification should have occurred in

this case, and it is always unlikely that two rare conditions should co-exist in the same patient. But if the underlying disease is a syringomyelia, then the fact of both patellae having undergone spontaneous fracture is consistent with the well-known tendency of this affection.

CASE II.

I. F., aged 15; no occupation; sister of the foregoing; admitted October, 1908.

History.

She had never had any bad illness, except that she had an operation performed on the left mastoid three years ago, from which she made a good recovery without any impairment of hearing.

About four years before admission a superficial ulcer appeared on the left big toe, and after some time another came on the right little toe. These ulcers gave her some pain in walking, and were very intractable in healing. Other smaller sores or ulcers affected the toes at various times subsequently. Eighteen months ago she had a large bunion on the right big toe, which became ulcerated. Since then she has had extensive sores on the left toes and foot, which slowly healed. Two months before admission a sore developed on the right foot as the result of knocking her foot slightly, and she applied hot fomentations to it. She did not feel the fomentations too hot, but they scalded her foot and were followed by extensive ulceration; the ulcers gave her slight pain in walking, and discharged freely, but did not prevent her getting about nor disturb her sleep. It was on account of the sores on this foot that she sought advice.

Condition on Admission.

She is a short, fat, thickly-set girl, with a red face and somewhat heavy expression; speech is deliberate, somewhat syllabic, but distinct; intelligence good, but she is slow in answering questions; skull of normal size and shape; face broad and

Fig. 2. Case I.—Skiagram of thumbs.

Fig. 3. Case I.—Diagram from skiagrams to show the condition of the patellae.

round; teeth irregular; no evidence of hereditary syphilis; the enamel of the teeth was markedly defective; no paralysis of any cranial nerve; no nystagmus; sight and hearing good; optic discs and retinae normal; pupils equal, and react normally to light and accommodation; chest well developed; thoracic and abdominal organs normal; urine normal; micturition and

both first metatarsals are strongly adducted, this accounting for the increased breadth of the feet. The left first metatarsus is shortened, broadened and enlarged at its head, whilst the corresponding first phalanx is very short, with marked hypertrophy of its base and lipping of its margins. The second toes in both feet and the left fourth toe were shortened, and were soft and

Fig. 4. Case I.—Diagram of sensational disturbance. Over the shaded areas there was loss of sensation to heat, cold and pain (pin-prick); light touch and firm pressure felt.

Fig. 6. Case II.—Diagram of sensational disturbance. Over the shaded area heat and cold were not distinguished, felt as touch only; sensation very deficient to pin-prick, generally felt as touch; over dotted area some alterations, but less marked, to pain. Light touch and pressure felt everywhere, except over toes of right foot (black shading).

defaecation normal; spine regular, no deformity, no scoliosis; sacrum and coccyx normal; relative length of thighs and arms to legs and forearms respectively, normal; hands rather broad and thick; no atrophy of muscles in upper extremities; sensation normal to light touch, pain, heat and cold over face, neck, trunk, upper extremities and thighs; over the lower part of the

legs and over the feet (see Fig. 6) heat and cold were not distinguished; there was also, but less marked, defective sensation to pin-prick, which was sometimes felt as a prick but more often as a touch; light touch and deep pressure were everywhere promptly recognized and localized, except over the toes of the right foot, the seat of the long-continued ulceration of the skin. The upper limit of defective sensation was well defined to heat and cold, and passed circularly round the limb; to prick it was indefinite, fading away insensibly above. There was no hyperidrosis; the feet inclined to be cold; there was some tendency to flushing of the skin over the left ankle; muscles of legs all well developed, strong, no paralysis of movement, and no incoordination. The muscles acted to both interrupted and constant currents, by direct and indirect stimulation; deep reflexes, in upper extremities normal; knee and ankle

Fig. 5. Case I.—Condition of legs after operation.

jerk much exaggerated, equal on both sides, no clonus.

Superficial Reflexes.—Abdominal normal; extensor plantar reflex in right foot (Babinski's sign) and often on the left, but is not always obtained on the latter.

Feet.—Very extensive superficial ulceration on dorsum of right foot, involving the great toe and all the other toes, chiefly on their dorsal surface. Under rest and suitable dressings this ulceration healed rapidly, as well as in a normal person, and was completely healed in four weeks. The feet are short and stunted, especially the right, the latter is inverted (Fig. 7). Skiagrams show (Fig. 8) that the right great toe is partly dislocated outwards on its metatarsal bone; there is thus pronounced hallux valgus, and a less degree of it on the left side;

“wobbly” as if boneless; the right fourth and fifth toes had disappeared. Skiagrams show that the second and third phalanges of both second toes, of the right fourth and fifth, and left fourth toes have disappeared, whilst the first phalanx of the right second and fifth and left fourth toes have undergone extreme atrophy. In the left foot there is slight pes cavus, partly due to internal angulation between base and shaft of the second, third, and fourth metatarsals. There is some thickening about the right ankle-joint. The knees and legs appeared normal, and the muscles well developed, with the exception of the small muscles of the feet, which were slightly wasted, but reacted to electricity.

