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# A practical clinical classification of spinal neural tube defects

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**Abstract** Current commonly used terminology to describe neural tube defects (NTD) is inconsistent, overlapping, contradictory and, at times, inaccurate making it difficult to convey the nature of the malformation and what needs to be done to optimally treat patients with these congenital abnormalities.

NTD can be broadly divided into those that are open with exposed neural tissue and leaking cerebrospinal fluid (CSF) and those that are closed with no exposed neural tissue nor loss of CSF. It appears that the loss of CSF during development is the underlying factor that leads to the entire central nervous system involvement with an open NTD and lack thereof with a closed NTD, wherein only the spinal cord is malformed. There are, however, rare transitional cases that bridge the gap between the two forms.

Agreeing on a nomenclature that is used in a standard fashion would be of help in addressing this group of congenital anomalies that have a great deal of variability and, at times, can be quite complex.

**Keywords** Spinal neural tube defects · Spinal dysraphism · Spina bifida · Open Neural Tube Defect · Closed Neural Tube Defect · Myelomeningocele · Meningocele · Myelocystocele · Limited dorsal myeloschisis

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## Introduction

The classification of spinal neural tube defects (NTD) is confusing and contradictory with a result that clinical care may be misdirected and outcome studies difficult to assess. The purpose of this communication is an attempt to simplify and rationalize the classification of spinal NTD in order to promote a more standard nomenclature that in turn leads to better understanding and treatment of patients with these congenital anomalies.

### Dysraphism

Raphe (or *rhaphe*) is a Greek word meaning "the line of union of two contiguous bilaterally symmetric structures" [1]. Dysraphism is defined as "defective fusion, especially of the neural folds" [2]. Thus, dysraphism refers to failure of normal midline fusion and is "a general term used to describe a collection of congenital abnormalities that include defects in the vertebrae, and spinal cord or nerve roots" [2]. These congenital abnormalities can vary from inapparent and insignificant (Figs. 1 and 2) to massive and incompatible with survival (Fig. 3).

### Spina bifida

Spina bifida is the term most commonly used for these dysraphic lesions and is defined as "embryologic failure of fusion of one or more vertebral arches; subtypes of spina bifida are based on the degree and pattern of malformation associated with neuroectoderm involvement" [3]. The most common terms are spina bifida aperta, spina bifida cystica, and spina bifida occulta. Spina bifida aperta and spina bifida cystica are frequently used interchangeably. The malformations

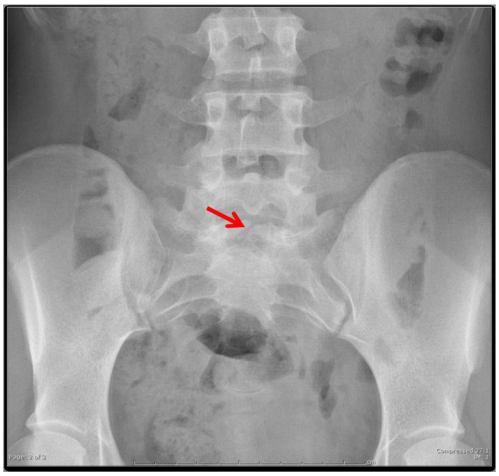


**Fig. 1** Incomplete posterior arch at L5 (*arrow*). This is a common incidental finding and is of no significance by itself

that are depicted by Figs. 4 (myelomeningocele) and 5 (meningocele) would be classified as spina bifida cystica and would imply that these are comparable lesions which they are not, as the former involves the entire central nervous system (CNS), producing marked deficits, while the latter involves only the spine and may produce no or minimal deficit. Figure 6 (myeloschisis) can be classified as spina bifida aperta but not spina bifida cystica since there is no cystic component; however, the clinical manifestations are similar to those of an infant with a myelomengingocele with involvement of the entire CNS. Furthermore, most, but not all, abnormalities classified as spina bifida occulta have external cutaneous manifestations that can vary from insignificant (Fig. 7) to very large, obvious (Fig. 8), and certainly not hidden or concealed as the term occult implies. Figure 8 could also be labeled as spina bifida cystica but once again the infant with this anomaly differs significantly in clinical manifestations from the infant that depicted by Fig. 4 (myelomeningocele). Since the terms spina bifida cystica and occulta can be misleading, the only consistent term is spina bifida aperta, which applies to a malformation very infrequently encountered—myeloschisis (Fig. 6). It is this author's contention that these terms be avoided.

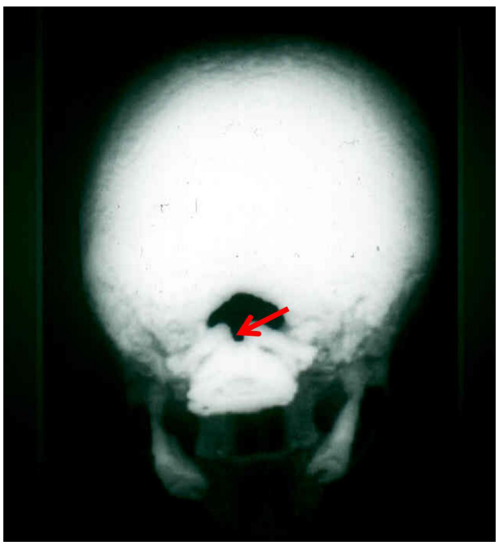


**Fig. 2** Incomplete closure of the posterior arch of C1 (*arrow*). This is a common finding that alone is of no clinical consequence

### Isolated bony defects in the posterior vertebral arch

Defects in the spinous process or laminae at L5, S1 (Fig. 1) or lower sacral levels are frequently noted as an incidental finding in the normal population. Such bony defects are overwhelmingly isolated, have no clinical significance, and could legitimately be termed spina bifida. The embryologic aberration—if it can be called such—that produces this mesenchymal defect in the posterior portion of one or more vertebral arches is unknown and does not appear to be linked to the process of neurulation. In fact, it may be a normal variation.

The only reason to mention the incomplete vertebral arch category is that it can be reported in imaging studies—most commonly plain spine X-rays—as spina bifida or spina bifida occulta. This can lead to a chain of events including one or more referrals for consultation and additional imaging studies. If the incomplete posterior arch(es) is an incidental finding in an asymptomatic patient with a completely normal examination and no cutaneous findings, a further diagnostic workup is not indicated.

## Neural tube defects

The defects of clinical significance involve the neural tube. Thus, the use of the term NTD would encompass all of the lesions in this group and focus the attention on the neural elements. The spectrum of NTD is considerable and often can be complicated.

NTD can be divided into two groups: one in which the NTD is open and the other in which it is closed. An open NTD has exposed neural tissue and is continuously or intermittently leaking cerebral spinal fluid (CSF). Extensive changes are almost always present in the CNS with a hindbrain malformation (referred to as Chiari II malformation) and migration abnormalities of the CNS very frequently present (Fig. 9). The second group of NTD is one that is closed with no visible neural tissue and not leaking CSF. In this group, the malformation is limited to the spinal cord, the brain is rarely malformed, and the Chiari II malformation and hydrocephalus are rarely encountered (Table 1).

