Henry Ford Hospital Medical Journal

Volume 3 | Number 4 Article 5

12-1955

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CONGENITAL ANOMALIES OF THE BRAIN AND SPINAL CORD

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Congenital anomalies of the brain and spinal cord may arise as a result of hereditary factors or from disturbances in the intra-uterine environment of the embryo. The parents of a child with hydrocephalus, cranium bifidum, or myelomeningocele are naturally concerned as to whether subsequent children will be similarly affected, and often question the physician concerning this possibility.

From the study of Record and McKeown, there is little doubt that following the birth of a child with anencephalus, spina bifida, or hydrocephalus, there appears to be a significantly increased probability of recurrence amounting to about five times normal expectation. The expectation of the population at large of having a child with such a malformation is about one in two hundred. Therefore, in families who have had one child with anencephalus, spina bifida, or hydrocephalus it would be expected, from the statistics of Record and McKeown, that one child in the next forty born to such a family might have one of these abnormalities.

According to Doctor James V. Neel of the University of Michigan Heredity Clinic, the present data on man still do not indicate whether the increased risk of defects of the nervous system in subsequent children is the result of genetic or non-genetic factors. It must be remembered that children with defects of the nervous system are usually unable to propagate, thus further limiting any hereditary tendency from expressing itself. Furthermore, if a first child is affected with a congenital anomaly of the nervous system, parents may tend to have no other children, thus further preventing the hereditary factor from expressing itself.

HYDROCEPHALUS

Before discussing congenital hydrocephalus I should like to discuss five different ways in which ventricular dilatation can be produced:

1. By an overproduction of cerebro-spinal fluid. This is usually due to a papilloma of the chorid plexus and is the least common cause of hydrocephalus.
2. By obstruction to the outflow of cerebro-spinal fluid within the ventricular system or at the foramina of exit in the posterior fossa: The most common cause of ventricular obstruction is tumor. However, a membrane can close the aqueduct of Sylvius or stenosis of the aqueduct of Sylvius can result in a specific type of hydrocephalus, which usually manifests itself symptomatically just after puberty.
3. Hydrocephalus by obstruction in the communicating channels: By "communicating channels" are meant the subarachnoid conduits through which the cerebro-spinal fluid flows to reach the absorptive mechanism on the surface of the cerebral hemispheres.

   Closure of the communicating channels can be caused by a developmental disturbance or by any pre or postnatal infection which might produce an arachnoiditis. The communicating is the commonest type of congenital hydrocephalus but the exact mechanism is certainly not yet understood. In such cases dye placed in the lateral ventricles would be recovered promptly on lumbar puncture, whereas in the obstructive type of hydrocephalus the dye

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would not appear.

4. Hydrocephalus due to failure in development of, or obstruction in, the absorptive mechanism: It is almost impossible to differentiate this condition clinically from a communicating type of hydrocephalus. The treatment of the two conditions, however, would be the same.
5. Hydrocephalus due to increase in protein in cerebro-spinal fluid: Gardner has recently put forth the ingenious theory that an increase in protein in the cerebro-spinal fluid might delay its absorption sufficiently to produce an increase in intracranial pressure or hydrocephalus or both. He suggests that the papilledema somtimes found in spinal cord tumor, small acoustic neurinomas and the Guillan-Barre' syndromes may result from this cause. Additional evidence is needed to prove this theory, however.

TREATMENT OF CONGENITAL HYDROCEPHALUS

When a child with evidence of hydrocephalus enters the clinic, the routine study is usually as follows:

The ventricle is tapped and as clear spinal fluid flows from the needle, it is gradually withdrawn until the fluid ceases to flow. The needle is then grasped flush with the scalp and withdrawn. This gives a comparatively accurate idea of the thickness of the brain substance. If it is less than one centimeter, it is probably unwise to attempt any type of operative procedure. If the brain substance is more than one centimeter in thickness one to two ccs. of dye are placed in the ventricle. After fifteen minutes a lumbar puncture is performed. If the dye is obtained it means that obstruction is not present in the ventricular system or at the foramina of exit of the fourth ventricle. It still does not tell whether the defect is in the communicating channels or in the absorptive mechanism.

