Scolloping of the vertebral bodies in von Recklinghausen's disease of the nervous system (neurofibromatosis)

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It is well known that intraspinal tumours may erode the bodies and arches of adjacent vertebrae. Rogers (1955) indicated how vascular intraspinal tumours may erode the backs of the vertebral bodies yet spare the intervertebral discs, in a manner resembling the erosion of vertebrae by an aortic aneurysm.

In von Recklinghausen's disease of the nervous system neurofibromatous tumours frequently arise from the sheaths of peripheral, cranial, and spinal nerves. The presence of superficial stigmata of the disease together with hollowing out or scolloping of the posterior surfaces of the vertebral bodies is likely to be interpreted as evidence of intraspinal tumour. In fact, this scolloping may occur in neurofibromatosis as an associated defect in the absence of any intraspinal tumour. Vertebral scolloping has been described in association with lateral thoracic meningocele by Cross, Reavis, and Saunders (1949) and by Bull (1950). In two-thirds of the reported cases of lateral thoracic meningocele there has been frank evidence of neurofibromatosis (Nanson, 1957). A case of scolloping without extraspinal meningocele was demonstrated radiologically by Braun in 1955 and another by Schröder in 1956: in neither instance was exploration or necropsy performed. Whitehouse (1958) noted the deformity in two children; he regarded it as a manifestation of meningocele, and called it intraspinal meningocele.

We have studied six patients with neurofibromatosis and scolloping of vertebrae who had neither meningocele nor intraspinal tumour. Two of these cases came to necropsy and two were submitted to laminectomy. Our observations lead us to suggest that the scolloping deformity is of developmental origin. In order that unnecessary exploration of the spinal canal be avoided, we feel that it is important to recognize that scolloping can occur in the absence of both intraspinal tumour and meningocele.

CASE REPORTS

CASE 1 A woman aged 54 years presented with an axillary sarcoma. Numerous café au lait patches and multiple superficial nodules had been present all her life.

She had no complaint referable to the spine. Figure 1 shows the condition of the vertebrae: an appreciable kyphoscoliosis with well-marked scolloping of the dorsal surfaces of the lower seven thoracic vertebral bodies. There was nothing to suggest spinal cord or nerve root involvement.

She died from pulmonary metastases, and permission for necropsy was refused.

CASE 2 A 41-year-old man was admitted from another hospital where an operation for horseshoe kidney and renal calculus had been performed a month earlier. Two days before admission he had sudden back pain with numbness and paralysis of the legs. This passed off and then recurred on the following day and he was unable to pass urine.

On examination he had numerous café au lait patches and multiple superficial nodules with a large plexiform neuroma in the right popliteal fossa. He had lost all sensation below the level of the umbilicus and his legs were completely flaccid. The abdominal reflexes were absent.

Radiographs of the spine showed 'gross erosion of the pedicles of T4 to T11 and in addition scolloping of T9, T10, and T11' (Fig. 2). These features were interpreted as being due to a large intraspinal neurofibroma. Subsequent exploration revealed an extradural haematoma but no evidence of tumour outside or within the dura. The cord and its vessels appeared normal. The case was ultimately classified as one of extradural haemorrhage.

CASE 3 A 35-year-old midwife with a family history of von Recklinghausen's disease had multiple superficial nodules and café au lait patches. In 1950 a large pigmented 'tumour' had been removed from her neck and shoulder. In 1958 she gave a two-year history of low back pain and frequency of micturition. For one year she had had episodes of pain and numbness of the left leg.

Power and reflexes in the lower limbs were normal, and

FIG. 1a

FIG. 1b

FIG. 2

FIG. 1a and b. *Lateral and anterior radiographs of thoracic spine to show kyphoscoliosis and well-marked scalloping of the posterior surfaces of the vertebral bodies.*

FIG. 2. *Lateral radiograph of the lower thoracic spine to show scalloping of T9, 10, and 11 vertebral bodies.*

FIG. 3

FIG. 4

FIG. 3. *Lateral radiograph of the lumbo-sacral spine to show well marked scolloping of L4 and L5 bodies.*

FIG. 4. *Photograph, taken at operation, of the bulbous sac-like dura mater in the lumbo-sacral region.*

FIG. 5a

FIG. 5b

FIG. 5a and b. *Lateral radiograph and myelogram of the lumbar spine to show scolloping of the vertebral bodies and the absence of a space-occupying lesion in the lumbar theca.*

FIG. 6a

FIG. 6b

FIG. 6a and b. *Lateral radiographs of the thoracic spine taken in 1952 and 1956. There is no appreciable alteration in the degree of scoliosis over the four-year period.*

there was no certain sensory change. Cerebrospinal fluid protein content was 61 mg. per 100 ml. Radiography of the lumbo-sacral region showed 'well marked scoliosis of the posterior aspects of L4, L5, and upper sacral segments with considerable erosion of the first left sacral intervertebral foramen' (Fig. 3). These changes were believed to indicate multiple intraspinal neurofibromata.