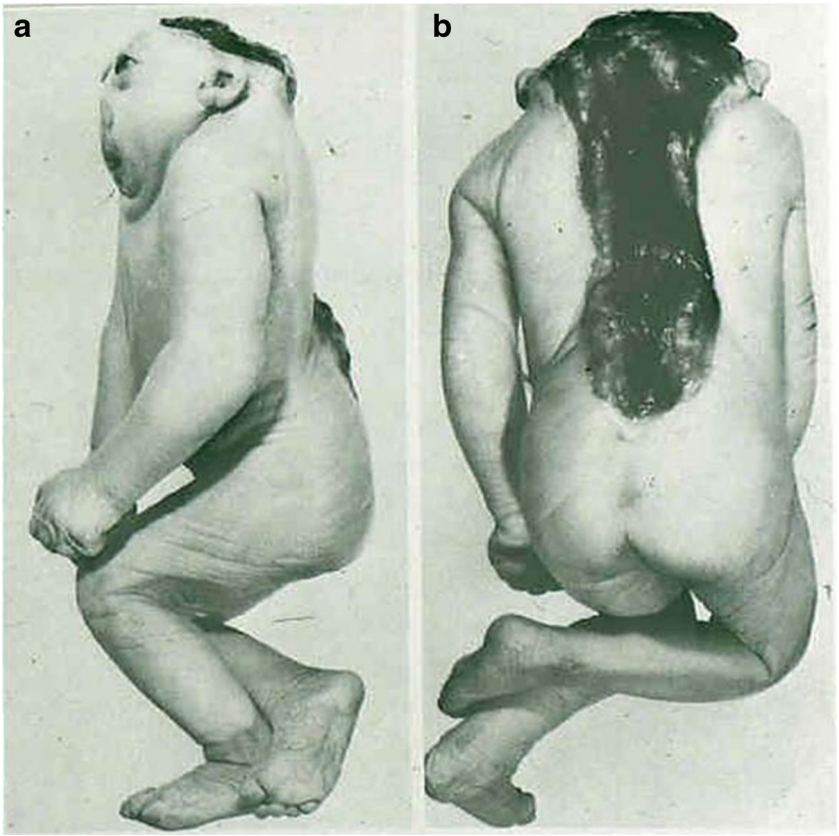



**Fig. 3** **a**, **b** Craniorachischisis is failure of closure of the cranium and spinal column. This deficit is incompatible with survival

Why the difference in CNS involvement between the two groups? Although other theories exist, the most likely explanation was provided by McLone and Knepper in 1989 [4] who postulated that the loss of CSF from the open NTD results in a lack of distention of the neurocranium, producing a secondary effect of a small posterior fossa that in turn causes the hindbrain malformation (Fig. 9) and various degrees of cerebral disorganization. This explanation is supported by the report of Adzick et al. [5] that prenatal closure of open neural tube

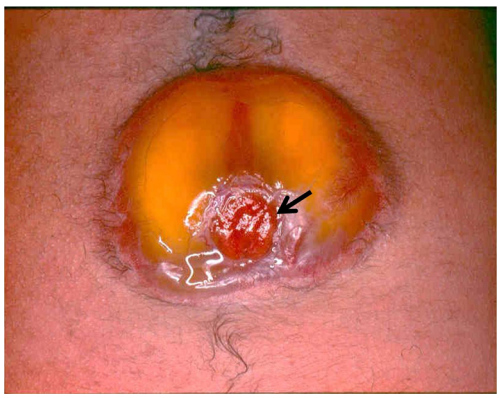


**Fig. 4** Open NTD—myelomeningocele. The size and shape of the myelomeningocele can vary significantly. In this example, the neural placode (*arrow*) is located at the caudal end of the cystic dilation of the dysplastic meninges. CSF will intermittently drain from the lesion

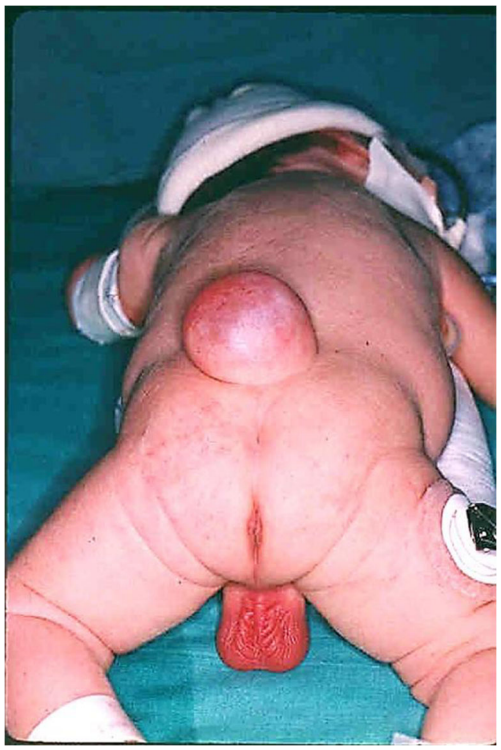


**Fig. 5** Closed NTD—meningocele. A typical lumbosacral meningocele in a newborn. The overlying skin was normal with the exception of mild telangiectasia. The neonate had no neurological deficit. The meningocele was repaired and the patient was completely normal

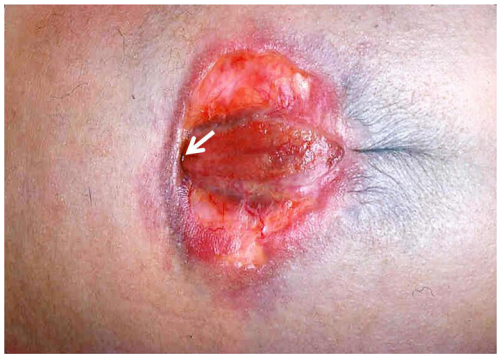


**Fig. 6** Open NTD—myeloschisis. This infant has a form of NTD referred to as myeloschisis. The neural placode lies on the anterior wall of the open spinal canal. The *arrow* points to the central canal of the spinal cord from which CSF is continuously leaking

defects has shown to decrease the incidence and severity of the hindbrain herniation. Further evidence was provided by Welch et al. [6] showing that lumboperitoneal shunts can lead to an acquired Chiari I malformation secondary to the pressure differential created between the cranial and spinal com-partments by the lumbar shunt, leading to downward migration of the cerebellar tonsils to produce a milder form of hindbrain herniation. Figure 10 depicts a newborn with a split cord malformation in which the NTD on the right side is closed while that on the left is open. The resulting clinical picture is that of an open NTD, supporting the position that the loss of CSF during development is the causative factor that distinguishes these two groups of NTD.

## Embryology of neural tube defects

Embryologically, NTD can be classified as preneurulation, neurulation, and postneurulation abnormalities.

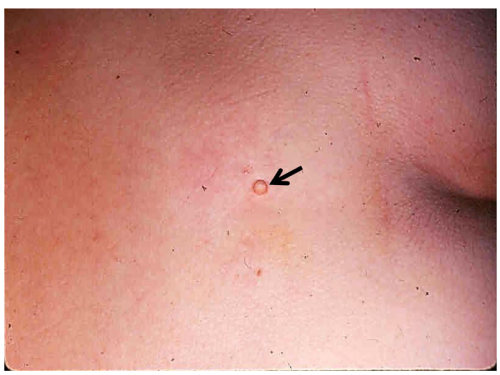


**Fig. 7** Closed NTD—lipomatous malformation. The small fibrolipomatous nodule in the lumbosacral region (*arrow*) was the only visible malformation of this infant with a low lying spinal cord tethered by a short thickened filum terminale containing adipose tissue that extended into the conus medullaris. Approximately one half of the patients with an abnormal filum terminale have no visible abnormalities