The operation of choice in the female is making a communication between the lumbar subarachnoid space and the fallopian tube after the method of Harsh. So far the results have been promising and the operation is physiological in that there is no loss of electrolytes.

The Matson procedure has been followed by a number of good results; however, because of loss of electrolytes there is some danger, particularly in hot weather. There is also the danger that in females particularly, an ascending infection may result in meningitis.

If obstructive hydrocephalus is believed present, air studies are carried out in an effort to demonstrate the location of the block. If this block cannot be relieved surgically a long tube may be placed connecting the lateral ventricle with the ureter, peritoneal cavity or fallopian tube. It is more common than not, however, that this long tube will obstruct, kink, or be drawn back up into the lateral ventricle. In our hands, the results in its use have been unsatisfactory.

Choroid plexectomy has been successful in some hands. An ideal operation for hydrocephalus of any type has not yet been produced.

DIFFERENTIAL DIAGNOSIS IN HYDROCEPHALUS

Congenital hydrocephalus may be simulated by a subdural hematoma. The shape of the head and the bulging of the fontanelle may be exactly the same in the two conditions. In a child with a subdural hematoma, which is not too long standing, however, the facial expression is, as a rule, much more intelligent than it is in the hydrocephalic infant. Subdural hematoma usually does not appear symptomatically before

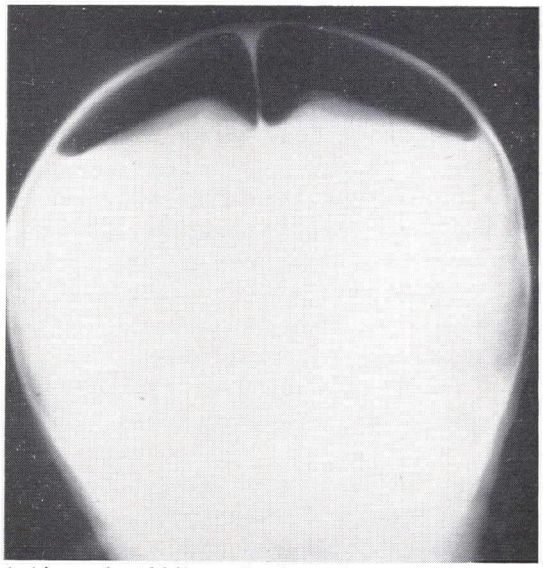

Fig. 1 Air study of bilateral subural hematoma in an infant.

the age of three months. Convulsions are common as are retinal hemorrhages in subdural hematoma, but rare in hydrocephalus. The diagnosis is made by subdural puncture.

Treatment of subdural hematoma is aspiration for a period of several weeks. If this is not effective osteoplastic craniotomy must be performed with partial removal of the membranes.

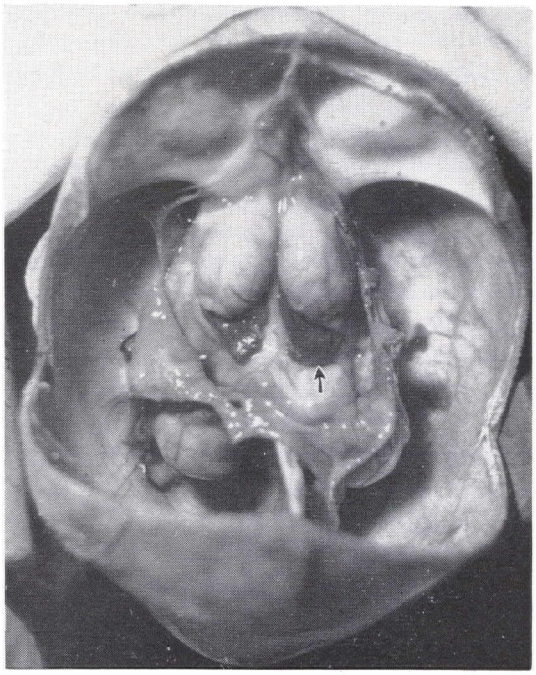

Fig. 2 Hydranencephaly. Arrow points to choroid plexus.