It should be added that in spite of the ulcers on the feet and the disease of the bones of the toes the patient was walking about when she came to the hospital. The readiness with which the ulceration healed under appropriate treatment is also noteworthy, and appears to have also taken place in the other cases quoted above.

The skiagrams show that the disease of the bones is mainly an atrophic one of a somewhat remarkable kind; some of the phalangeal and metatarsal bones appear to have undergone a process of atrophy and to have gradually disappeared. Indeed, the appearances of some of the toes of the second case suggest that the bone changes are not always secondary to ulceration spreading from the skin, but that the atrophy of the phalangeal bones is a primary process, for in these toes the soft parts are left

Fig. 7. Case II.—Photograph of feet.

ntact, while the phalanges have disappeared, or are represented by small atrophic remnants. In this respect they show well the characters described by Tedesco⁴ as present in syringomyelia, and which he regards as constituting a special type of bone affection. These characters are: (1) A general transparency of the bone shadows as a whole; (2) diminution and softening of the cortical layer of the diaphyses; and (3) rarefaction of the spongy bone while its gross structural form is retained, so that the external contour or form of the bone is preserved. When the process of atrophy is far advanced, however, the bone gradually disappears, as the skiagrams show. The increased brittleness which accompanies this process of osteoporosis accounts for the occurrence of fractures. Spontaneous fractures, however, do not occur very frequently in syringomyelia—not so much so as one would expect. At the same time, a certain amount of hypertrophy of bone, with lipping of and outgrowths from the ends of the bones, together with periosteal thickenings, go on side by side with atrophy; and similarly in the cartilages processes of absorption and new formation take place. Effusion of fluid into the joints is frequent; there seems little tendency to ankylosis. According to Kienbock,⁵ the x-ray appearances are against the view which has been brought forward that the bone changes in syringomyelia depend on disappearance of calcium salts.

As to causation, little support is given by these cases to the theory which makes traumatism play an important part in the production of bone and joint lesions in this disease. We are not now speaking of the joint lesions produced by infective processes through perforating, or other deep ulceration of the skin and subcutaneous tissues. In neither case could any history of severe or even repeated

slight injuries be obtained, such as would be likely to directly damage the joints, and the alternative theory which assigns the changes to trophic and vasomotor disturbances seems the more probable one.

In the bone and joint lesions of syringomyelia, the joints most frequently affected are the elbow and shoulder joints. It is possible that the cases under discussion are representative of a distinct but rare group of the disease, in which the sacro-lumbar region of the cord is alone involved, and in which bone lesions are the predominant feature.

Lastly, with regard to the distribution of the disturbance of sensation; in each case this was similar, in that its upper limit passed circularly round the limbs at right angles to their long axis. Catola and Lewandowsky⁶ remark on this point that this form of sensory disturbance is not infrequent in syringomyelia, as well as the customary root arrangement, and they attribute it to a partial interruption of the long tracts in the spinal cord, which they think occurs in this disease, and, as cases of tumour of the cord show, can produce a sensory loss with this distribution.

It is our pleasing duty to express our indebtedness to Mr. J. E. Jones, Skiagraphist to

the Bristol General Hospital, for the excellent skiagrams.

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Fig. 8. Case II—Skiagram of feet

A CASE OF ORAL SEPSIS WITH PECULIAR GENERAL SYMPTOMS.

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In view of the interest which at present attaches to all the complications of oral sepsis, the following case seems worth recording:

Mr. T., aged 35, holding a responsible position in a well-known firm of surgical instrument manufacturers which necessitates a good deal of travelling (never beyond the British Isles), had been in good health except for several attacks of influenza and a nervous breakdown at the age of 16 which caused him to abandon a career as a teacher. No previous history of rheumatism, gout, syphilis, gonorrhoea, or alcoholism could be obtained. On March 27th he had aching in the head, back, and

limbs. Thinking that it was an attack of influenza, he went to bed and applied some domestic remedies. Next morning he was much better, and in a few days was able to return to work. On April 2nd, whilst out on business, he was taken ill with a violent headache, and had some difficulty in getting home. After two days in bed he felt better, and went to the seaside to recuperate, but whilst there on April 9th, 12th, and 15th, he had similar attacks during which he went to bed, being able to get about in the intervals.

He was first seen by one of us on April 16th, and gave a very clear account of the attacks, which, he said, were all alike. The first thing he noticed was the appearance of a red spot on one limb (near a joint) which enlarged till it was about $\frac{1}{2}$ in. in diameter. At first painless, it soon began to burn, and then the neighbouring joint became painful and swollen. In the course of a few hours he felt chilly, and his temperature rose to about 103° F.; with the temperature the headache and pain increased, so that the joint could not be moved. About one hour later perspiration commenced, at first slight, but soon so profuse that the bedclothes were saturated. With the onset of perspiration the pain became less, and vanished in about an hour, whilst the perspiration itself continued for about four hours. After an attack he passed a good night, and felt comparatively well next morning.