ANATOMIC ANOMALIES COMMON TO MYELOMENINGOCELE OF INFANCY AND SYRINGOMYELIA OF ADULTHOOD SUGGEST A COMMON ORIGIN

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THE fact that an infant with myelomeningocele may have a syrinx in association with the Arnold-Chiari malformation,[1,2] together with the fact that this same association occurs in the adult with syringomyelia[3-6] led to a search of the literature for other anatomic anomalies shared by the infant with myelomeningocele and the adult with syringomyelia. This search revealed that each may have hydrocephalus, hydromyelia (dilatation of the central canal), syringomyelia (syrinx or cavity paralleling the central canal), Arnold-Chiari malformation, Dandy-Walker malformation, membranes enclosing the foramens of the fourth ventricle, glial heterotopia, and skeletal anomalies—scoliosis, hemivertebra, fused vertebrae, anomalous ribs, deformity of the feet, enlargement of the skull and of the vertebral canal.

Hydrocephalus

In most cases of myelomeningocele there is increased intracranial pressure and progressive dilatation of the ventricles, indicating an uncompensated hydrocephalus. This hydrocephalus may be of the communicating or of the noncommunicating variety.[7] It is not widely known that syringomyelia in the adult may be accompanied by dilated ventricles but with normal intracranial pressure, i.e., compensated hydrocephalus. Netsky[8] found dilatation of the ventricles in three of four cases of syringomyelia in which the brain was examined at necropsy *(Fig. 1)*. Encephalography likewise will show that the ventricles frequently are dilated in patients with syringomyelia and that they may or may not communicate with the subarachnoid space.[3-5] Thus, we find that hydrocephalus occurs both in the infant with myelomeningocele, and in the adult with syringomyelia, and that it may be communicating or noncommunicating. The conditions differ in that the hydrocephalus in the infant usually is severe and uncompensated, whereas in the adult it is mild and compensated.

Hydromyelia and Syringomyelia

In 1876, Leyden[9] stated his belief that syringomyelia in the adult is a "rest" of a congenital hydromyelia that "cuts itself off" from the central canal. Recently Greenfield[10] also has pointed out that there is a link between hydromyelia and syringomyelia, and stated that cases are apt to be assigned to one or the other category according to whether or not the cavitation has produced the typical

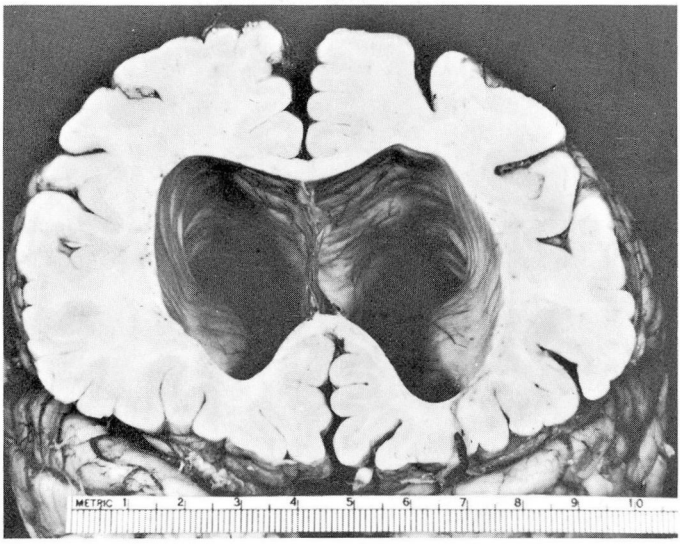


Fig. 1. Hydrocephalus in a woman of 47 years with symptomatic syringomyelia for 15 years. The cord was cystic from its junction with the medulla to its sacral portion. (Courtesy of Netsky, M.G.: A.M.A. Arch. Neurol. & Psychiat. 70: 741-777, Dec. 1953.)

symptoms of syringomyelia during life. Netsky[8] among others has shown that syringomyelia in the adult may constitute merely a diverticulum of a hydromyelia so that both conditions coexist.

The infant with myelomeningocele usually has hydromyelia *(Fig. 2)*,[2,10-12] and in addition may have a true syrinx. Lichtenstein[1] described a case in which a syrinx communicated with the central canal of the cord near its juncture with the fourth ventricle *(Fig. 3)*. In a 26-week-old infant with myelomeningocele, Cameron[2] observed a syrinx that extended from the level of the sixth cervical vertebra to the third thoracic vertebra and measured 0.4 cm. in maximum diameter. At its upper end only it communicated with the hydromyelic central canal. Therefore, the infant with myelomeningocele, as well as the adult with clinical syringomyelia, may have hydromyelia and a communicating syrinx.

Because the ventricles and the central canal constitute a single cavity, the term "hydrocephalomyelia" has been proposed[3] to describe the state in which both are dilated.