At exploration, the posterior part of the sacrum was found to be expanded and almost paper thin. There were no sacral spinous processes. When the dura was exposed it was found to be expanded into a bulbous sac-like structure (Fig. 4). When opened there was no tumour present. The dorsal surface of the bodies of L5 and the sacrum were abnormally irregular and concave. (The patient also had cervical vertebral deformities which have been reported elsewhere by Heard, Holt, and Naylor (1962)).

CASE 4 This 20-year-old girl began to complain of backache while confined to bed for treatment of a peptic ulcer. On direct questioning a five-year history of slight back pain was obtained; she also admitted to slight frequency and urgency of micturition. She had a history of intermittent epileptiform seizures. None of her family had suffered from neurofibromatosis.

Examination showed a healthy girl with numerous café

au lait patches and a few small, soft skin tumours. There were no abnormal objective signs in the central nervous system. Radiographs of the lumbar vertebrae showed gross scoliosis of the bodies with thinning of the pedicles (Fig. 5a). The body of L1 had a rather streaky appearance suggestive of an angioma. Lumbar puncture produced clear fluid with normal manometry and 29 mg. protein per 100 ml. A myelogram confirmed that there was no space-occupying lesion in the lumbar theca (Fig. 5b).

CASE 5 A boy of 19 years was first seen in 1956 when he had for four years suffered intermittent back pain. There was no family history of von Recklinghausen's disease.

On examination he had multiple café au lait patches and a large bathing trunk naevus. There was marked lower dorsal kyphoscoliosis. There were no abnormal neurological signs. In addition to gross vertebral angulation and wedging the radiographs showed four or more of the bodies to be the site of marked scoliosis. Comparison of radiographs of the lower thoracic spine taken in 1952 and 1956 showed an increase in angulation and partial disintegration of a vertebra but scoliosis of the adjacent bodies was unchanged (Fig. 6a and 6b). Lumbar puncture findings were normal. His symptoms were eased by a spinal brace.

FIG. 7

FIG. 8

FIG. 7. A block of the necropsy specimen of the lower thoracic and upper lumbar spine viewed from the posterior aspect. There was severe deformity of the spine. The concavities (scolloping) of the posterior surface of the vertebral bodies are seen.

FIG. 8. Gough-Wentworth thin section of the lower thoracic and upper lumbar spine cut in the sagittal plane. Two of the lower thoracic vertebrae show well-marked scolloping.

In 1960 he was readmitted with a large left-sided retroperitoneal mass. Gravely ill and wasting rapidly he died on 10 May 1960.

At necropsy the immediate cause of death was found to be haemorrhage into the pleural cavity from a degenerating retroperitoneal sarcoma. This tumour was $28 \times 18 \times 17$ cm. in size adjacent to the left side of the vertebral column. The spinal cord and intrathecal nerve roots were normal. There were no intrathecal tumours. There was no macroscopic evidence of tumour within the spinal canal. When the spinal cord was removed it was evident that there was a gross irregular widening of the spinal canal, with marked scolloping of the dorsal surfaces of the bodies. This is illustrated in Figs. 7 and 8.

On histological examination the retroperitoneal tumour was a spindle-cell sarcoma and malignant infiltration of the vertebrae and ligaments was seen at several sites.

CASE 6 This child, aged 6 years, presented with a history of developmental delay, clumsiness of the right arm and stumbling on the right leg for several years. A right facial weakness had been noted at the age of 4 years.

On examination he had multiple café au lait patches, with hydrocephalus and right hemiparesis. Investigations suggested possible aqueduct stenosis. During the next two years he had two epileptiform seizures and was then readmitted with papilloedema. He died after left-sided

ventriculocisternostomy. Radiographs of the spine during life had demonstrated scolloping in the lower lumbar region (Fig. 9).

His mother had von Recklinghausen's disease with multiple superficial nodules and pigmented patches.

At necropsy the brain showed diffuse oedema with herniation of the cerebellar tonsils and uncinate processes. The optic chiasma and optic nerves were swollen and there was diffuse enlargement of the pons and medulla. Coronal sections of the brain revealed a large tumour, the body of which was situated in the left subthalamic region. The tumour, a spongioblastoma polare, extended upwards into the optic chiasma and downwards into the brainstem.