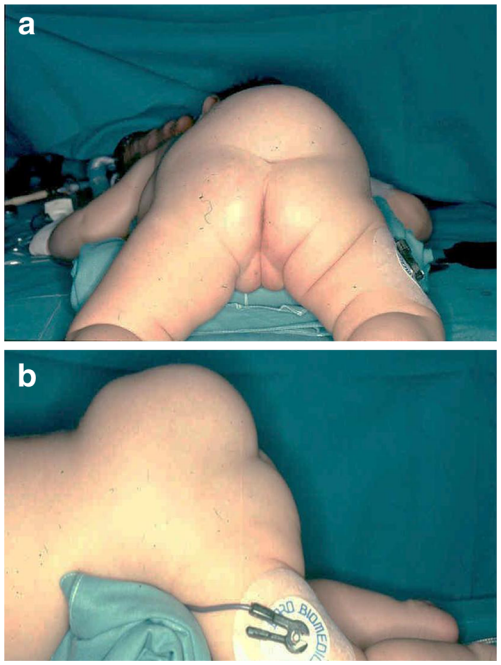



**Fig. 8** **a**, **b** Closed NTD—lipomatous malformation. The infant has a closed form of NTD with a lipomatous malformation that extends from the subcutaneous region to intimately involve the low lying tethered spinal cord (**a** + **b**). The overlying skin is normal. The abnormality is limited to the lower end of the spinal cord without involvement of the rest of the CNS

Preneurulation NTD encompass those that involve the neurenteric canal (Fig. 11a). At this early point in gestation, the tissue that is to form the neural tube is still in the form of a plate. Neurulation defects occur later and are overwhelmingly open NTD with myelomeningoceles (Fig. 4) being the most common form. Postneurulation defects result from faulty development and coalescence of the caudal cell mass that connects with the closing neural tube (Fig. 11b), thus leading to a closed form of NTD. These occur mainly in the lumbosacral region. To make matters more complicated, primary neurulation may begin at multiple locations and not just at one site as has been previously thought [7], and multiple neurulation abnormalities may be present in the same patient.

## Open neural tube defects

### Myelomeningocele

The dysplastic meninges and spinal cord protrude through a defect in the posterior vertebral arches to extend beyond the spinal canal. These are by far the most common form of open neural tube defect, leak

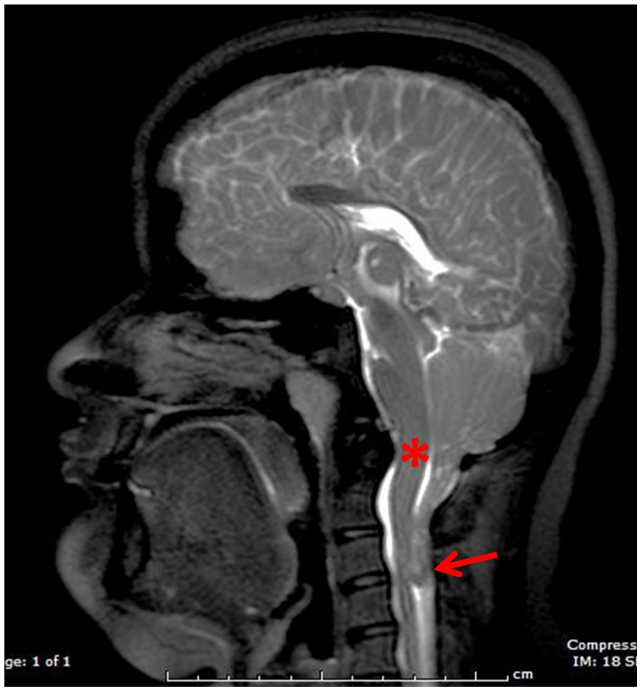



**Fig. 9** CNS malformation associated with open NTD. Sagittal T2-weighted MRI of a teenager with a repaired open NTD and shunted hydrocephalus. The small posterior fossa is associated with a hindbrain herniation (Chiari II) with downward displacement of the cerebellum and brainstem. The cerebellar tonsils extend to C4 (*arrow*) and the medulla is in the cervical canal (*asterisk*). Migration abnormalities can be present to a varying degree in both hemispheres leading to malformation of the lamination and gyral pattern. It would appear that the loss of CSF from the open NTD in utero produces the changes noted secondary to lack of distention of the neurocranium during development

CSF intermittently or continuously, and are associated with malformation of the entire CNS (Fig. 4).

**Table 1** A practical clinical classification of spinal neural tube defects

| |
|---|
| Open neural tube defect |
| Myelomeningocele |
| Myeloschisis |
| Hemimyelomeningocele |
| Closed neural tube defect |
| Meningocele |
| Posterior—lumbar, sacral, and thoracic |
| Posterior—cervical and limited dorsal myeloschisis |
| Anterior—sacral |
| Myelocystocele |
| Lipomatous malformation |
| Abnormal filum terminale |
| Congenital dermal sinus |
| Split cord malformation |
| Neurenteric cyst |
| Neural tube defect associated with caudal regression |

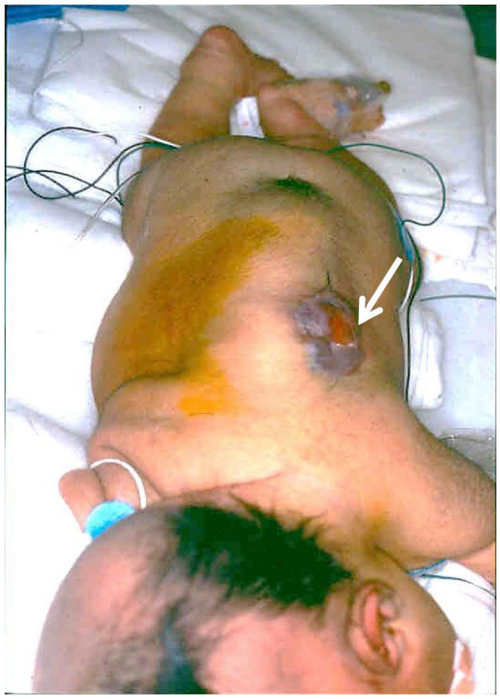


**Fig. 10** Open NTD—hemimyelomeningocele. This newborn infant has a hemimyelomeningocele, a very rare lesion. A bony spur divided the spinal cord into two segments, one that was open (*arrow*) and the other fully closed. The patient has normal neurologic function in the right lower extremity, but has a profound deficit on the left. The remainder of the CNS is malformed with the presence of a Chiari II malformation and hydrocephalus. Thus, from a clinical standpoint, this patient has the diffuse CNS involvement seen with a typical myelomeningocele

### Myeloschisis

This is a form of open NTD with no cystic component. The neural placode is plastered against the ventral wall of the spinal canal and continually drains CSF. The loss of CSF produces the hindbrain herniation (Chiari II malformation) and brain migration abnormalities, as it does with a myelomeningocele (Fig. 6).

### Hemimyelomeningocele

This is a very rare form of NTD that appears to reflect disruptions in both the preneurulation and neurulation processes that together result in a split cord malformation, closed on one side and open on the other (myelomeningocele). The loss of CSF from the myelomeningocele component sets the clinical picture of entire CNS involvement (Fig. 10).

