Myelocystocele with cerebellar heterotopia

Case report

ANDERS SUNESON, M.D., AND HANNU KALIMO, M.D.

Department of Neurosurgery, Södersjukhuset, Stockholm, and Division of Neuropathology, Department of Pathology, University of Uppsala, Uppsala, Sweden

The authors describe a case with well differentiated cerebellar tissue contained in a hydromyelic dilation of the spinal cord, that is, myelocystocele, in connection with a cervicothoracic meningocele. The embryology is discussed.

Key Words • congenital abnormality • myelocystocele • cerebellar heterotopia • meningocele

In 1907, Wolbach first described heterotopic glial nests in the subarachnoid space. Since then various cases of heterotopia within the central nervous system (CNS) have been reported, some of them in connection with other congenital malformations, such as myelomeningocele. In 1978, a case was described with heterotopic cerebrum, brain stem, and cerebellum located intracranially. Our case differs from all the others because it consists of a well differentiated heterotopic cerebellum situated within a hydromyelic dilation of the spinal cord, that is, it was a myelocystocele, in connection with a cervicothoracic meningocele.

Case Report

This 21-month-old boy was admitted for evaluation of a hairy swelling in the cervicothoracic region of the back with a preliminary diagnosis of a cervicothoracic myelomeningocele.

History. He was the youngest of four children born to healthy parents; his birth weight was 3610 gm. The elder sisters, brother, and parents had no signs of congenital disease. During pregnancy the mother took no drugs with the exception of diuretics for edema. No hypertension nor eclampsia was observed, nor any infection. The patient’s developmental milestones were normal up to the time of admittance. He sat at 7 months of age, walked at 1 year, and said a few sentences at admittance. Immediately after birth a little swelling in the cervicothoracic region was noticed. Later the parents noted that the swelling was slowly growing and had become hairy. An x-ray film of the back carried out before admittance revealed a spina bifida extending from C-4 to T-1.

Examination. The patient was a bright little boy without any pathological neurological findings except for a soft, fluctuating, hairy swelling, 2 × 2 cm in size, in the middle of his back in the cervicothoracic region. On the top of the swelling were two small darker patches with very thin skin. The swelling felt pulsatile on compression. Air myelography showed a juxtamedullary tumor at the back of the spinal cord, at C-4 to T-1, with well defined margins (Fig. 1). The preoperative diagnosis was a spina bifida with a dermoid cyst or a lipoma.

Operation. A longitudinal oval skin incision was made around the swelling, which was found to be entirely situated between the skin and muscle fascia. It was only a small stalk-like connection penetrating through the fascia in the midline. Cross section of the subcutaneous swelling revealed macroscopically that it consisted of a cystic structure filled with a yellowish opalescent fluid under some pressure. The fluid differed quite obviously from normal cerebrospinal fluid (CSF). No patent connection from the subcutaneous cyst through the stalk to underlying structures could be identified. Attempts to probe this stalk were made but were not successful. Just caudal to this stalk a little arachnoid cyst was protruding through the fascia and underlying muscle (Fig. 2 upper left). The muscle and its fascia were divided in the midline,
FIG. 1. Air myelography demonstrating the juxtamedullary tumor at the back of the spinal cord which was later proved to be a myelocystocele.

and a laminectomy of the two cervical arches (C2-3) immediately above the spina bifida was performed to expose the dura. The dura was quite normal except for some swelling of the whole dural sac in the spina bifida region and the two small openings in the dorsal midline through which the stalk and the little arachnoid cyst penetrated.