Arnold-Chiari Malformation

Chiari,[13] in 1891, gave a complete description of three types of deformity of the hindbrain caused by hydrocephalus of the forebrain. Despite the fact that this antedated Arnold's[14] inadequate description of the deformity, his pupils Schwalbe

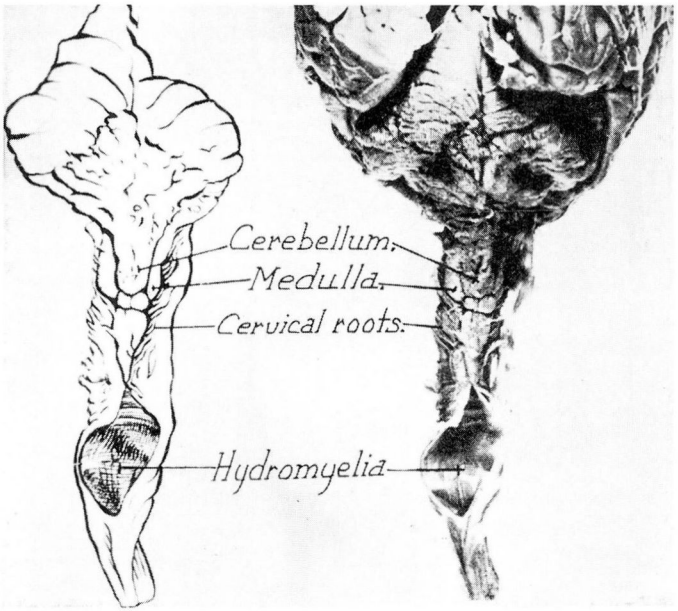


Fig. 2. Hydromyelia in an infant with myelomeningocele. (Courtesy of D'Errico, A.: South. M. J. 35: 247-252, March 1942.)

and Gredig[15] subsequently entitled Chiari's type 2 the "Arnold-Chiari malformation." In his more commonly quoted second paper, Chiari[16] described 14 cases of his type-1 deformity, all in adults or in adolescents, only one of whom had myelomeningocele. He also described seven cases of his type-2 deformity, all in infants with myelomeningocele. In type 1 the herniation included the cerebellar tonsils, but not the fourth ventricle, a deformity now frequently referred to as "pressure coning." In type 2 the more pronounced herniation included the vermis and the fourth ventricle as well, with an associated posterior bulging of the medulla, and because of the accompanying caudal dislocation of the upper cervical cord, the emerging nerve roots pursued a cephalad course to their points of exit. Chiari believed that type 2 was merely a severe form of type 1, and that both were caused by hydrocephalus of the forebrain. He found hydromyelia in both types.*

In 17 instances of the "Arnold-Chiari malformation" in adolescents and adults described by Gardner and Goodall,[3] none were associated with myelomeningocele. There was a syringomyelic type of sensory loss in 12 patients, and a cystic cervical cord was demonstrated at operation in 10 of these 12 as well as in three others who did not present the clinical picture of syringomyelia. These 17 hindbrain malformations resembled Chiari's type 1, in that the cerebellar hernia consisted of

**Chiari's type 3 consisted of a herniation of the hindbrain through the bony defect of a cervical spina bifida.*

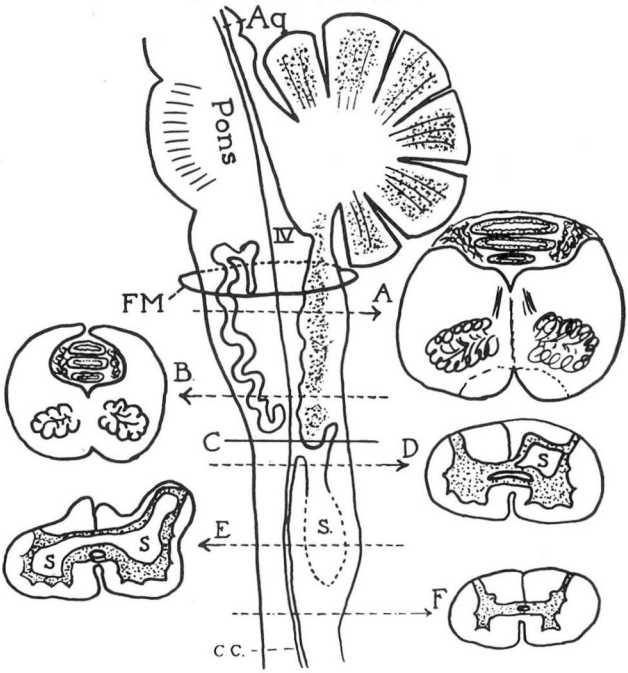


Fig. 3. Schematic drawing of a midsagittal section through the hindbrain and upper cervical cord of an infant with hydrocephalus, Arnold-Chiari malformation, and myelomeningocele. FM indicates the foramen magnum, S the syrinx, c.c. the central canal, and C the level of transection at necropsy. Note the absence of the foramen of Magendie. (Courtesy of Lichtenstein, B. W.: Arch. Neurol. & Psychiat. 49: 881-894, June 1943; A.M.A. Arch. Neurol. & Psychiat.)