## Closed neural tube defect

### Meningocele—posterior lumbar, sacral, or thoracic

As this is a closed form of neural tube defect tube without the loss of CSF during development, the only

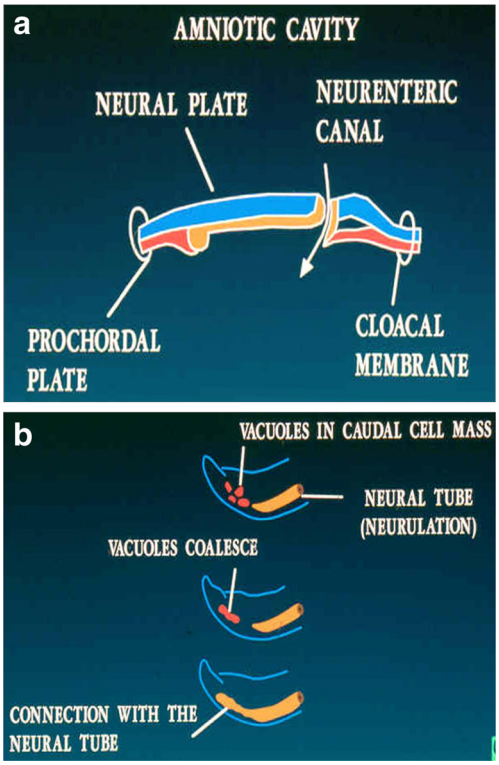



**Fig. 11** **a** Drawing depicting the relationship of the neurenteric canal (endoderm—*yellow*) to the neural plate (ectoderm—*blue*). Mesoderm is in *red*. Abnormalities during this stage (preneurulation) lead to a variety of NTD. **b** Drawing depicting the coalescence of the caudal cell mass to connect to the distal end of the closing neural tube. It is thought that the majority of the closed NTD in the lumbosacral region result from a perturbation of this process

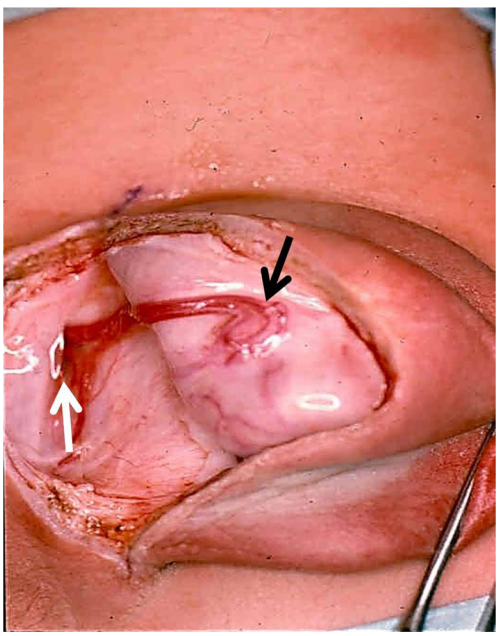


**Fig. 12** Closed NTD—meningocele. After opening the meningocele dome, a fibroglial nodule, densely adherent to the meningocele sac, was noted (*black arrow*) and resected flush with the spinal cord to minimize subsequent tethering. The *white arrow* points to the pedicle where the meningocele connected to the lumbar subarachnoid space

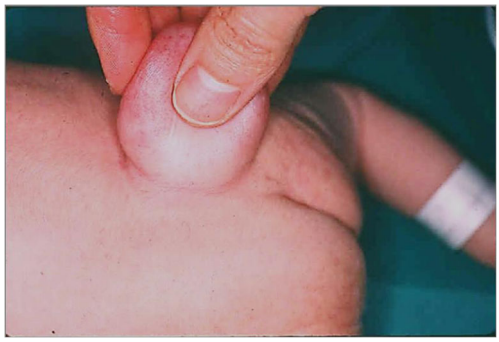


**Fig. 13** Closed NTD—posterior lumbosacral meningocele. This meningocele was covered with normal skin and was easily compressible. The spinal cord is within the spinal canal. This neonate had no neurological deficit

abnormality is within the spine. Meningoceles in the lumbar or sacral region result from maldevelopment in the post or secondary neurulation sequence (Fig. 12). The embryology of rare posterior thoracic meningocele is less well understood.

These lesions are pedunculated to varying degrees, can vary considerably in size, are usually reducible to some extent, transilluminate well, and are fully covered with cutaneous elements, that can be completely normal (Fig. 13) to having varying degrees of dysplasia (Fig. 14). These infants usually have a normal neurologic examination without deformity of the lower extremities or sphincter abnormality.

Although the meninges herniate through a defect in the posterior vertebral arches, the spinal cord resides within the spinal canal. Probably most, if not all, posterior meningoceles are not "pure" in that they contain either singly or in combination, aberrant nerve roots adherent to the inner wall of the meningocele herniation (Fig. 12), occasional ganglion cells, and even a glial nodule that may represent a diverticulum from the central

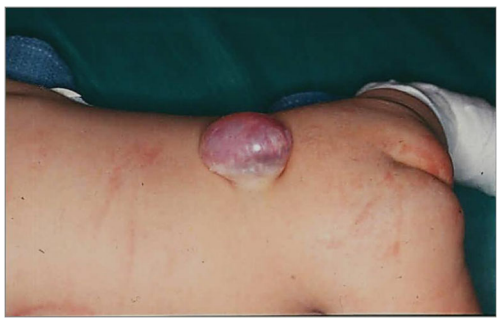
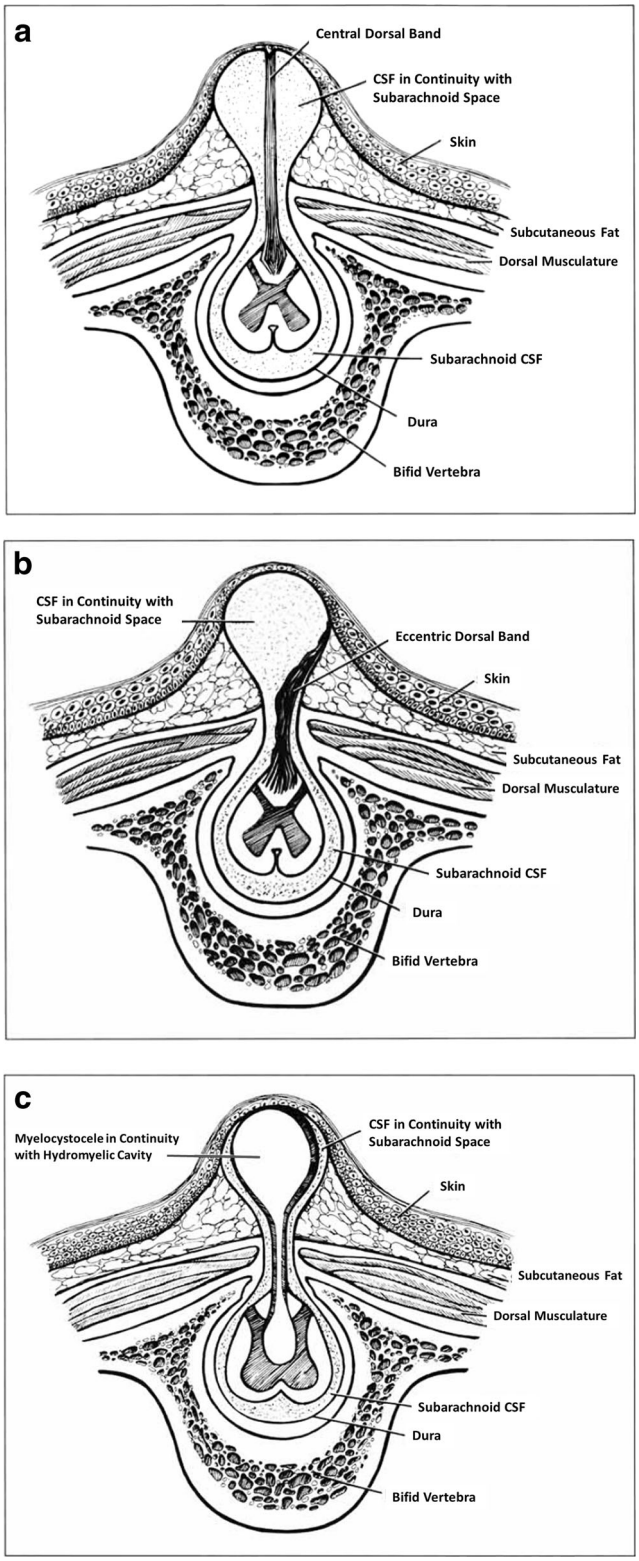


