LUMBOSACRAL SPINA BIFIDA CYSTICA WITH CRANIOVERTEBRAL ANOMALIES: REPORT OF TWO CASES PRESENTING WITH NEUROLOGICAL DISORDER IN ADULT LIFE

BY

JOHN D. SPILLANE and LAMBERT ROGERS

From the Department of Neurology and the Surgical Unit, Welsh National School of Medicine, the Royal Infirmary, Cardiff

Neurological disorders associated with spina bifida are usually either present at birth or arise in early life. There are two main types. First, lesions of the cauda equina; secondly, lesions at the foramen magnum. Occasionally, symptoms do not make their appearance until adult life and then the developmental nature of the disorder may not be recognized. We propose to give an account of two patients with both lumbosacral spina bifida cystica and craniovertebral anomalies who developed a neurological disorder in adult life as a result of lesions of the cerebellum and brain-stem respectively. We are not aware of any previous record of this association.

Case Reports

Case 1.—G. B. L., aged 34 years, a carpenter and joiner, was admitted to the Cardiff Royal Infirmary on August 9, 1956, for investigation of his complaint of weakness of the legs of one year's duration. He was born with a lumbosacral meningocele (Fig. 1) and wore a protective truss. He did not suffer from enuresis and his only complaint had been of transient sensations of weakness and numbness of the legs whenever he accidentally compressed the meningocele. Sometimes he experienced occipital headache and discomfort if he slept in a supine position.

Difficulty in walking was first noticed one year previously. The left foot, and, shortly afterwards, the right foot began to drag and he frequently stumbled or tripped over kerbs, steps, or on uneven surfaces. Paraesthesia was present in both feet and as weakness of the legs progressed he found his sense of balance was also deteriorating. Nevertheless, he was able to ride a bicycle until two months before admission and he continued at his work. In the last month he could only ascend stairs with much effort and he had begun to feel "pins and needles" in the fingers of each hand. He had no complaints referable to the cranial nerves and sphincter function remained normal.

Examination.—His general condition was good and body configuration was normal. His neck was not short. Gait was slow and laboured; he shuffled along, stumbled

when he turned, and swayed when he closed his eyes. The only abnormality in the cranial nerves consisted of fine horizontal nystagmus on right and left lateral gaze. There was no spontaneous nystagmus or oscillopsia and the fundi were normal. The jaw jerk was normal; speech was normal and there were no bulbar signs.

The range of neck movements was normal but he disliked fully extending his head because of vertigo. Power in the arms was normal, and there was no wasting, incoordination, or sensory impairment. The arm reflexes, however, were abnormal. Both biceps and radial reflexes were absent; the triceps jerks were exaggerated. There was brisk finger flexion on attempts to elicit biceps and radial reflexes.

FIG. 1.—Case 1: Lumbosacral meningocele.

FIG. 2

There was moderate weakness of both lower limbs especially of hip flexion; dorsiflexion of the feet was normal. There was no wasting and muscular development of the legs was good. Both knee jerks were much exaggerated; the left ankle jerk was reduced and the right was absent. Both plantar reflexes were extensor. There was a slight degree of clawing deformity of the right foot. Superficial sensation in both lower limbs to mid-thigh level was impaired. Appreciation of passive movements of the great toe was defective and vibration sensation was considerably impaired at the shins.

Radiological Examination.—In the lumbosacral spine posterior spina bifida from L3 to S1 (Fig. 2) was seen.

The foramen magnum (Fig. 3) was grossly enlarged (antero-posterior diameter 5 cm., transverse diameter 5.5 cm.). There was no evidence of hydrocephalus, chronic intracranial hypertension, or basilar impression.

An anterior view of the cervical spine showed partial posterior spina bifida of C2, C5, and C6. A lateral view showed enlargement of the antero-posterior diameter of the upper cervical spinal canal. Measurements were: C1, 27 mm.; C2, 25 mm.; C3, 17 mm.; C4, 16 mm. Average normal readings are: C1, 22 mm.; C2, 20 mm.; C3, 17 mm.; C4, 16 mm.

Lumbar puncture yielded a clear spinal fluid with a protein content of 90 mg. %, and a slow rise and fall of fluid on jugular vein compression.

Opaque myelography showed that there was no obstruction to the flow of myodil upwards but confirmation was obtained of the increased capacity of the upper cervical spinal canal.

