ARNOLD-CHIARI DEFORMITY WITHOUT BONY ANOMALIES

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SINCE the early and original contributions of Arnold and Chiari on the malformation that now bears their names, numerous articles have appeared pertaining to the mechanism, diagnosis and treatment of this anomaly. The occurrence of this deformity has usually been found in infants to be associated with extreme degrees of spina bifida and cranium bifidum. When encountered in adults this pathologic entity has generally been seen in conjunction with some type of bony anomaly of the cranio-vertebral junction, such as basilar impression (platybasia) or the Klippel-Feil deformity.

A survey of the literature, however, reveals several cases in which this malformation has been proven, either at operation or autopsy, to exist without spina bifida or coexisting bony anomalies of the spine. The case reported here illustrates the fact that this anomaly may exist in the absence of either bony deformities or spina bifida, and furthermore that it may occur in the very young, our patient being the youngest of those recorded. It is because of this fact and in view of the unusual progress and diagnostic difficulties presented by this case that we set forth our experiences in dealing with this problem of the Arnold-Chiari malformation without demonstrable bony anomaly.

CASE REPORT

History. A 5-year-old white female was seen in consultation on April 28, 1947. She was the first born of the family. Her birth and developmental history were entirely normal, and she had had none of the usual childhood diseases. In November 1946, the mother first became aware of the fact that the child was less active than usual, that she appeared listless and preferred not to run and play as she had formerly. She offered no specific complaints other than that her head ached and that exercise caused pain in her neck. By December 1946 such general lassitude and weakness of the lower extremities had developed that her schooling was discontinued. It was then that the mother noted a change in the child’s gait, described as a tendency to drag her feet when walking. Following tonsillectomy in February 1947 there was noted for the first time a weakness in the upper extremities similar to that seen in the lower. In the latter part of March, 1947 the child had to be confined to bed because of profound weakness of all extremities. She was unable to elevate her arms above the shoulder level. She could not hold her head erect nor was she able to hold her body erect upon attempting to stand upright. Two weeks before the initial examination the child had, on two occasions, regurgitated liquids through her nose. At about this same time there developed huskiness of her voice. This illness had not been attended by fever; she had not complained of any specific pain and had not experienced any cranial nerve disturbances other than the dysphagia.

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Neurological Examination. The appearance of this child was similar to that of an adult in an advanced stage of myasthenia gravis. She was carried into the examining room by her father, and when placed in the erect position she immediately sagged against the support of his arms, the knees flexed and were adducted, the arms hung limply by her side, and the chin rested against the chest. When she attempted to look at the examiner she did so by elevating her eyes and lids but not her head. She breathed through her mouth with her tongue partially protruded, and there was a salivary drooling. Her breathing was rapid and labored and was performed entirely by the intercostal muscles. When held in an upright position she was able to offer some support of herself on her toes and heels. The handgrips and finger movements were likewise weak. Individual muscle tests revealed that the most marked handicaps were apparently confined to the group supplied by the upper and middle cervical cord. There were no fibrillary twitchings noted and the only demonstrable muscle atrophy was confined to the intrinsic muscles of the left hand. This produced a modified ulnar type of deformity. There was no involuntary nuchal rigidity, but she complained of pain when her head was moved.

Examination of the vertebral column revealed no palpable deformities of the cervical spine, and there were no findings to suggest a spina bifida at a lower level. The cranial nerve study was not noteworthy aside from obvious palate and tongue weakness which was not attended by atrophy or fibrillar twitching of the tongue. The sensory examination was entirely normal for all modalities. There were no abnormal cerebellar objective evidences. Reflex studies elicited brisk triceps responses bilaterally, but the biceps reflexes and the abdominal reflexes were absent. There was a marked exaggeration of the knee and ankle reflexes. Transient ankle clonus was present and the Babinski reflex was bilaterally positive.