Another condition, which must be differentiated from hydrocephalus is hydranencephaly. This is a condition in which the infant may appear normal at birth. Within a week or so, however, it becomes obvious that he is retarded. The head may then enlarge. Transillumination reveals evidence of complete absence of the brain in the fronto-parietal area. The cause of hydranencephaly is probably congenital obstruction of the foramina of Monro.

CRANIUM BIFIDUM

The occurrence of cranium bifidum with encephalocele in proportion to surgically significant spina bifida is about 1:7. In comparison with spina bifida occulta, simple cranium bifidum is an extremely rare lesion. The patient with an encephalocele, particularly when it is occipital, is apt to have other anomalies of the brain such as microgyria, hydrocephalus, or brain stem deformities. In spite of this, Ingraham and Swan estimate that 34 percent of infants with encephalocele may expect a relatively normal life, following appropriate surgery.

The occipital encephaloceles are sometimes exceedingly complicated and carry the poorest prognosis. In one child, who died at operation when an abnormal circular sinus was opened, the brain stem at autopsy, was found to be rotated ninety degrees.

From a surgical point of view the most interesting of these lesions have been nasal encephaloceles. Two of these were identical, the tips of both frontal lobes having been contained in an unsightly sac, which presented itself at the root of the nose.

CRANIOSTENOSIS

Craniostenosis is a term used by Virchow to designate various types of skull deformity resulting from, or at least associated with, premature closure of the cranial sutures. It was Virchow's theory that when precocious synostosis of two cranial bones through ossification of the suture line is encountered, the growth of these bones

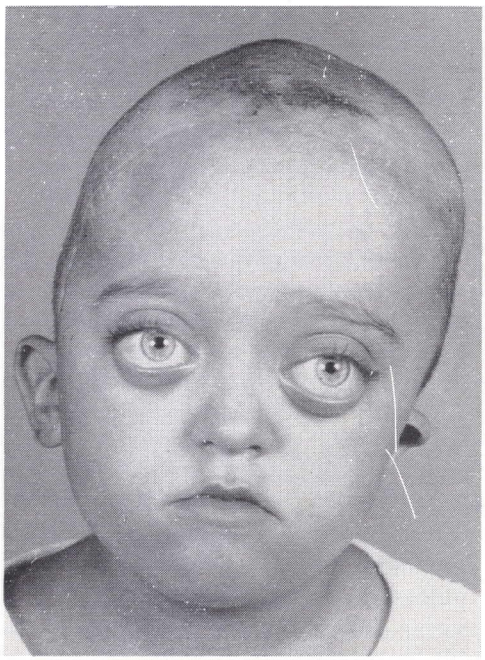

Fig. 3 Oxycephalic type of craniostenosis (10 days postoperative).

perpendicular to the suture line is hindered while overgrowth takes place elsewhere. Thus with closure of the coronal sutures, growth in an anteroposterior direction is inhibited while lateral overgrowth is stimulated and a brachycephalic type of craniostenosis results. Likewise, if the sagittal suture closes prematurely, the skull becomes long and narrow, the dolichocephalic type occurring. If all of the sutures synostose prematurely but the anterior fontanelle remains open for a period of time the oxycephalic or "tower" skull results. Cruzon's disease is an unsightly type of craniostenosis in which the child shows exophthalmos, a bird-like nose and a protruding lower jaw. This facial deformity results from premature synostosis of the facial bones as well as those of the skull.