FIG. 2.—Case 1: Radiograph of lumbosacral spine; spina bifida L3 to S1.

FIG. 3.—Case 1: Radiograph of base of the skull, showing gross enlargement of the foramen magnum.

FIG. 4.—Case 1: Upper cervical laminectomy showing protrusion of right cerebellar tonsil through the foramen magnum. Note the tortuous venous plexus.

FIG. 3

FIG. 4

It was considered that the disability resulted from compression of the upper cervical cord by protruding cerebellum. There was no clinical evidence of a cervical syrinx but a minor degree of cauda equina disturbance was presumed to be present.

Operation.—Cervical laminectomy under general anaesthesia was performed with removal of part of the occipital bone forming the back of the foramen magnum. The spinous process of the axis was abnormally large and the posterior arch of the atlas unusually thin, having a circular cross section of only $\frac{1}{8}$ in. diameter and constricting the dural tube which bulged above and below it. When the dura was opened the right cerebellar tonsil occupied the upper part of the spinal canal, was elongated and appeared pendunculated (Fig. 4). Beneath it the posterior spinal veins were congested and tortuous and the right inferior cerebellar artery was looped downwards. The exposure, particularly the removal of the thin posterior arch of the atlas, relieved the compression. The dura was left open to further the decompression.

Subsequent History.—The patient returned to his work as a carpenter three months after this operation. There is residual spasticity in the lower limbs but he leads a nearly normal life.

Case 2.—I. B., a housewife aged 49 years, was referred to Cardiff Royal Infirmary in October, 1956, with the complaint of increasing weakness of the legs. This had developed gradually over the previous 12 months and her gait was now slow and unsteady. She felt that the left lower limb was more affected than the right; in the past few months weakness had also made itself apparent in the left upper limb. She experienced short stabs of abdominal pain occasionally and at night she had been troubled with flexor spasms in the lower limbs. For two months her fingers and hands had become numb and clumsy and paraesthesia was intermittent. Her speech was becoming slurred.

FIG. 5.—Case 2: Scar of operation for removal of lumbosacral meningocele in infancy.

FIG. 6.—Case 2: Atrophy of right side of the tongue.

Her previous history was interesting. She was born with a lumbosacral meningocele which was removed at the age of 1 month. Her development was normal except that the left leg was thinner and $\frac{1}{8}$ in. shorter than the right. There was nocturnal enuresis persisting to adult life and periodic urinary incontinence by day. She played games in school but was rather clumsy. Occasionally she was unsteady on her feet. She had always found that her neck was "stiff"; she did not possess the normal amount of cervical mobility. She tended to turn her trunk when she wished to look sideways. At the age of 19 years she fell down a stairway in a store in which she was working and immediately lost the use of her legs. They were paralysed for five months and she was confined to her house for a year. No further particulars could be obtained about this episode and she had always understood that the paralysis resulted from injury to her spine. Her arms were not affected. Her legs were numb and there was defective urinary control.

She recovered, returned to her work, and led a moderately active life. Her gait was never wholly normal, she tired easily, could not dance, and a tendency to stumble persisted. She married but had no children. She had never suffered from blurring of vision, diplopia, or transient episodes of weakness or numbness of trunk or limbs. Her husband had noticed that at meals of late she frequently coughed and choked slightly. Swallowing was becoming difficult and there was nasal regurgitation at times.

On examination the scar of the lumbosacral operation was visible (Fig. 5) and there was some lack of development of the muscles of the left calf and foot. She was of slight build and walked unsteadily in a slow fashion. Her speech was slightly slurred and nasal, the optic discs and fundi were normal and the corneal reflexes were brisk. Palatal movements were weak and the right side of the tongue showed atrophy and fibrillation (Fig. 6). The jaw jerk was normal.

Movements of the neck were slightly limited in all directions but rotation and lateral flexion to the left were distinctly restricted. There was moderate diffuse

FIG. 7.—Case 2: Antero-posterior tomograph of the craniocervical junction showing fusion of the odontoid to the left lateral mass of the atlas.