Course in Hospital. The child was immediately hospitalized with a presumptive diagnosis of an infiltrating tumor of the upper cervical spinal cord and medulla oblongata. Roentgenograms of the spine revealed no evidence of a congenital anomaly. There were no suggestions of a widening of the interpedicular measurements, laminal erosions or traumatic damages; nor did X-rays of the skull depict abnormalities. A lumbar puncture was attempted and although the operator was certain the needle was within the canal, clear fluid was obtained only by resorting to abdominal compression, and the CSF pressure was recorded at 30 mm. of water. Fluid sufficient for a protein determination was finally removed and 1 cc. of lipiodol was then introduced. The protein was reported as 63 mg. per cent, the cell count 0. An attempt was made to fluoroscope the movement of the lipiodol but it was found to be immobile, apparently having been introduced into the subdural space. A 2nd attempt was made to determine the CSP dynamics, with similar results except that on the 2nd occasion jugular compression did increase the flow of the spinal fluid measurably.

At this point a neurological consultant confirmed the previous findings and corroborated the probable diagnosis of glioma of the brain stem and cervical cord. A trial of roentgen therapy was agreed upon, inasmuch as there had been a distinct improvement clinically. Even before changes secondary to the X-ray therapy could be expected, the child became more active, was able to engage in minor activities on the ward and no longer exhibited the positive Babinski reflexes. At the completion of the course of roentgen ray therapy she was ambulant with increasing power, no longer exhibited difficulty in swallowing or breathing, and could elevate her arms above her head. She was discharged from the hospital to be observed at frequent intervals.
Subsequent Course. The patient was seen at frequent intervals during the next 6 months, and at each visit the picture presented was dissimilar. On one occasion she had reverted to the condition seen on the first examination, with the added finding of nystagmus on lateral gaze. On another occasion she appeared bright, sufficiently strong to walk unaided, and presented no very positive neurologic findings. During one of her more marked periods of regression, she was re-admitted for further CSF study. The manometric studies were entirely normal, and the protein was reported as 54 mg. per cent. A trial of prostigmine was given without beneficial effect and she was discharged definitely improved, this improvement lasting 1 month.

For a short period following her return home she improved markedly, to the extent that her parents considered sending her to school. Within 1 month, however, her condition became progressively worse. In addition to the original symptoms and the nystagmus on lateral gaze, there was paresis of the tongue and palate movements with complete areflexia in the upper extremities. There developed disturbing paroxysms of coughing due to swallowing difficulties. She was readmitted to the hospital for the 3rd time Nov. 15, 1947 for encephalography. This procedure was elected because of the technical difficulties encountered previously with the lipiodol studies and in the hope of visualizing the upper cervical and lower cranial spinal fluid pathways.

Encephalogram Report. With the patient under pentothal anesthesia and in an upright position, a lumbar puncture was performed. Fractionally, after considerable time had elapsed, a total of 20 cc. of clear fluid, containing 30 mg. per cent protein, was obtained, and 25 cc. of oxygen introduced. Routine skull roentgenograms as well as stereoscopic views of the cervical spine were obtained with the patient semi-erect on the table. There were only a few small gas shadows over the convexity of the brain, but none visualized in any of the ventricular cavities. On the cervical films the air column was seen to extend as high as the 2nd cervical level, at which point it appeared to be cut off. For a distance of 2 spinal segments below this point the column was distinctly reduced in width. The findings suggested an Arnold-Chiari deformity, and an exploratory operation was advised.

Operation. On Nov. 18, 1947, under intratracheal ether anesthetic, a combined suboccipital craniotomy and upper cervical laminectomy was carried out through a midline incision extending from the greater occipital protuberance to the level of the 4th spinous process. As the cervical arches were exposed, an anomalous, tremendously dilated and tortuous vein, apparently draining into the posterior internal vertebral venous plexus, was encountered. This vein lay over the 1st laminal arch, on the left, and after hemostasis of this large vessel an almost complete erosion of this arch was noted. This was in sharp contrast to the normal-appearing 1st arch on the right.