Premature closure of several of the sutures may only result in ugly deformities of the skull. Craniostenosis, however, usually takes place before birth and, if sufficient sutures are involved, increased intracranial pressure will develop as the brain expands. In time, this results in mental deterioration and visual loss. The object of treatment is to allow expansion of the skull by surgical creation of new suture lines before irreparable damage has taken place. It is, therefore, imperative that the pediatrician recognize this condition early and refer the infant to a surgeon for treatment as soon as possible.

SPINA BIFIDA

Spina bifida is a term loosely used by physicians when they wish to refer to a saccular protrusion which usually contains nerve roots or spinal cord and its membranes. The herniation of nervous tissue through a vertebral defect should more correctly be described as a myelomeningocele, or if it consists only of spinal cord membranes it should be called a meningocele. Spina bifida may be classified as follows:

Spina bifida occulta in which there is a vertebral defect but no obvious protrusion. This common condition is found in about 25 percent of normal individuals. It almost always occurs in the sacral region, which is the last portion of the vertebral column to fuse.

Meningocele where is a protrusion of membranes of the spinal cord only, through the bifid vertebrae. The sac is usually covered by healthy skin but somtimes is formed by a transparent, parchment-like membrane. The base or neck of the sac may be broad but occasionally is narrowed down to a fine stalk. There is usually no neurological defect associated with a true meningocele. As a surgical curiosity, a meningocele may protrude anteriorly through a vertebral defect, simulating an abdominal tumor, or laterally into the chest through an intervertebral foramen. We have seen two cases of the latter in which the preoperative diagnosis has been neurofibroma.

Myelomeningocele where there is a protrusion of spinal cord or nerve roots covered at least by a membrane. This is a more common lesion than meningocele in a proportion of about 7:1. In more than half of the cases of myelomeningocele there is a severe neurologic defect such as paralytic club feet, relaxation of the sphincters, or hydrocephalus. The myelomeningocele occurs least commonly in the dorsal region, which, embryologically, is the first part of the vertebral canal to close.

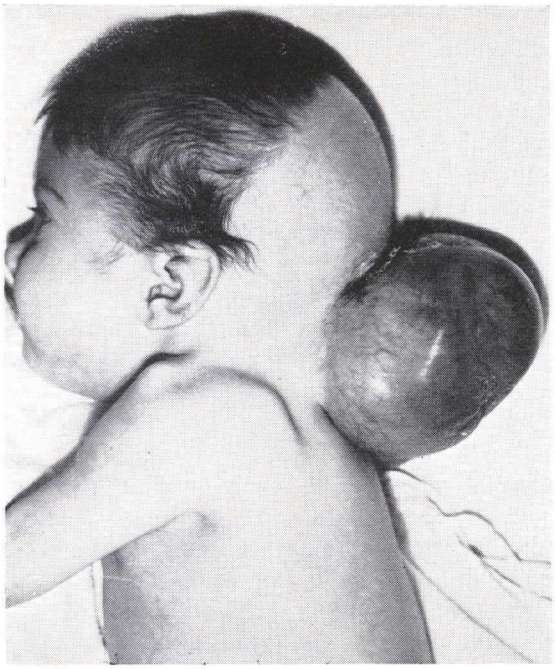

Fig. 4 Combined occipital encephalocele and cervical myelomeningocele.

Myeloschisis is the term applied to a condition represented by a wide open, weeping, shiny, reddish mass which has protruded entirely through the vertebral defect. This is the most marked of the developmental defects of the spinal cord.

Embryological studies carried out by Patten, in the Department of Anatomy at the University of Michigan indicate the extraordinarily early age at which this defect is established. The youngest of the embryos in his series which shows this defect was 8 mm. in crown-rump length, with a presumptive fertilization age of only a little more than five weeks. The traditional interpretation of such defects is based on the developmental arrest concept, but Patten's quantitative assessment of the neural plate tissue in the region of the defect seems to indicate—to the contrary—that local overgrowth of the neural plate is responsible. Myeloschisis is almost invariably fatal within a few months after birth.