FIG. 8.—Case 2: Lateral tomograph of the craniocervical junction showing fusion of spinous processes of atlas and axis and fusion of the odontoid to the clivus.

weakness of the left upper limb with slight wasting of the extensor muscles of the forearm and the dorsal interossei. The reflexes of the upper limbs were symmetrically exaggerated; the finger jerks were brisk. There was moderate weakness of both lower limbs, left more than right. Both knee jerks were exaggerated but the ankle jerks were absent. There were no plantar reflexes; the abdominal reflexes were absent. There was no defect of coordination in upper or lower limbs. All forms of superficial sensation were impaired in the left upper limb below the elbow and in both lower limbs below the knee; the impairment was most marked in the peripheral parts of these limbs. There was no area of dissociated sensory loss. Vibration sensation was absent in the left leg and impaired in the right. Appreciation of passive movements was defective in the fingers of the left hand and in the toes of both feet.

Radiological Examination.—There was a spina bifida involving the first and second sacral vertebrae. The foramen magnum and upper cervical spinal canal were of normal dimensions. There was a bony abnormality involving the atlas and axis, which was studied by tomography (Figs. 7 and 8). There was no basilar impression; the odontoid was fused to the left lateral mass of the atlas and also to the clivus where a false joint could be seen; the spinous processes of the atlas and axis were fused. The atlas was not "occipitalized", there was no atlanto-axial dislocation, and the remaining cervical vertebrae were normal.

Lumbar puncture yielded a clear fluid with normal pressure and a protein content of 24 mg. per 100 ml. The Wassermann reaction was negative and manometry was normal.

Opaque Myelography.—The myelogram flowed

freely upwards through the cervical region and no abnormality was detected.

Operation.—Upper cervical laminectomy under general anaesthesia revealed a fusion of the posterior arch of the atlas with the spinous process of the axis. There was no constriction of the dura, which, when opened, revealed a normal cord and cerebrospinal junction.

Subsequent History.—During the ensuing year her condition progressively deteriorated. The eyes remained normal; speech became more nasal and less distinct; there was loss of the left corneal reflex with hypoalgesia of the left trigeminal field; gross wasting and fibrillation of both sides of the tongue, dysphagia, palatal paresis, and marked wasting of the muscles about the shoulder girdle with fasciculation were noted. The muscles of both hands wasted and the arm reflexes disappeared. Spastic paraplegia was present with all forms of sensation impaired below the 2-3. The pressure and composition of the cerebrospinal fluid remained normal. She died of suppurative bronchitis on October 17, 1957, two years after the onset of the illness.

Necropsy.—Dr. E. E. Payne made the necropsy examination. The spinal cord extended into the sacral canal: there was no lumbar enlargement of the cord but the dural sac in this region was dilated. The cord was tethered to the sacrum. Thin sagittal sections of the cervical spine and posterior fossa mounted on paper according to the Gough-Wentworth method (1949) showed fusion of the odontoid with the left lateral mass of the atlas. The tip of the odontoid was also fused to the clivus and the anterior arch of the atlas was displaced upwards and anteriorly. There was slight diffuse atrophy of the cerebral hemispheres with slight ventricular

FIG. 9.—Case 2: Transverse section of the sacral spinal cord showing maldevelopment of posterior segments of the cord.

dilatation. There was no tumour or distortion of the brain-stem or cerebellum. A spindle-shaped area of degeneration approximately $1\frac{1}{2}$ in. in length was seen in the anterior half of the medulla extending from its junction with the cervical cord below to the pons above.

Microscopical examination of the nervous system could not include the medulla as this had been sectioned *in situ* in preparing the thin sagittal sections of the cervical spine and base of the skull. The brain was microscopically normal. In the mid-brain many of the large ganglion cells contained pigment. In the thoracic cord there was chromatolysis and reduction in number of the anterior horn cells with sclerosis in the posterior and lateral columns. The architecture of the lumbosacral cord was grossly distorted: fat and fibrous tissue replaced the posterior segments (Fig. 9). Sections of muscle from the thenar groups, forearm extensors, and tongue showed fibre atrophy and sarcolemmal proliferation.