The dura and the underlying arachnoid, including the arachnoid cyst, were opened by a midline incision. Cerebrospinal fluid flowed from the subarachnoid space outside a hydromyelic dilation of the spinal cord. The above-mentioned stalk was attached to this hydromyelic dilation, inside which a round pedunculated tumor could be seen through the transparent dorsal wall of the dilation (Fig. 2 upper right). The thin dorsal wall was excised including the base of the stalk of the subcutaneous malformation. At this point the hydromyelic character of the intraspinal dilation could be verified. It was filled with clear water-like fluid of the same character as the CSF in the spinal subarachnoid space, but the only connection that could be found between the hydromyelic dilation and the normal CSF-filled spaces in the CNS was the central canal. Both the cranial and caudal openings to the normal parts of the central canal were identified and probed with a thin rubber catheter several centimeters upward and downward. There was no difference in fluid pressure between the CSF in the spinal subarachnoid space and the water-like fluid in the hydromyelic dilation. Arteries and veins running from the top of the round tumor to the right margin of the hydromyelic dilation were divided between clips. The round tumor was about 2 cm in diameter, grayish in color, and had a smooth surface. Its pedunculated base was attached to the ventral wall of the hydromyelic dilation (Fig. 2 lower). In the belief that the tumor was a cystic glioma of the spinal cord, we divided its peduncle near the base and the whole tumor was extirpated. The dura, muscle, and skin were closed in layers.

FIG. 2. Upper Left: Diagram of the subcutaneous malformation with its stalk penetrating through the muscle and its fascia in the midline. The arachnoid cyst is visible just caudally. Upper Right: The dura and the arachnoid opened between stitches, exposing the hydromyelic dilation of the spinal cord with contents. Lower Left: After the hydromyelic dilation was excised, the two openings into the central canal and the pedunculated miniature cerebellum were exposed. Lower Right: Schematic longitudinal midline section of the spinal cord with the hydromyelic dilation. The dilation contains the pedunculated tumor. On the top of the dilation is the base of the stalk leading to the subcutaneous malformation. The connection between the hydromyelic dilation and the normal parts of the central canal is also shown. Short dashes indicate the excision of the dorsal wall of the dilation.
Postoperative Course. Immediately after the operation the patient showed a markedly reduced tonus of the entire body without any distinct pareses. All reflexes of the extremities were present but were weak. A positive Babinski sign was present on both sides. No sphincter disturbances could be detected before or after the operation. During the following weeks he showed a marked recovery. Eleven days after the operation pneumoencephalography revealed a normal cerebellum in the posterior fossa with no signs of Arnold-Chiari malformation (Fig. 3). Four weeks after the operation the patient was discharged. At follow-up review 18 months postoperatively, the boy had no signs of neurological disturbances except for a persistent positive Babinski sign on the right side. He was able to sit, walk, and run normally.

Pathological Examination. The cystic structure beneath the skin was surrounded by meningeal cells and abundant collagenous fibers. The walls of the irregularly shaped cyst were composed of thick bands of aberrant glial tissue with occasional nerve cells, and they were partially lined by ependyma. A few structures resembling choroid plexus were also encountered (Fig. 4).

The pedunculated tumor removed from the hydromyelic dilation showed well differentiated cerebellar architecture; the folia were well developed and they were separated from each other by sulci of variable depth (Fig. 5). Microscopically the cortical structure was practically normal. The molecular and granular layers were in general similar to those usually seen in normal cerebellum at this age. At the junction of these layers there were numerous Purkinje cells. These typically had a large nucleolus and abundant cytoplasm which contained much Nissl substance and formed distinct neuronal processes (Fig. 6 left). The granular cell layer was composed of normal tightly packed small neurons with scanty cytoplasm. A rich network of silver-positive neuronal processes was visible between the granular cells (Fig. 6 center). The external granular layer was lacking, as it is in the normal cerebellum at this age. In the center of the folia, as well as in the central areas, many myelinated fibers were seen, but their number was, however, clearly less than in a normal cerebellum (Fig. 6 left). Remarkably, a collection of neurons, evidently equivalent to the dentate nucleus, was seen in the central white matter (Fig. 6 right).
Myelocystocele with cerebellar heterotopia

Fig. 5. Heterotopic cerebellar tissue, depicting the formation of folia and sulci, and a good cortical differentiation into layers. Arrow points to the collection of neurons equivalent to the dentate nucleus. H & E. Bar: 4 mm.