the tonsils, but in some there was herniation of the fourth ventricle as well, a feature that Chiari included under his type 2 *(Fig. 4).*

Thus we see that the Chiari type-1 malformation in the adult with syringomyelia may have some of the features of Chiari's type-2 deformity of infancy. Conversely, the infant born with a mild myelomeningocele may exhibit Chiari's type-1 deformity, as was illustrated by Chiari's[16] own experience mentioned above, and also by case 19 of Russell.[17] Therefore, although the Chiari malformation present in the adult with syringomyelia usually is type 1 and that in the infant with myelomeningocele usually is type 2, nevertheless, transitional forms do occur both in the adult and in the infant. This intermingling of the features of the two types lends support to Chiari's conviction that type 2 is merely a severe form of type 1.

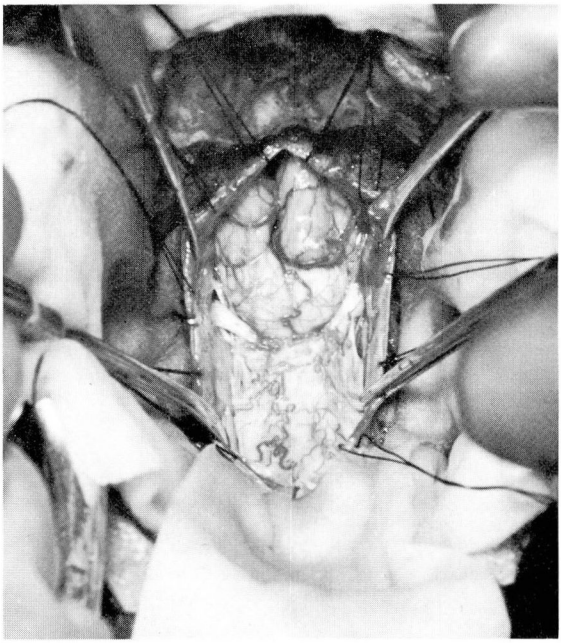


Fig. 4. Herniation of the cerebellar tonsils, posterior bulging of the medulla, and upward-slanting cervical nerve roots in a patient with syringomyelia. This represents a transitional form between Chiari's type-1 and type-2 malformations. (Courtesy of Gardner, W. J.; Abdullah, A. F., and McCormack, L. J.: J. Neurosurg. 14: 591-607, Nov. 1957.)

Dandy-Walker Malformation

It is not generally recognized that an infant with myelomeningocele may prove to have an enormous dilatation of the fourth ventricle instead of the anticipated Arnold-Chiari malformation *(Fig. 5)*. Dandy[18] showed a photograph of such a patient, and Benda[19] also described this association. In Benda's patient, the fourth ventricle formed a huge sac that, together with a separation of the cerebellar hemispheres, constituted his "Dandy-Walker syndrome." Examination of the spinal cord of this infant "revealed myelomeningocele and congenital syringomyelia of the hydromyelic type . . ."

When encountered in the adult, the Dandy-Walker malformation is mild and may be accompanied by the typical clinical picture and operative findings of syringomyelia as described by Gardner, Abdullah, and McCormack.[4] Thus, the Dandy-Walker malformation has been encountered both in the infant with myelomeningocele and in the adult with clinical syringomyelia.