Following the removal of the 1st and 2nd cervical spinous processes and arches, a wide, firm fibrous band, which appeared to be a reduplication of the posterior occipito-atlantal ligament, was noted. This extended from the upper level of the 1st arch to the midpoint of the 2nd, and measured 3 mm. in width. After resecting this tough ligamentous band, a deep transverse notch was noted in the dura overlying the greater cistern. Freeing this constricting band did not result in reexpansion of the dura. Accordingly, the right posterior horn of the lateral ventricle was tapped and no increased ventricular pressure was noted nor did evacuating the fluid from this cerebral cavity improve the dural deformity.

The bone over the suboccipital region having already been removed, the dura was opened over both cerebellar hemispheres and as far inferiorly as the level of the
Fig. 1. Photograph of operative findings. (a) Occipital-cervical junction. (b) Caudally displaced and distorted cerebellar tonsils. (c) Third cervical level of spinal cord.
3rd cervical arch. This exposure revealed both cerebellar tonsils displaced caudally into the cervical canal to the level of the midportion of the 3rd cervical arch (Fig. 1). Both tonsils were literally glued together by thin arachnoidal adhesions which were easily separated. Both inferior cerebellar tips were yellow and atrophic in appearance. These structures were flattened to no more than 1 mm. thickness at their caudal ending. With the freeing of the arachnoidal adhesions and separation of the tonsils, there then occurred a brisk escape of clear fluid from the 4th ventricle. By retracting and separating the cerebellar tonsils, an adequate visualization of the 4th ventricle was afforded and it was noted that the lower portion of the 4th ventricle was elongated and displaced downward into the vertebral canal (Type II, Chiari).

Inspection of the site of junction of the medulla oblongata and the cervical cord disclosed a small area of cystic degeneration in the midline and a distinct cleft in the cervical cord corresponding to the level of the similar indenture noted in the dura. This cystic area corresponded to the lowermost portion of the 4th ventricle. Both the 1st and 2nd cervical nerves were observed to ascend obliquely to their exit from the spinal canal rather than to run in their usual transverse direction.

Inspection and palpation of the cerebellar hemispheres disclosed no unusual findings, and exploration of the cerebellopontine angles revealed no tumor. At this point, however, the explanation for the peculiar appearance of the cerebellar hemispheres became apparent: there was no structure present corresponding to the vermis. With a continuous escape of fluid from the 4th ventricle, it appeared obvious that no obstruction of the aqueduct existed.

Freeing the obstruction at the level of the greater cistern required removal of the 3rd cervical arch and amputation of both cerebellar tonsils. This procedure effected an adequate decompression, and the operation was terminated by a partial approximation of the dura over the cervical cord and cerebellar hemispheres, leaving that cistern open. The wound was meticulously closed in layers to prevent a cerebrospinal fistula, and the patient was returned to her room in excellent condition.

Postoperative Course. The course following surgery was uneventful. On the 1st postoperative day a spinal puncture was done to ascertain if a free communication of cerebrospinal fluid had been accomplished. It apparently had, as 20 cc. of slightly bloody fluid was removed. By the 3rd postoperative day the child was able to swallow liquids without difficulty, and our earlier fears of a respiratory catastrophe were relieved. The nystagmus and pathologic reflexes were not noted after the 4th day. When allowed up on the 6th day, the child was able to hold her head erect, to elevate her arms above her head, and to walk unaided, though she did so with a wide base. Upon discharge from the hospital on the 10th postoperative day she was able to dress herself, to walk unassisted, and was eating solid foods without regurgitation.

When last seen 1 year after operation, the child had improved markedly. She had regained full motor power and coordination, no longer complained of any discomforts and had been able to engage in the physical activities of her schoolmates without limitation. The only positive neurologic findings consisted of persisting areflexia in the upper extremities and hyper-reflexia in the lower extremities without pathologic toe signs. There was still persisting evidence of minor atrophies of the left hand, but the handgrip and finger movements were strong.