SURGICAL TREATMENT

The purpose of operations upon myelomeningoceles is to free all nervous tissue from the tethering effect of the sac, returning the nerve roots and cord to the vertebral canal without increasing the neurological defect. This is often more easily said than done. Care must be taken not to transect functioning cord tissue which has attached itself to the sac and doubled back into the spinal canal. A nerve stimulator is of value here in determining what nervous tissue can be sacrificed. In spite of extreme care, however, the nervous defect is often increased by the operative procedure.

We like to wait as long as possible before operating upon most myelomeningoceles. This is so that hydrocephalus may not be precipitated by the procedure.

We never operated upon patients with paralysis of the legs or sphincters or with

impending hydrocephalus. Occasionally where a sac is extremely large, it may be removed to facilitate nursing care in spite of paralysis.

Where rupture of the sac appears eminent and paralysis is not present, operation must be performed as an emergency.

LIPOMAS OF THE CONUS MEDULLARIS AND CAUDA EQUINA

This particular congenital anomaly of the spinal cord has been recorded in the literature surprisingly seldom. The reason may be that a lipoma of the cord is often confused with the fatty mass which is commonly associated with a myelomeingocele. These lipomas of the cord represent a clinicopathological entity: From birth a non-fluctuant mass is present over the sacral area. Roentgenograms almost invariably show a bifid deformity of the sacrum and of the lower two lumbar laminae. Urinary incontinence or nocturnal enuresis may appear at any time. When exposed at operation the subcutaneous lipoma narrows to a stalk which traverses the bifid vertebra entering the dura though a sizable oval opening to merge with the conus medullaris and cauda equina. With the tethering action of the lipoma it is easy to see how neurological symptoms could arise with growth of the spine. Bassett published nine cases in 1950 from the University Hospital. He showed that where the spinal cord is fixed by a lipoma, the stalk of which penetrates the dura in the lumbosacral area, the nerve roots from the cord segment just above the point of fixation course upward to enter the intervetebral foramina. We have seen this same phenomenon associated with a congenital tumor of the filum terminale which was merged with the dura in the lower end of the dural sac.

CONGENITAL DERMAL SINUS

If in a child with meningitis, the causative organism is not the meningococcus and the source of infection is unknown, the midline of the back and occiput should be searched for evidence of a congenital dermal sinus. Likewise, if meningitis has recurred in the absence of obvious evidence of chronic infection close to the meninges, it is more than possible that the individual is harboring a congenital dermal sinus.

Walker and Bucy first used the term, "congenital dermal sinus," in 1934 and called attention to the fact that the chain of events—infection in a midline dermal sinus followed by meningitis and paralysis—constitutes a clinicopathological entity. They concluded that, at that time in embryological development (late in the first month of intrauterine life) when the cutaneous ectoderm and the neural ectoderm should have become separated, the cleavage between them was incomplete at the particular point where the sinus occurred. Thus the neural tube carried down with it a narrow invagination of skin, the connective tissue layer of which was continous with the connective tissue of the covering of the spinal cord, the meninges. Given such a developmental background, one can easily understand how meningitis can occur if the sinus should become infected.

Congenital dermal sinuses may lead to dermoid and epidermoid tumors of both the brain and the spinal cord. Dermoid and epidermoid tumors of the spinal cord, without a congenital dermal sinus, are not at all rare. Of the two the epidermoid cyst is more frequently encountered, however. Pain is conspicuously absent in dermoid and epidermoid tumors, probably because of the very gradual enlargement of the tumor, its soft smooth character, and its relatively complete fixation. Tumors of this

type may be very extensive. It is often impossible to remove the entire wall of the cyst without causing irreparable damage to the spinal cord or to the cauda equina. It is probable, moreover, that complete removal of the epithelial lining of the epidermoid cyst is necessary in order to prevent recurrence, just as it is with a sebaceous cyst. Sufficiently long periods of case observation have not as yet been recorded to establish this point. It is certain, however, that irreparable damage to the cord should be avoided during the first operation.