Commentary

We here have two adults with lumbosacral spina bifida and craniovertebral anomalies presenting with a progressive neurological disturbance. In Case 1 there was spastic paraplegia, altered arm

reflexes, nystagmus, and ataxia; radiological examination revealed an enlarged foramen magnum and upper cervical canal. Laminectomy disclosed compression of the cord from a herniated cerebellum projected through and itself compressed by an abnormal posterior arch of the atlas. In Case 2 there was progressive paraplegia and bulbar palsy together with sensory loss in the left trigeminal field, in the left upper limb and below th. 2-3. Wasting and fasciculation of the muscles of the upper limbs were observed. At operation there was no herniation of the cerebellum or deformity of the medulla and no syrinx or abnormal dural adhesion or membrane.

In the first case the diagnosis of cerebellar herniation was considered most probable in view of the enlargement of the foramen magnum. In the second there was no compressing lesion of the medulla or cervical cord. Disseminated sclerosis was excluded in virtue of the muscular wasting and fibrillation; pathological examination of the brain and thoracic cord disclosed no areas of characteristic demyelination. The paralysis of the legs at the age of 19 years

was considered to be due to injury to the congenital lumbosacral malformation, an occasional occurrence in patients with spina bifida cystica. (Concussion of the sac in Case 2 also tended to induce weakness and numbness of the lower limbs.) Motor neurone disease was excluded by reason of the sensory loss in the left upper limb and the left side of the face. The original sensory disturbance in the lower limbs may have been due to interference with the cauda equina but as the disease advanced all forms of sensation were impaired below upper thoracic levels. Although the classical zones of dissociated sensory loss of syringomyelia were not found it was thought that a degenerative lesion was present in the medulla and cervical cord which was in some way related to the congenital anomalies at the upper and lower ends of the spinal canal.

In retrospect it is unfortunate that the technique of post-mortem examination of the cervical spine and cord by the thin section method was adopted in this case. We thought it would help in disclosing the relationships between the craniovertebral anomaly and the lesion in the neuraxis. Macroscopic degeneration was revealed in the medulla but histological studies were then impossible for the reasons already given.

There are two aspects of these cases which are noteworthy, first, the problem of delayed neurological manifestations of spina bifida, and secondly, the question of the association with abnormalities at the foramen magnum.

It is well known that when they occur the neurological complications of lumbosacral spina bifida occulta may not make their appearance for some years—until late childhood or adolescence (Craig and Mulder, 1956). Laminectomy may then enable us to remove a pad of fat compressing the dura or sever adhesions or bands tethering or exerting traction upon the cauda equina or conus medullaris. In neither of our patients, however, was this the explanation: the neurological disability arose not at the caudal end of the spinal canal but at the craniovertebral junction and was therefore not due to the spina bifida.

Cerebellar herniation through the foramen magnum may be found in patients with or without craniovertebral anomalies and meningocele. In association with craniovertebral abnormalities in adult patients it has been observed by Chamberlain (1939), Gustafson and Oldberg (1940), List (1941), Craig, Walsh, and Camp (1942), Scoville and Sherman (1951) Garcin and Oeconomos (1953), Cogan and Barrows (1954), and by others. With a normal craniovertebral junction it has been reported by Aring (1938), McConnell and Parker (1938), Ogryzlo (1942), Bucy and Lichtenstein (1945),

Gardner and Goodall (1950), and by others. Both types of case have been described by Spillane, Pallis, and Jones (1957). Although the term "Arnold-Chiari malformation" is usually applied to the deformity of the hind-brain in hydrocephalic infants with meningocele, Chiari (1895) also described cases in which there was no meningocele and some of these patients reached adult life (six out of 14). He referred to these as type 1 and pointed out that the fourth ventricle was not displaced below the foramen magnum. The cerebellar "peg-shaped appendage" was often asymmetrical and sometimes unilateral. It is this type of malformation which was found in our first case. There was nothing to suggest that the cerebellar displacement was a result of "traction" from below or pressure from above. It is presumably an anomaly of development associated but not in any way dependent upon the grosser anomaly at the sacral end of the vertebral column.

Summary

Two cases are reported of the association of lumbosacral spina bifida cystica with craniovertebral anomalies.

Neurological disability arising from the lesion at the cranio-vertebral junction began at the age of 33 years in the first case and 48 years in the second case.

Operation disclosed a Chiari malformation (type 1) in the first patient. In the second case the appearances were normal but at necropsy a year later the only lesion consisted of a softening of the medulla and cervical cord.

There was no evidence in either case that anchorage, pressure, or traction on the neuraxis was primarily responsible for the lesion in the nervous system.

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