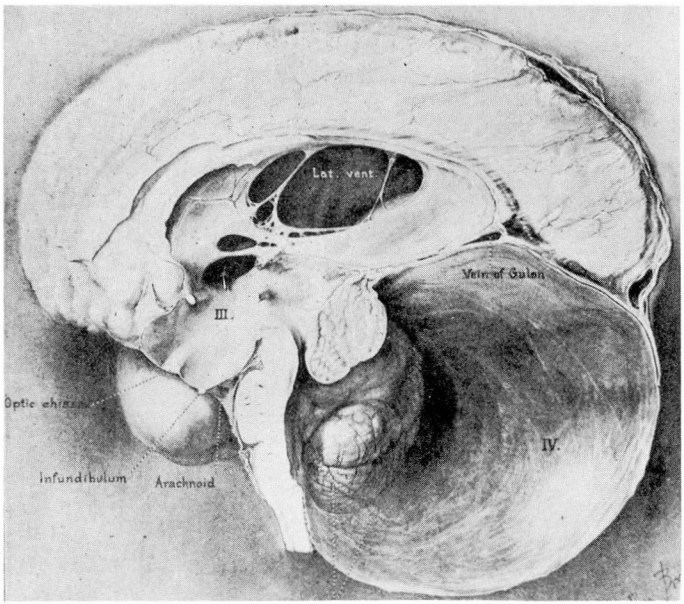


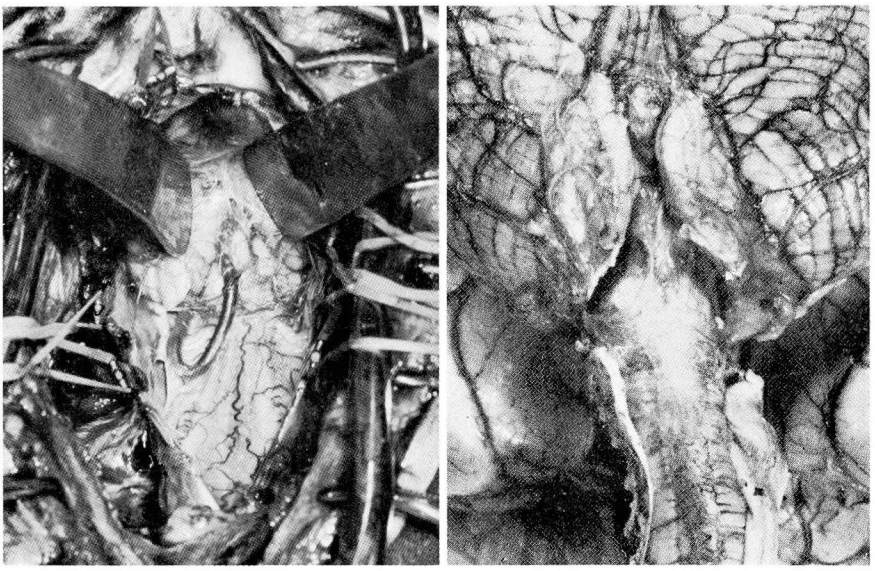
Fig. 5. Sagittal section through the brain of an infant with hydrocephalus due to congenital occlusion of the foramens of Luschka and Magendie. The enormously dilated fourth ventricle constitutes Benda's[19] "Dandy-Walker Syndrome." (Courtesy of Surg. Gynec. & Obst.: Dandy, W. E.: Surg. Gynec. & Obst. 32: 112-124, Feb. 1921.)

Membranes Enclosing the Foramens of the Fourth Ventricle

Membranes enclosing the distended foramens of a dilated fourth ventricle are the diagnostic feature of the Dandy-Walker malformation whether encountered in the infant with myelomeningocele or in the adult with syringomyelia. The attachments and the microscopic appearance of these membranes indicate that they represent persisting portions of the embryonic rhombic roof.[20] It is not generally known that similar membranes may enclose these foramens in the Arnold-Chiari malformation as well, both in the infant with myelomeningocele and in the adult with syringomyelia. When present in the Chiari type-1 deformity, either in an adult (case 3 of Gardner, Abdullah, and McCormack[4]) or in an infant (case 19 of Russell[17]) the membranes are readily demonstrable *(Fig. 6, A and B)*. In the Chiari type-2 malformation, the foramens of Luschka are concealed in the herniated mass compressed within the spinal canal. A sagittal section in some cases, however, will clearly show a membrane enclosing the foramen of Magendie *(Fig. 7)*.

Glial Heterotopia

Most authors in discussing myelomeningocele have described an associated glial heterotopia. Cooper and Kernohan[21] found extruded heterotopic glial nests



A B

Fig. 6. A, Membrane obstructing the foramen of Magendie in a Chiari type-1 malformation in an adult with syringomyelia. B, Similar finding in an infant with a Chiari type-1 malformation, a lumbosacral meningocele, and a tethered cord. Each foramen of Luschka also was closed by a bulging membrane in this case. (Courtesy of Russell, D. S.: M. Res. Council Special Rep. Ser. No. 265; H. M. Stationery Office, 1949, 138 pp.; the Controller, H. M. Stationery Office, 1959.)

in the meninges in a series of 50 necropsies, in 34 of which there were associated congenital anomalies of the nervous system, the most common ones being: spina bifida, hydrocephalus, syringomyelia, and hydromyelia. Gardner, Abdullah, and McCormack[4] described foci of glial heterotopia in the leptomeninges of the pons, the medulla, and the spinal cord of an adult with syringomyelia. Glial heterotopia, therefore, is another anatomic anomaly that is present in the infant with myelomeningocele and in the adult with syringomyelia.

Skeletal Anomalies

In association with syringomyelia, skeletal anomalies such as scoliosis, hemivertebra, fused vertebrae and cervical ribs have been described. Gardner and Goodall[3] found scoliosis in eight of 17 patients, fused vertebrae in two patients, and hemivertebra in one patient. Similar but more severe skeletal anomalies are found in infants with myelomeningocele. Cameron,[2] in 26 such infants, found kyphoscoliosis in nine, sometimes associated with multiple hemivertebrae; and there were absent, deformed or supernumerary ribs in four patients. Fused vertebrae are difficult to recognize in infants, because ossification is incomplete, but Ingraham and Scott[11] found fusion associated with myelomeningocele, as did Feller