DISCUSSION

Many authors in discussing the Arnold-Chiari malformation have stressed the combination of the Chiari deformity and the usually described
bony lesions, pointing out the necessary accompaniment of the latter in order to conform to the Arnold-Chiari syndrome. In 1938, McConnell and Parker reported 5 cases of the Arnold-Chiari malformation. The youngest patient was 10 years of age, the oldest 32. In none of these patients was there demonstrable evidence of a spina bifida deformity, but all had an associated internal hydrocephalus. No mention was made by these authors of cervical spine pathology. In the same year, Aring reported a single case in an adult without vertebral changes, and suggested that his case might represent a variant of a congenital spinocerebellar ataxia. Adams, Schatzki and Scoville reported 2 instances of this malformation, 1 in a 17-year-old boy without demonstrable evidences of a spina bifida, and the 2nd in a 13-year-old boy who had had a myelomeningocele repaired at birth. Similarly Penfield and Co-burn have recorded a case of a 29-year-old woman in whom symptoms due to this anomaly did not appear until 26 years after the repair of a dorsal myelomeningocele. These 2 cases illustrate the latent period of development of symptoms which may follow repair of a spina bifida and support the theory of Russell and others that the Arnold-Chiari malformation may develop in association with a spina bifida. This deformity may occur late in life without demonstrable cause, as evidenced by the case reported by Bucy and Lichtenstein. In this instance, a 40-year-old negress without demonstrable bone pathology or cause for the lesion was found to have the classical Arnold-Chiari deformity. Ogryzlo reported 3 adults, operated upon by Dr. McKenzie, who had this anomaly yet did not present vertebral changes. In 2 instances a preoperative diagnosis of congenital herniation of the cerebellum had been made.

Most authors have stressed the fact that an internal hydrocephalus accompanies this malformation, and some have expressed the belief that the hydrocephalus may be responsible for the deformity. In the case reported here, even though clinical evidence was present intermittently, and even though at operation there appeared a complete block of the cerebrospinal fluid at the cisterna magna, a hydrocephalus from ventricular estimation was not present. Although increased intracranial pressure may produce the so-called cerebellar pressure cone, the extreme prolapse to the degree frequently encountered in this malformation is rarely seen in the usual hydrocephalus.

The Arnold-Chiari malformation is apparently a congenital deformity and may be associated with a wide variety of other anomalies in addition to the bony deformity. The incomplete development of the cerebellar vermis has repeatedly been encountered, as in the case reported here. In Case V of McConnell and Parker, there was a congenital absence of the foramina of Magendie and Luschka. Lichtenstein reported an associated aqueductal stenosis.

The etiology of the malformation in the presence of spina bifida has apparently been well established as due to the traction force exerted against the medulla and cerebellar tonsils with fixation of the spinal cord at the site
of the spina bifida. With continued elongation of the vertebral column, these structures are pulled downward through the foramen magnum. As Bucy and Lichtenstein have suggested, this same traction force may produce the malformation in the absence of firm fixation of the brain in the cranial cavity. This latter supposition has not been confirmed by anatomic studies. The Arnold-Chiari malformation therefore may occur in association with a number of other congenital anomalies and need not in each instance be combined with either a spina bifida or other cranio-vertebral deformities.

**SUMMARY**

1. A case of Arnold-Chiari malformation without demonstrable spina bifida, hydrocephalus or other cranio-vertebral anomalies is reported.

2. Attention is called to the fact that this malformation may occur in association with a number of congenital anomalies and need not in each instance be combined with spina bifida, hydrocephalus or cranio-vertebral deformities.

3. This malformation should be considered in evaluating acquired pyramidal tract disturbances.

**REFERENCES**