Fig. 6. Photomicrographs of the cerebellar tissue. Left: The cortical differentiation into the molecular and granular layers is practically authentic. Three of the Purkinje cells at the junction of the layers appear normal, whereas three others show degenerative changes. The granular layer is composed of tightly packed small neurons, and a few myelinated fibers (arrow) lie in the center of the folium. LFB-cresyl violet. Bar: 50 μm. Center: The network of neuronal processes is illustrated by silver staining. Bar: 50 μm. Right: In the sparsely myelinated white matter a collection of neurons is visible that is evidently an equivalent of the dentate nucleus. LFB-cresyl violet. Bar: 100 μm.
Discussion

The cerebellum normally develops from the alar plates of the mesencephalon to be located within the posterior fossa overlying the fourth ventricle. The pathogenetic mechanisms that led to the development of a heterotopic miniature cerebellum on the posterior aspect of the basal plates in the lower cervical cord can only be speculated upon. A faulty inductive stimulus on the undifferentiated cells of the fetal mantle layer is a possible explanation, but very little is known about the process of induction in the development of the normal cerebellum.

Different forms of neuroectodermal heterotopias have been described to occur within the CNS and the leptomeningeal space. Most of these cases have been collections of neural or glial cells, the differentiation of which has not at all been comparable to that seen in our case. The malformation that best corresponds to our case was described by Billings and Danziger; that was a heterotopic cerebellum of normal infantile structure protruding into the fourth ventricle of a 9-month-old boy. In both cases the heterotopia lay within the ventricular system. In two other malformations with differentiated cerebellar heterotopia, the tissue was located within a separate arachnoidal cyst outside the parenchyma itself. In the recent report by Marubayashi and Matsukado, an intracranial, extracerebral brain heterotopia was described; in that case the malformation consisted also of cerebrum and brain stem. We have also learned of a 1 1/2-year-old retarded boy, operated on in Gothenburg, Sweden in 1959, who had a heterotopia with cerebral and well differentiated cerebellar component located in a widened left Sylvian fissure.

In our case the miniature cerebellum lay within a hydromyelic dilation of the central canal, referred to in the literature as "myelocystocele" or "syringocele." This entity is considered a variety of spina bifida cystica, which is seen only in the cervical or thoracic region. It is associated with a relatively wide defect in the vertebral arches through which a swelling protrudes into the subcutis. The structure of this swelling is similar to that in our case, that is, it contains neuroectodermal tissue surrounded by a meningeal sac. There is a narrow-based connection between the myelocystocele and the subcutaneous aberrant tissue, which can safely be dissected without damage to the spinal cord, as in our case. Thus this malformation should not be classified as a myelomeningocele, in spite of the presence of subcutaneous neural tissue, because myelomeningoceles are nearly always broad-based with tethered nerve roots or spinal cord and consequently exhibit neurological symptoms. This should rather be considered a myelocystocele associated with meningocele and aberrant neural tissue, which designation takes into account the two different components. The hamartomatous cerebellar heterotopia is an additional superimposed malformation.

An interesting question in our case is whether the miniature cerebellum was functioning; the nearly complete structural organization including the cortical layering and the intrinsic nuclei might also indicate functional connections to the surrounding neural structures. The patient showed a temporarily reduced motility and tonus in all extremities immediately after the operation. Those symptoms could have been due to severed neural connections that later became compensated for by other connections during the postoperative recovery. However, they might equally well have resulted from a minor trauma (such as spinal shock) to the underlying spinal cord caused by manipulation during the extirpation of the miniature cerebellum. In either case, most probably the patient had a normally functioning cerebellum in his posterior fossa. Its presence was verified by pneumoencephalography, and normal functions can be inferred from the practically normal neurological status of the patient 1 1/2 years after the operation.

Acknowledgments

The authors thank Mrs. Margit du Rietz and Mr. Frank Bittkowski for their excellent photographic work.

References

6. Lindgren S: Personal communication, 1977

Address reprint requests to: Anders Suneson, M.D., Department of Neurosurgery, Sodersjukhuset, Stockholm, Sweden.