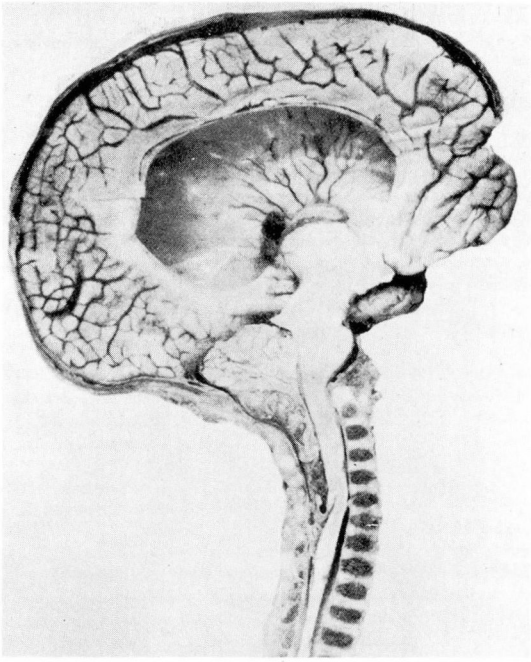


Fig. 7. Membrane obstructing the foramen of Magendie in an infant with myelomeningocele and Chiari type-2 malformation. (Compare the attachment of the membrane at the obex to that in *Figure 5.*) (Courtesy of Moncrieff, A., and Norman, A. P.: Greenfield, J. G.: *Neuropathology:* Edward Arnold (Publishers) Ltd., 1958, 640 pp.)

and Sternberg.[22] Clubfoot is encountered frequently in the infant; whereas, in the adult with syringomyelia, pes cavus is a common finding.

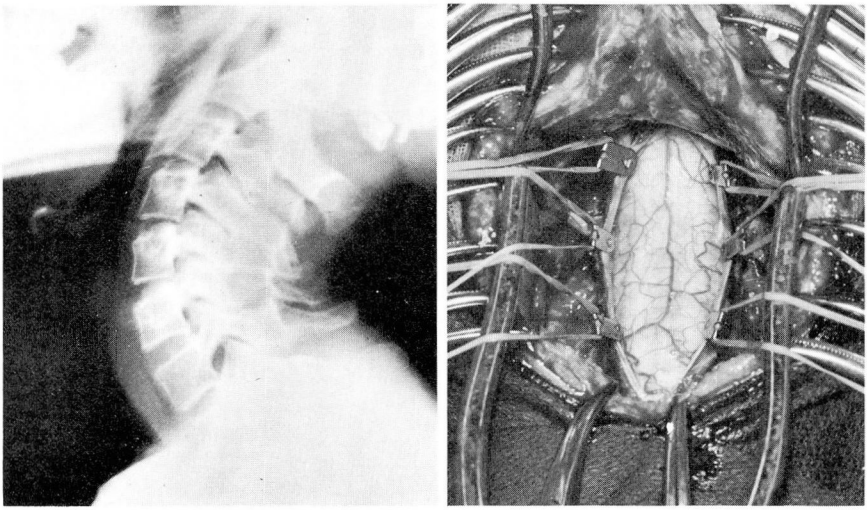
Expansion of the Cranio-Vertebral Axis

In the infant with myelomeningocele the cranial vault usually is enlarged. The same may be true of the adult with syringomyelia, although the enlargement is less frequently observed and less severe. Netsky[8] described a patient with syringomyelia who had a large brachycephalic skull and evidence, on roentgenograms, of long-standing internal hydrocephalus. Gardner, Abdullah, and McCormack[4] also found this in one of their patients.

Dilatation of the lumbar canal at the level of the spina bifida is always present in infants with myelomeningocele. There may be an associated dilatation of the cervical portion of the canal as was described by Feller and Sternberg[22] in their case of lumbar myelomeningocele mentioned above.

I have observed dilatation of the cervical portion of the vertebral canal in a 16-year-old boy with syringomyelia, and also in an 11-year-old girl *(Fig. 8, A and*

B). This increased size of the lumen of the bony spine and skull obviously is not due to a primary defect in osteogenesis, but represents an expansion secondary to increased intraluminal pressure.



A B

Fig. 8. A, Dilatation of the vertebral canal of an 11-year-old girl with syringomyelia. B, Same case at operation; a syrinx always is distended in this fashion in the living subject as compared to its usual collapsed state at necropsy. The distention of the cervical cord in this case appears to have been responsible for the dilatation of the vertebral canal just as the distention of the ventricles in congenital hydrocephalus is responsible for the enlargement of the cranial vault.

Discussion

The neural tube of the embryo is a closed cavity consisting of the primitive ventricles and the central canal of the cord. Its lumen is extremely large compared to the thickness of its walls, thus constituting physiologic hydrocephalus and hydromyelia, or hydrocephalomyelia. A study of Weed's[23] reproductions of pig embryos, shows that this distention of the neural tube reaches its most advanced stage shortly after the appearance of the choroid plexus *(Fig. 9).* It then subsequently diminishes, i.e., the hydrocephalomyelia becomes compensated as the rhombic roof and the subarachnoid space become more permeable. The opening of the foramens of the fourth ventricle in fetal life permits, for the first time, perfectly free communication between the ventricles and the subarachnoid space.