The midline dermoid and epidermoid tumors of the brain have been well described by Matson and Ingraham. Most of these tumors are connected to the surface by congenital dermal sinuses, exactly in the same manner as are those of the spinal cord, thus making it possible for infection to precipitate the first sign or symptom. All of the sebaceous material must be removed from these cysts in the uninfected case, and probably the wall as well, or an infective meningitis may develop on the basis of chemical irritation. The risk of this type of meningitis may not be as great as it was before the days of the antibiotics when Critchley and Ferguson reported three cases of incompletely excised cholesteatoma of the brain, all of whom succumbed to meningitis. The cholesteatoma of the brain are not so difficult to remove completely as are those of the spinal cord where so little trauma can have such devastating effect.

Considerable confusion exists between congenital dermal sinuses in the lumbosacral region and pilonidal sinuses and cysts. The former are present from birth and appear as midline dimples, usually surrounded by hair or angiomatous skin. According to Doctor Carl V. Weller, a pilonidal cyst is an acquired lesion. It is almost always found in the sacrococcygeal region and usually appears in young, hairy males. A pilonidal cyst is apparently a reaction to the mechanical implantation of the hair of the patient in and beneath the skin of the midline of the postanal region. A similar lesion has been found between the fingers of barbers where hair follicles are normally absent.

Treatment of Congenital Dermal Sinus: In our experience, the patient with a congenital dermal sinus communicating with the spinal meninges is invariably a child. He has entered the hospital with meningitis or has just recently recovered from it. Penicillin, streptomycin and aureomycin are usually started immediately. If the opening of the sinus tract is inflamed, wet dressing may be applied for short periods. When the acute infection has subsided the sinus tract should be completely excised after it has been well exposed through a carefully performed laminectomy.

DIASTEMATOMYELIA

Diastematomyelia is a cumbersome term which has been used for more than 50 years to describe a cleft in the spinal cord which splits the cord in two, each half surrounded by its own dura. "Diplomyelia" is often used as a synonym for diastematomyelia but, in my opinion, actually refers to a doubling of the spinal cord. The latter condition is rarely of surgical significance and is more common than diastematomyelia. Diplomyelia may exist in the absence of any symptoms.

Diastematomyelia is accompanied by a midline bony spicule which has arisen from the posterior surface of the body of a vertebra and has passed through and impaled the spinal cord or cauda equina. With the relatively greater growth of the spine, traction may later be produced on the spinal cord or on its nerve roots.

The symptoms of diastemotomyelia are usually manifested first in children shortly after they have learned to walk. The first symptom is, as a rule, that of a disturbance in gait. Such a disturbance may consist in unilateral or bilateral weakness of the lower extremities, or in an increasing spastic paralysis. Bladder disturbance or a trophic ulcer of one or more toes may then be noticed or, as a matter of fact, may be the first symptom to be observed.

Examination of the back may reveal a tuft of hair or a lipoma lying over what is revealed by x-ray examination to be a spina bifida. On careful examination of the anteroposterior view of the spine, an abnormal line of increased density may be noted in the middle of the spinal canal. This is usually found in the region of a bizarre spina bifida occulta and represents a bony abnormality which has grown backwards through the spinal cord and its meninges. So far as we know, this abnormal line of increased density was first pointed out by Hamby in a case from which he had removed the midline bony spicule. Neuhauser, and his associates, have since been able to make the diagnosis preoperatively in a number of cases in the Children's Hospital in Boston. Of 11 cases operated upon at that hospital by Matson, Woods, Campbell and Ingraham, the correct preoperative diagnosis was made in eight by X-ray studies. In some cases pantopaque studies were carried out and revealed that the dural sac had been divided into two halves at the point of the bony spicule.