**Fig. 14** Closed NTD—lumbar meningocele. A smaller pedunculated lumbar meningocele in a newborn. In this case, the skin was dysplastic; however, there was no loss of CSF. The neurological examination was normal




◀ **Fig. 15** **a**–**c** Diagrams of limited dorsal myeloschisis. **a** A cervical meningocele with a fibroglial band extending from the dorsal surface of the spinal cord to the dome of the meningocele. From Steinbok and Cochrane ([9], p. 321) (reproduced with permission). **b** A cervical meningocele with a fibroglial stalk eccentrically located and extending from the dorsal surface of the spinal cord to the lateral aspect of the meningocele sac. From Steinbok and Cochrane ([9], p. 321) (reproduced with permission). **c** A cervical meningocele with a hydromyelic central canal herniating from the dorsal surface of the spinal cord to produce a myelocystocele. From Steinbok and Cochrane ([9], p. 320) (reproduced with permission)

canal of the spinal cord (Fig. 15b). Even though fragmentary neural elements are beyond the confines of the spinal canal, the spinal cord is not.

## Meningoceles—posterior cervical and limited dorsal myeloschisis

A very small percentage of meningoceles are difficult to classify and may consist of more than one form of neurulation abnormality. Such a group is posterior cervical meningoceles. Posterior lumbar and sacral meningoceles are thought to result from failure of normal postneurulation development, but this theory does not explain meningoceles in the cervical region where the neural tube is formed by the primary neurulation process. It is postulated that this form of NTD occurs during gastrulation (i.e., preneurulation) as an abnormal midline endomesenchymal tract that bisects the notochord and neural plate and produces a secondary disorder in the primary neurulation process. This abnormality has been termed limited dorsal myeloschisis and consists of a small area of dorsal midline attachment between neural and cutaneous ectoderm that persists after final fusion of the edges of the neural plate [8].

Steinbok and Cochrane [9] have proposed a unifying hypothesis: that of limited dorsal myeloschisis consisting of a stalk with or without dysplastic glial elements extending to the dome or side of the meningocele sac (Fig. 15a, b). Other forms of limited dorsal myeloschisis have an extension of a diverticulum from the central canal containing CSF resulting in a dorsal myelocystocele as well (Fig. 15c). A more extensive disruption of the notochord and neural plate could presumably result in the formation of a neurenteric cyst, split cord malformation, or the presence of both conditions. In this category, the spinal cord lies within the spinal canal. That being the case, it is better to think of these lesions as meningoceles with associated limited dorsal myeloschisis (Fig. 16). These cervical NTD can be found alone or in conjunction with encephaloceles or other NTD on the spinal axis (Fig. 17).

At birth, these lesions in the cervical region present as cystic masses that may be fully epithelialized (Fig. 18a), partially covered with dysplastic epithelial tissue (Fig. 18b), or open (Fig. 18c). Those cervical meningoceles associated with a Chiari II malformation are likely to be open or have been open NTD at one

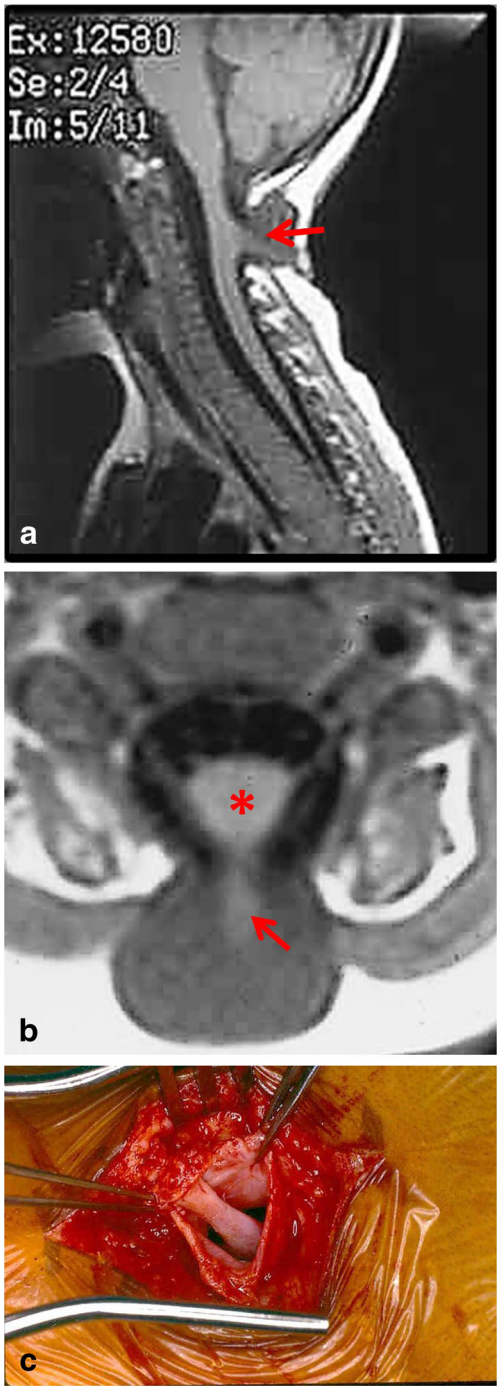



**Fig. 16** **a**–**c** Limited dorsal myeloschisis. **a** A sagittal T1-weighted MRI scan of a 3-year-old boy with a cervical meningocele fully covered with normal skin showing a dorsally tethering fibroglial stalk (*arrow*). The cerebellar tonsils are in normal position and hydrocephalus was not a factor. This patient had no significant neurological deficit either pre- or postoperatively. **b** An axial T1-weighted MRI showing the presence of a dorsally tethering fibroglial stalk (*arrow*), extending into a meningocele. The spinal cord (*asterisk*) resides within the spinal canal. **c** The fibroglial stalk is in the process of being disconnected from the dome of the meningocele. Its base extends to the dorsal surface of the spinal cord and would correspond to the diagrammatic representation shown in Fig. 15a

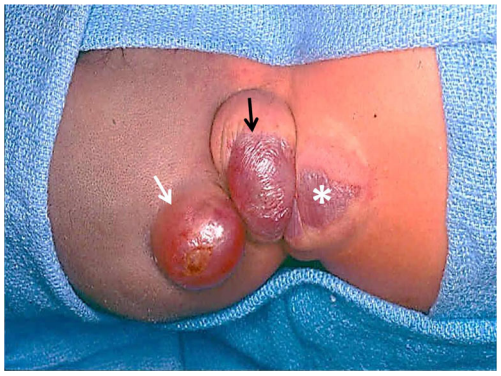


**Fig. 17** Multiple NTD. A newborn with a combination of a cervical meningocele (*asterisk*), cervical myelomeningocele (*black arrow*), and occipital encephalocele (*white arrow*)

point as characterized by eschar or dysplastic skin over the dome of the cyst, while those that are fully epithelialized with normal-appearing skin are more likely to be closed NTD and not associated with more extensive CNS involvement. These posterior cervical NTD may be a transition between the open and closed group of NTD.