The beating of the choroid plexus, which is a relatively enormous structure in the embryo *(Fig. 10),*[24] acts as an unvalved pump to generate a pulse wave in the ventricular fluid.[25] This ventricular fluid pulse wave, funneled into the central canal in embryonal life, is shunted into the subarachnoid space after the foramens of the fourth ventricle open. The bypassed central canal, then com-

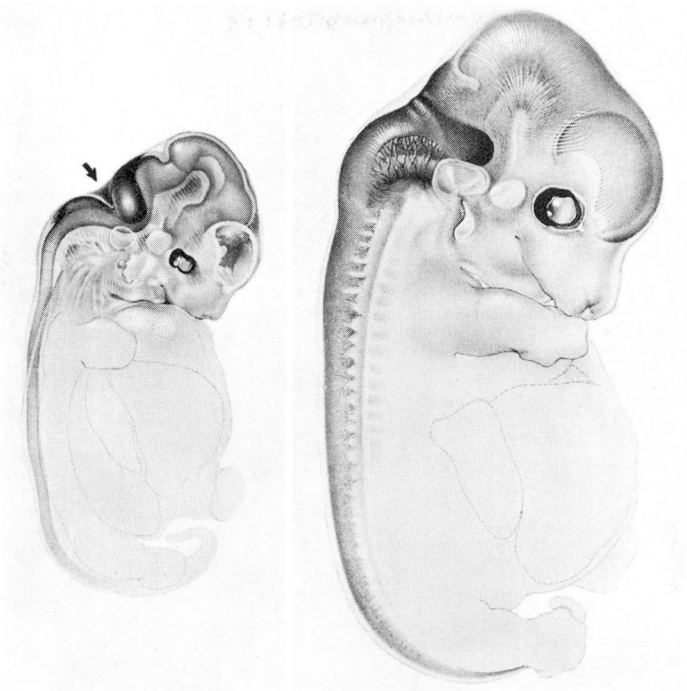


Fig. 9. An 18-mm. and a 26-mm. pig embryo with the cerebrospinal fluid spaces outlined by precipitated crystals of Prussian blue. In the 18-mm. embryo the ventricular fluid is just beginning to push through the permeable rhombic roof (arrow). In the 26-mm. embryo, as a result of dissection by the extruded fluid, the subarachnoid spaces have expanded to their adult configuration (L. H. Weed[23]). While the crown-rump length and the anteroposterior diameter of the embryo have increased less than 50 per cent, the lumen of the neural tube by comparison has enlarged tremendously. Hydrocephalomyelia, therefore, is a normal state in embryonal development. (Courtesy of The Carnegie Institution of Washington: Weed, L. H.: Contributions to Embryology, Vol. V, No. 14, Inst. Pub. No. 225, 1917, 116 pp.)

pressed from without and containing no choroid plexus, subsequently narrows to become a vestigial structure *(Fig. 11, A and B)*. The central canal may enlarge in postnatal life if the foramens of the fourth ventricle become occluded so that ventricular fluid once more can distend it. This is shown by the development of hydrocephalus, hydromyelia, and syringomyelia when the foramens are sealed experimentally by reaction to the intracisternal injection of kaolin[26] *(Fig. 12)*.

It was Chiari's belief that the deformity of the hindbrain which he described is the result of hydrocephalus of the forebrain. Weed[23] has pointed out that should the rhombic roof not be adequately permeable in embryonic life, obstructive hydrocephalus will ensue which in turn may result in communicating