The operation for diastematomyelia follows the same approach as that described

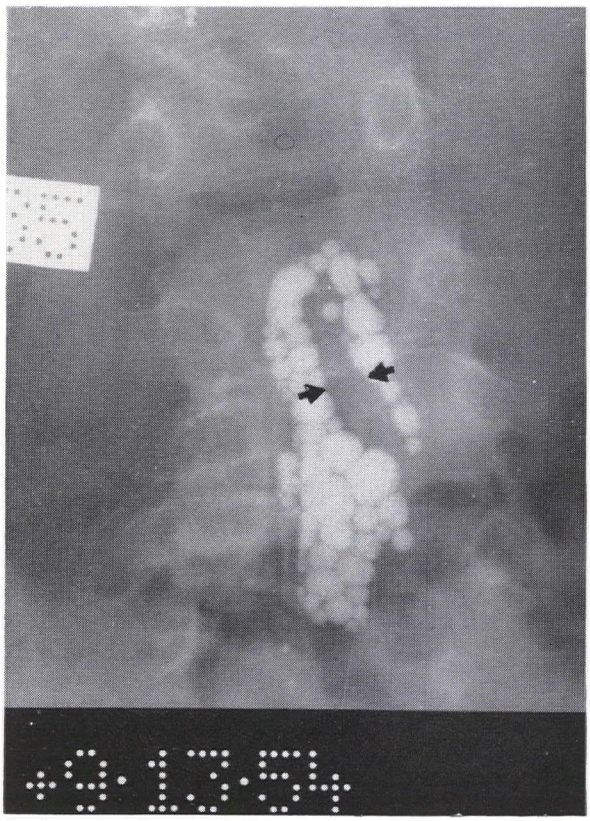

Fig. 5 Myelogram of case of diastematomyelia in which dural septum (arrows) was not excised at first operation.

for spina bifida occulta. When the bony spicule is encountered, it is dissected subperiosteally down to its attachment to the body of the vertebra from which it has arisen. We now believe that it is necessary to open the dura and obliterate the dural septum after the bony spicule has been removed. In one case where we failed to excise the septum, recurrence of symptoms took place five and a half years later. A myelogram (Fig. 5) was then performed and the typical midline defect of diastematomyelia was seen. At operation it noted that scar tissue had replaced the excised spicule of bone and was compressing the cord posteriorly as well. The scar-filled dural septum was excised and the dura closed posteriorly. Rapid improvement followed this procedure.

TERATOMAS OF THE SPINE

Although the teratoma of the sacrum is not specifically an anomaly of the nervous system, it is ordinarily treated by the neurosurgeon. The teratomas of the cervicodorsal and lumbar spine, however, are associated with a vertebral defect and sometimes with a meningocele or myelomeningocele.

The sacral teratoma, however, is a distinct entity. There is no connection with the meninges or spinal cord. It has invariably been associated, in our experience, with a defect on one side of the lower part of the sacrum and a total absence of the coccyx. In 20 such sacral teratomas which we have operated upon in this clinic only three were malignant.

Comment: These lesions may assume a tremendous size. Most observers in the past have concluded that sacral teratomas were in all instances vestiges of a reduced parasitic twin. More recently, however, Patten has pointed out another possible origin:

"The extraordinarily high incidence of trigerminal teratomata in precisely the region where the primitive streak of the embryo was formerly located may well mean that some of these masses originated by neoplastic growth of the tissue in this territory. In the embryo, the region of the primitive streak is a proliferation center from which cells entering all three of the germ layers are derived. It is quite logical, therefore, if it became involved in neoplastic growth that it would give rise to a teratomatous mass in which derivatives of all three of the germ layers were represented."

Patten is careful to emphasize that his interpretation in no way precludes the involvement of twinning in a certain portion of sacral teratomata. The possibility of an additional method of origin in this region, however, may well help explain the high incidence of teratomata in the sacral region as compared with other regions in which unequal conjoined twinning might be involved.

Surgical Treatment: If the teratoma is exceedingly large, necrosis may threaten, in which case the lesion must be excised immediately. The prognosis with teratomas is ordinarily excellent. We have experienced one recurrence of a benign teratoma where some of the tumor was inadvertently allowed to remain. It was successfully excised at a second operation.

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