## Meningocele—anterior sacral

Anterior sacral meningoceles are truly occult lesions because there are no visible abnormalities. These lesions form as a result of faulty embryogenesis involving the caudal cell mass and, as a result, are often associated with rectal anomalies; malformation of the uterus and vagina; duplication of the renal pelvis or ureter; bony pelvic and vertebral abnormalities; and dermoids, teratomas, or hamartomas associated with the cyst. The embryologic abnormality allows herniation of the dura mater beyond the spinal canal leading to the development of the anterior meningocele (Fig. 19). The cysts slowly enlarge over decades and displace the rectum, bladder, and ureters, giving rise to difficulty with bladder and bowel function and dystocia in females. These lesions are small in infancy and very slowly and progressively enlarge secondary to hydrostatic pressure and CSF pulsations.

A variant of the more complex anterior sacral meningocele is that of an anterior meningocele associated with an anterior sacral bone defect and anorectal anomalies. These three associated malformations are referred to as Currarino's triad. This triad may form by a somewhat different embryogenesis than that of the more extensive anterior sacral meningocele complex [10] or possibly the same process but with less perturbation.

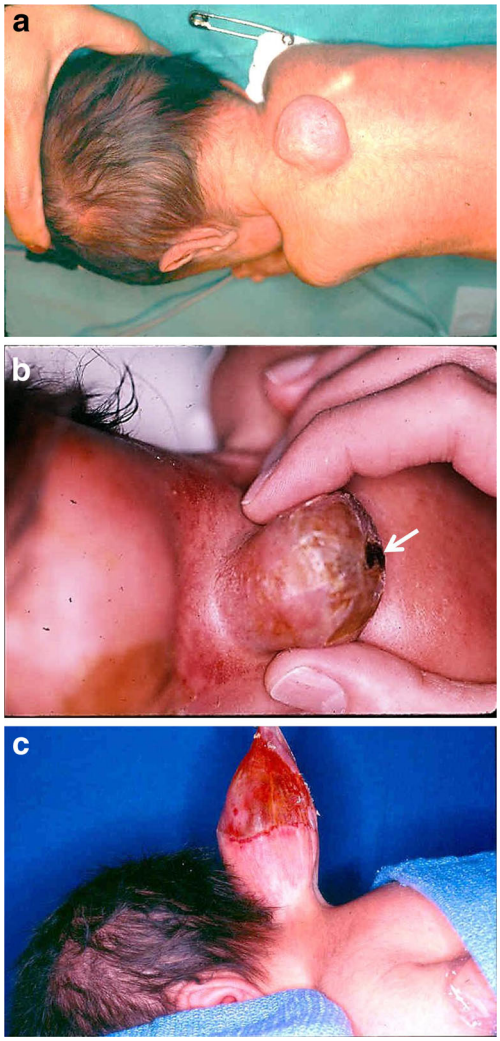



**Fig. 18** **a**–**c** Closed and open NTD—posterior cervical meningocele. **a** A neonate with a cervical meningocele completely covered with normal skin. The CNS was normal and probably reflects that CSF was not lost during gestation. **b** A neonate with a broad-based cervical meningocele, covered by normal skin at the base, but with dysplastic tissue and an eschar at its dome (dark area—*arrow*). Infants with lesions such as this may have a Chiari II malformation and hydrocephalus. One wonders if cervical meningoceles such as this may have been an open NTD at one point. Lesions such as this may be a transition between open and closed NTD. **c** A neonate with a large pedunculated cervical meningocele covered by normal skin at the lower half and dysplastic skin at the upper. This lesion was open at the time of delivery and was leaking cerebrospinal fluid. This neonate had diffuse malformation of the CNS

## Myelocystocele

As noted above, when discussing posterior cervical meningoceles, a myelocystocele can be seen with limited dorsal myeloschisis and consists of a herniation of the dilated central canal of the spinal cord containing CSF that protrudes through a defect in the posterior vertebral arches to extend beyond the spinal canal; however, the spinal cord remains within the spinal canal. These abnormalities can occur

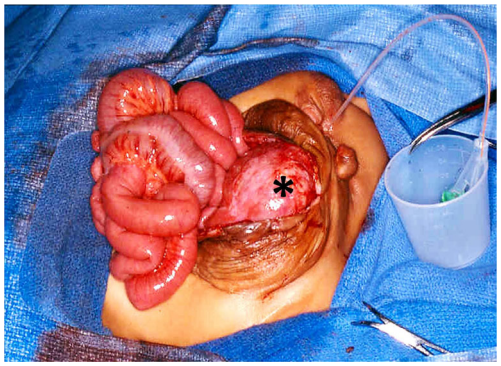



**Fig. 19** Anterior sacral meningocele. The meningocele (*asterisk*) contained a dermoid cyst and CSF. The intestines are displaced for exposure. The catheter is draining urine from the bladder

anywhere along the spinal cord but appear to be more frequent in the posterior cervical region and in the lumbosacral region associated with caudal regression abnormalities (Fig. 20). With a myelocystocele, there are two separate fluid compartments—one the outpouching of the dilated central spinal canal and the other the subarachnoid space—both of which contain CSF.

In referring to Stedman's, one finds the terms myelocele [11], myelocyst [12], and myelocystocele [13] defined as follows:

1. Myelocele:
   (a) Protrusion of the spinal cord in spina bifida (Greek *myelos*, marrow + *kele*, hernia).
   (b) The central canal of the spinal cord (Greek *myelos*, marrow + *koilia*, a hollow).
2. Myelocyst: Any cyst that develops from a rudimentary central canal in the central nervous system (Greek *myelos*, marrow + *kystis*, bladder).
3. Myelocystocele: Spina bifida containing spinal cord substance (Greek *myelos*, marrow + *kystis*, bladder, + *kele*, tumor). Note that *kele* in this definition is tumor, while *kele* is defined as hernia when used with myelocele.

The terms as defined by Stedman's are, to say the least, confusing. A myelocele by Stedman's definition is synonymous with a myelomeningocele for the first definition and a dilatation of the central canal of the spinal cord for the second. A myelocyst would seem to coincide with the second definition of a myelocele.

It would seem better to avoid the use of the terms myelocele and myelocyst and just use the term myelocystocele as this term is in common clinical usage while myelocele and myleocyst are not.