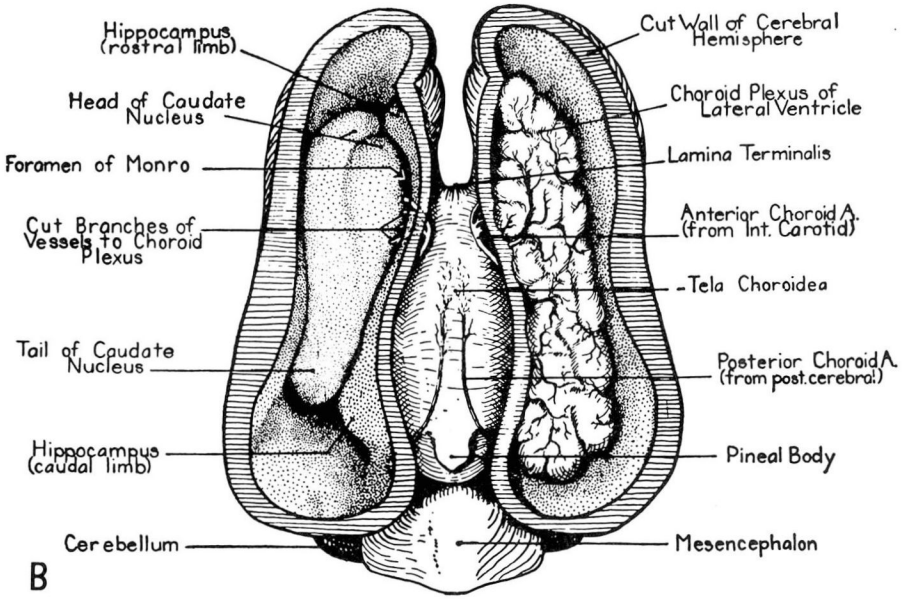


Fig. 10. The relatively enormous size of the choroid plexus and of the lateral ventricle is well shown in this diagram of the brain of a three-month human fetus cut horizontally. (Courtesy of Patten, B. M.: *Human Embryology;* modified after His; McGraw-Hill Book Company, Inc., 1946, 776 pp.)

hydrocephalus. Since in postnatal life hydrocephalus due to obstruction of the fourth ventricle results in a herniation of the hindbrain through the foramen magnum, the same thing may be anticipated in prenatal life—indeed more readily, since the immature dentate ligaments offer little resistance to the caudal dislocation of the hindbrain.*

In obstructive embryonal hydrocephalus, the central canal of the cord takes part in the dilatation. This cavity, normally a slit, is thus converted into a distended cylindrical tube that displaces and distorts the sclerotomes on each side of it *(Fig. 13).* A distorted sclerotome can scarcely be expected to develop into a normal bone. A pair of sclerotomes separated from each other by a distended neural tube may fail to unite, resulting in bilateral hemivertebra.

**The reason why the fourth ventricle is compressed in the Arnold-Chiari malformation and dilated in the Dandy-Walker malformation will be dealt with in a subsequent communication.*

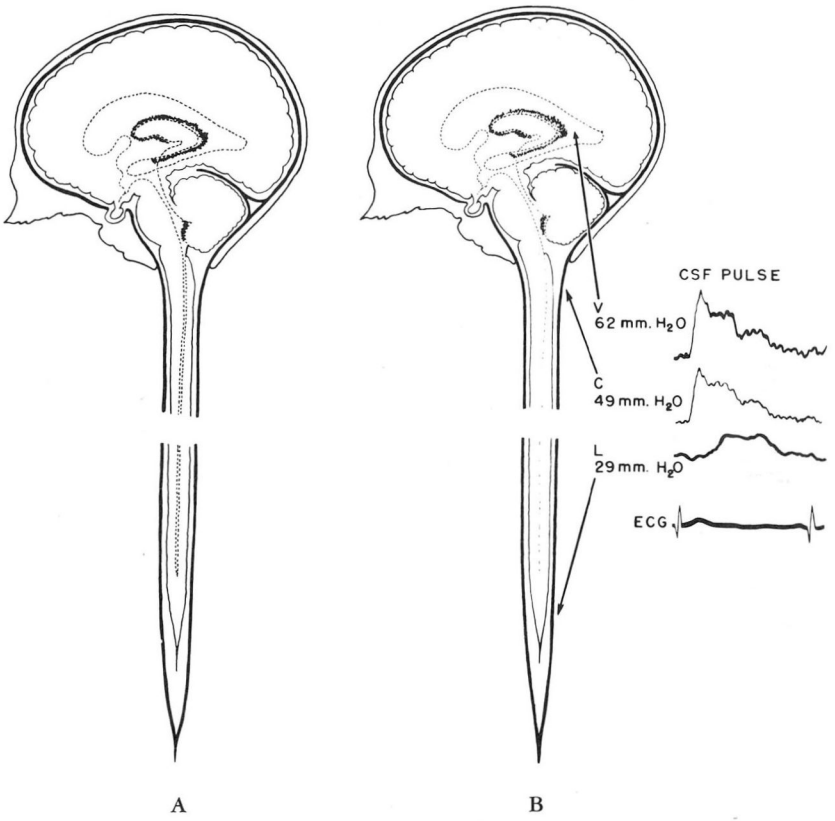


Fig. 11. A, Prior to the opening of the foramens of the fourth ventricle, the central canal of the cord, distended by ventricular fluid, constitutes an offshoot of the ventricular system. B, After the foramens open, the pulse wave of the ventricular fluid is shunted into the expansible spinal subarachnoid space on the outer surface of the cord and the central canal gradually narrows. The figures indicating the height of the pulse waves at various levels are from Bering.[25]