The lower thoracic and lumbar spine was removed together with the whole of the spinal cord. There was no macroscopic evidence of tumour in the spinal canal. The spinal dura mater, nerve roots, and spinal cord appeared normal.

Large thin sections of the spine cut by the Gough-Wentworth technique (Fig. 10) showed the abnormal concavity of the posterior surface of the vertebral bodies; the posterior longitudinal ligament did not follow the concavity, but bridged the gap, giving a bow-string appearance. Histology of the vertebral bodies (L3 to S2) did not show any evidence of tumour either within or about the bone or ligaments.

FIG. 9

FIG. 10

FIG. 9. *Lateral radiograph of the lumbo-sacral spine to show scoloping of the vertebral bodies.*

FIG. 10. *Thin sagittal section (by the Gough-Wentworth technique) of the lumbar spine L3, 4, 5, and S1. The scoloping of the L4, L5, and S1 bodies is well shown. The posterior longitudinal ligament and dura mater bridge the concavity giving a bow-string appearance.*

DISCUSSION

The realization of the high frequency of bone changes in von Recklinghausen's disease of the nervous system is recent. Although earlier workers gave an incidence of 7%, Holt and Wright (1948) found 29% of their cases displayed an abnormality of bone. Heard (1960) found bone changes in 45% of a group of 79 cases. However, the frequency of significant complications, for example, sarcoma and the more major bone abnormalities, is likely to be abnormally high in hospital patients.

Of all the bone abnormalities in von Recklinghausen's disease those of the vertebrae are most often noted.

In a series of bone abnormalities studied by Heard (1960) 48% affected the vertebrae, which accords with a figure of 43% observed in a series described by Miller in 1936. An accurate assessment of the incidence of bone changes would require total skeletal surveys in all cases as well as precise criteria for the diagnosis of von Recklinghausen's disease of the nervous system. The deformity is commonly a lower dorsal kyphoscoliosis and may be so gross as to cause paraplegia. Miller found that in 20 patients who had paraplegia in association with von Recklinghausen's disease severe angulation of the spine was responsible for the paraplegia in more than half of the cases.

Changes in bone in neurofibromatosis may be the

result of erosion or infiltration by tumour, or from involvement of epiphyseal plate with subsequent growth disturbance. Alteration in innervation or in vascularity has been postulated as the cause of the bone deformity where a neurofibromatous mass is found in the same region as a bone deformity. A small number of cases have been reported in which an associated renal tubular defect was the cause of an osteomalacia; bone softening may also follow disuse. Congenital abnormalities of bone are common and may represent a primary developmental defect in bone formation.

It is clear that the concavities illustrated here were not the result of tumour erosion or infiltration. Although in one case a malignant tumour involved parts of the vertebrae, this was an incidental feature.

In diseases which affect the spine an alteration in the shape of the vertebral bodies is a common finding. This is so in degenerative, infective, metabolic, and neoplastic disorders. In spondylosis, a disturbance in the form and function of the intervertebral discs alters the stresses and strains within the vertebral bodies with a change of their shape; the resulting slight concavity of the anterior surface is accentuated by the formation of osteophytes at the upper and lower edges.

In spondylitis due to bacterial invasion, distortion in shape of the vertebral body tends to be late in appearing and most frequently involves the anterior surface with the formation of an angulated concavity. In senile osteoporosis and other metabolic disturbances which produce bone softening, the discs expand into the vertebrae giving rise to the well known 'fish vertebra' deformity. In secondary neoplasms there is found irregular infiltration, usually osteolytic, terminating in wedging and collapse. In none of these conditions is there any close analogy to be drawn with the deformity of 'scolloping'.

The similarity in appearance between scolloping and the deformity produced by aneurysm and vascular tumours tempts one to search for a com-

parable cause. It has been postulated (Whitehouse, 1958) that the primary fault is dural, an expansion of the spinal dural sac eroding the vertebral canal. Cerebrospinal fluid manometry was normal in our patients so that altered hydrodynamics cannot be implicated. Our necropsy findings revealed no neurofibromatous involvement of the dura. Further, the dura did not always exactly follow the bone deformity (see Fig. 10) which implies that the deformity of the bone is a primary defect.

In our opinion the likely explanation of scolloping is that it is a primary abnormality in the development of the vertebrae, which is associated with neurofibromatosis, itself an inherited Anlage defect.

SUMMARY

Six cases are reported of scolloping of the dorsal surface of the vertebrae in von Recklinghausen's disease of the nervous system. The scolloping is not due to erosion by intraspinal tumour nor accompanied by meningocele. The deformity is regarded as a primary developmental anomaly.

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