The common clinical usages for CSF dilatations within the spinal cord are referred to as hydromyelia, syringomyelia, and hydrosyringomyelia. Hydromyelia is a distention of the central canal with the ependymal lining of the canal intact,

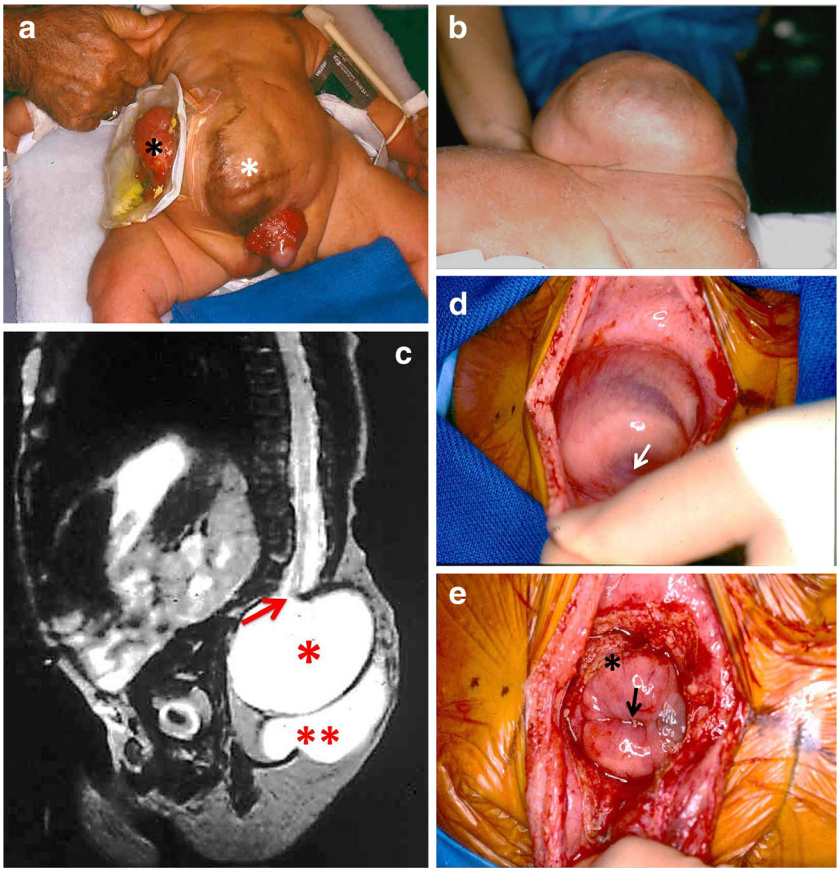



**Fig. 20** **a**–**e** Neural tube defect associated with a caudal regression. **a** This infant has a colostomy (*black asterisk*) to address the imperforate anus. The abdominal wall defect (*white asterisk*) has been repaired. The genitalia are ambiguous and the bladder is exstrophic. The deficits correspond to the omphocele, extrophy of the bladder, imperforate anus, and sacral agenesis (OEIS) grouping. **b** A myelocystocele is present. The mass contains neural elements, CSF, and adipose tissue. **c** Sagittal T2-weighted MRI showing the spinal cord (*arrow*) opening into the myelocystocele and represents the distal central canal of the spinal cord containing CSF (*single asterisk*). The *double asterisk* is the surrounding subarachnoid space also containing CSF but separate from that in the myelocystocele. **d** The myelocystocele has been opened. The *arrow* points to the opening of the central canal of the spinal cord. **e** The edges of the neural placode have been separated from the surrounding tissue. The outside edges will be apposed imbricating the neural tissue eliminating the myelocystocele. The *arrow* points to the central canal and the *asterisk* is on the margin of the neural tissue

syringomyelia occurs when the ependymal lining is ruptured, and the CSF collection extends into the parenchyma of the spinal cord, and hydrosyringomyelia is a combination of both. The collections of CSF in the spinal cord of clinical significance are most frequently hydrosyringomyelia and often referred to as a spinal cord syrinx.

## Lipomatous malformations

This category includes all closed spinal NTD with excessive lipomatous tissue present within or attached to the spinal cord or filum terminale and compromises those lesions that have a variety of names but are most frequently referred to as lipomyelomeningoceles. The adipose tissue associated with this group of lesions is frequently referred to as lipomas, implying a benign tumor of fatty tissue with progressive increase in the number of cells. These malformations are really hamartomatous—as the number of cells does not increase—each individual cell, however, can enlarge. The term lipomyelomeningocele is misleading in another sense: in some cases, the spinal cord extends beyond the spinal canal and is indeed a myelomeningocele (Fig. 21a), while in many others, the spinal cord is within the spinal canal, making this lesion a meningocele (Fig. 21b). The use of the term lipomyelomeningocele can also imply the faulty concept that a lipomyelomeningocele is a myelomeningocele with fat which it is definitely not.

These lipomatous malformations are by far the most common form of a closed spinal NTD and vary from an enlarged filum terminale containing adipose tissue to a huge fatty mass occupying most of the dorsal lumbosacral region and containing the spinal cord and CSF (Fig. 8a, b). As this group of lesions is one of a continuum wherein the embryologic maldevelopment, clinical findings, and prognosis are variable to a degree but

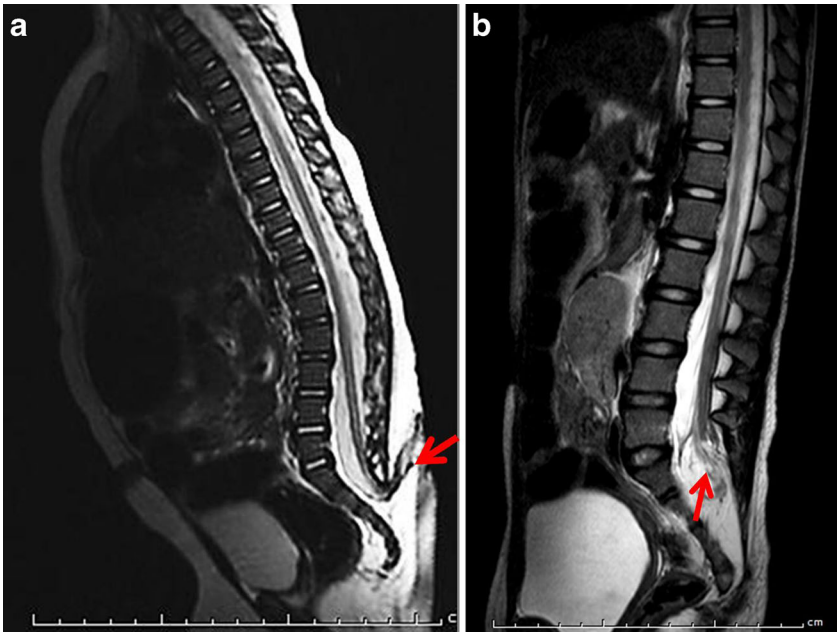



**Fig. 21** **a**, **b** Closed NTD with lipomatous malformation. **a** T2-weighted sagittal MRI of subcutaneous lipomatous tissue that extends into the low lying tethered spinal cord. In this case, neural tissue (*arrow*) and CSF extend beyond the spinal canal. This lesion could be termed a *lipomyelomeningocele*. Because the disordered embryogenesis, clinical manifestations, and prognosis for patients with this form of closed NTD differ considerably from those of patients with a myelomeningocele, it is better to refer to these lesions as lipomatous malformations to eliminate the misconception that a lipomyelomeningocele is a myelomeningocele with fat. **b** This infant has a low lying and tethered spinal cord with lipomatous tissue extending into the conus medullaris and nerve roots of the cauda equina. As the spinal cord is within the confines of the spinal canal (*arrow*), this could be classified as a meningocele but clinically differs little from the situation in which the spinal cord extends beyond the confines of the spinal canal, as shown in **a**

distinctly differ from those of an open NTD (i.e., myelomeningocele)—it seems more appropriate to refer to this group of closed NTD as lipomatous malformations. Although the NTD are limited to the spinal cord, sparing the rest of the CNS, the degree of severity has a wide range making it hard to assess the need for surgical intervention, surgical techniques, and various treatment modalities on long-term outcome [14, 15].