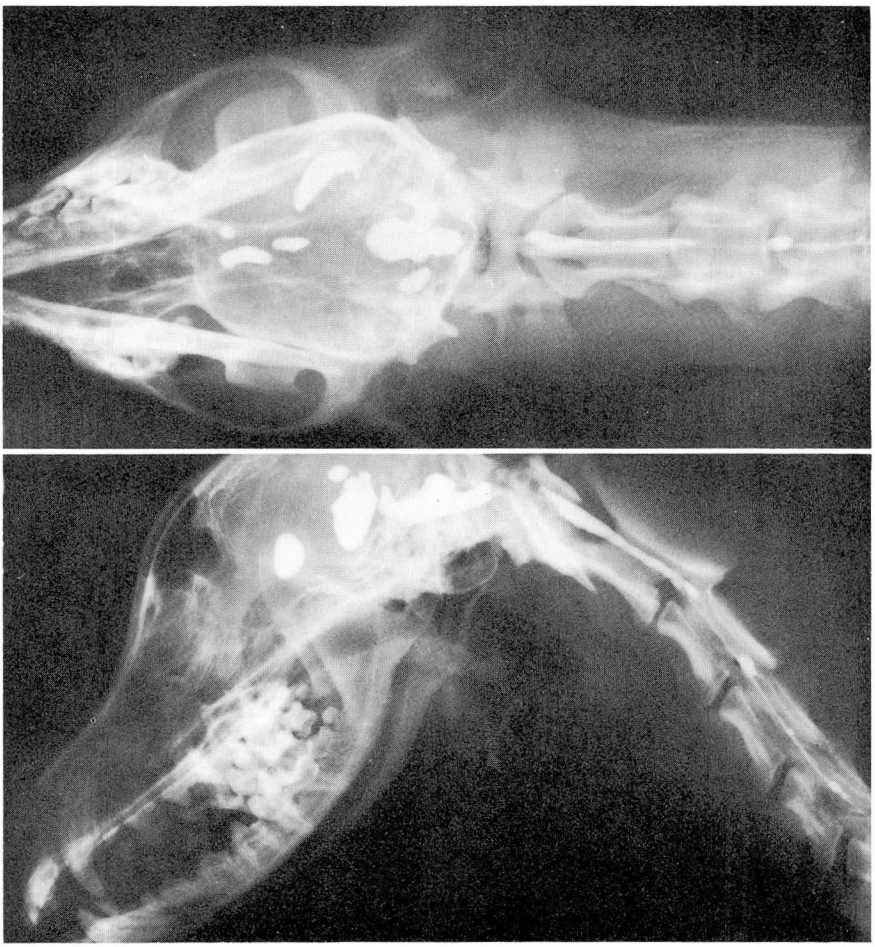


Fig. 12. In experimental hydrocephalus, injection of Pantopaque into the obstructed fourth ventricle shows that it communicates with the dilated central canal. This represents acquired hydrocephalomyelia. (Courtesy of McLaurin, R. L.; Bailey, O. T.; Schurr, P. H., and Ingraham, F. D.: A.M.A. Arch. Path. 57: 138-146, Feb. 1954.)

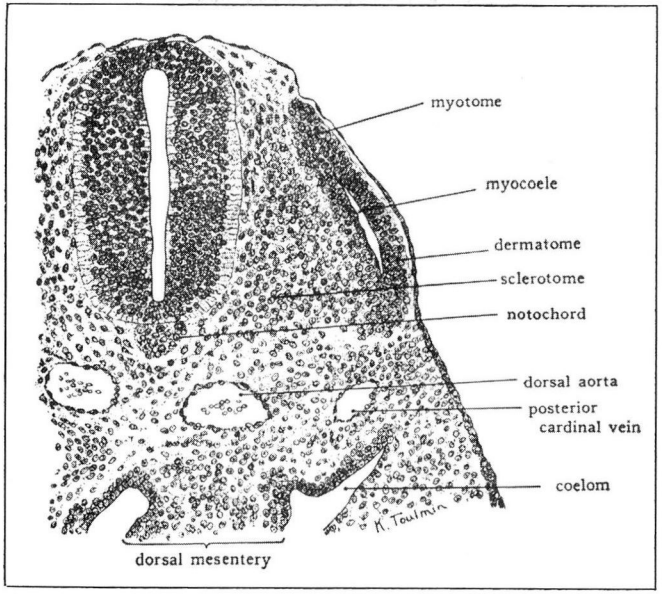


Fig. 13. Drawing from a transverse section of a 30-somite pig embryo. Note the slitlike neural tube bordered on each side by the skeletal anlagen. (Courtesy of Patten, B. M.:*Human Embryology;* modified after His; McGraw-Hill Book Company, Inc., 1946. 776 pp.)

Summary

The several anatomic anomalies, shared by the infant with myelomeningocele and the adult with syringomyelia, support the belief of earlier writers that there is a relationship between these states.

Hydrocephalomyelia is present in the embryo prior to the opening of the foramens of the fourth ventricle. In postnatal life it is found in cases where these foramens have failed to open adequately, as in the infant with myelomeningocele, and in the adult with syringomyelia. This suggests that in the latter conditions, the embryonal state of hydrocephalomyelia has been preserved by the inadequate permeability of the foramens of the fourth ventricle; that a normal individual results when embryonal hydrocephalomyelia becomes compensated at the normal time; that syringomyelia results when it becomes compensated too late; that myelomeningocele results when it does not become compensated at all.

This hydrodynamic theory will explain not only the anomalies of the central nervous system common to these states but the anomalies of the skeleton as well.

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