## Abnormal filum terminale

An abnormal filum terminale not associated with other forms of neural tube malformations is the "mildest" form of a closed NTD and can result in a tethered spinal cord. The filum is short, with a diameter greater than 2 mm, and contains an increased amount of connective tissue and/or fat with the conus below the L2 level. A filum up to 2 mm in diameter containing adipose tissue is a normal variant and by itself of no consequence. The degree of abnormality of the filum is variable, and at times, it can be difficult to know if the filum is producing a meaningful tethering of the cord sufficient to explain existing symptoms or can result in a future deficit. Clinical symptoms are localized to the conus medullaris with the most frequent deficit being a neurogenic bladder. An abnormal filum is often seen in conjunction with lipomatous malformations (Fig. 22a, b).

The embryological deficit may relate to failure of normal involution of the terminal spinal cord following completion of secondary neurulation. Approximately one half of all patients with an abnormal filum terminale have no cutaneous markers for dysraphism. Fibrous bands extending from an area of cutis aplasia (Fig. 23)—sometimes called meningocele manqué or atretic meningocele—to a thickened filum terminale can also be included in this category.

## Congenital dermal sinuses

A congenital dermal sinus consists of a tract lined by stratified squamous epithelium found on or near the midline and is thought to result from an abnormal adhesion between the ectoderm destined to form the neural tube and that which will form the overlying skin. The process appears to be somewhat different from that associated with limited dorsal myeloschisis. Depending upon the extent of incomplete separation, the tract may end in the subcutaneous tissue or extend toward its ultimate embryologic terminus—the conus medullaris for lesions in the lumbosacral region or central canal of the spinal cord for tracts at the thoracic or cervical level. Congenital dermal sinuses are most frequently found in the lumbosacral area with those more rostral occurring

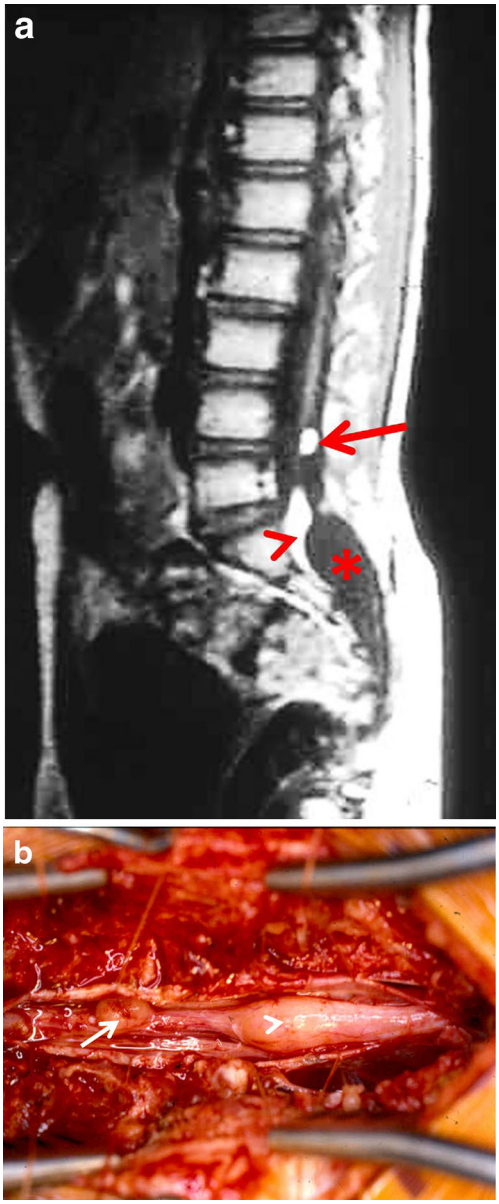



**Fig. 22** **a**, **b** Closed NTD—abnormal filum terminale. **a** T1-weighted sagittal MRI showing a low lying tethered spinal cord with a lipomatous mass at the conus medullaris (*arrow*) and at the filum terminale (*arrowhead*). In addition, an arachnoid cyst was present in the sacral region (*asterisk*). **b** Operative photograph of the findings noted in **a**. Lipomatous tissue of the conus medullaris (*arrow*) and the filum terminale (*arrowhead*). The arachnoid cyst has been fenestrated and is not visible. The filum was divided and the fatty tissue excised

very rarely. As is true with other forms of dysraphism, congenital dermal sinuses frequently, although not invariably, have one or more associated cutaneous markers (Fig. 24a, b). The congenital dermal sinus can dwindle into a connective tissue band or nodule, or enlarge to form one or more dermoid cysts (Fig. 25a–d). Because the connection between the cutaneous ectoderm and neural ectoderm is small during embryologic development, the disturbance of the mesoderm condensing about the tract is minimal. As a result, the

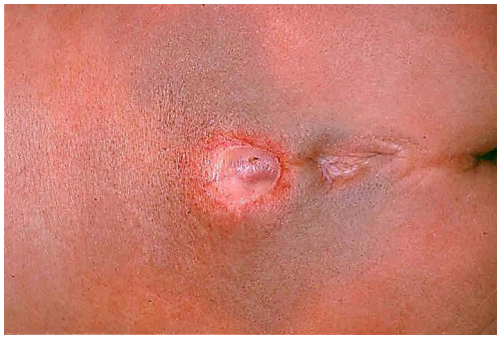


**Fig. 23** Closed NTD—abnormal filum terminale. Two areas of cutis aplasia and associated pigmentary changes in the lumbosacral region. Often present are fibroglial bands that tether the low lying spinal cord in addition to an abnormal filum terminale

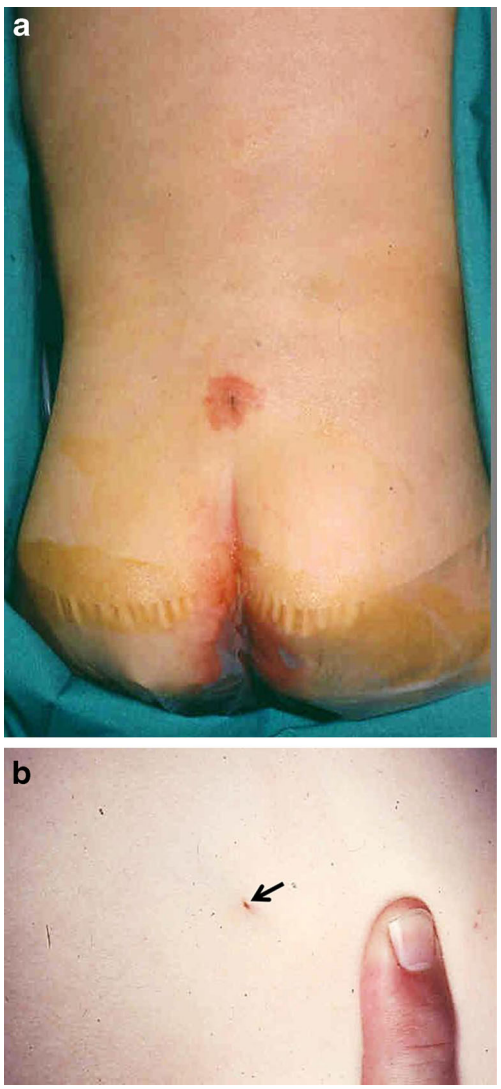



**Fig. 24** **a**, **b** Closed NTD—congenital dermal sinus. **a** A congenital dermal sinus extending from the lumbosacral area to the conus medullaris is present. Hairs are extending from the orifice of the tract. Capillary telangiectasia surrounds the sinus openings. **b** This congenital dermal sinus (*arrow*) is unusual in that no other cutaneous markers were present as is usually the case. This patient had two previous bouts of bacterial meningitis before the tract was identified

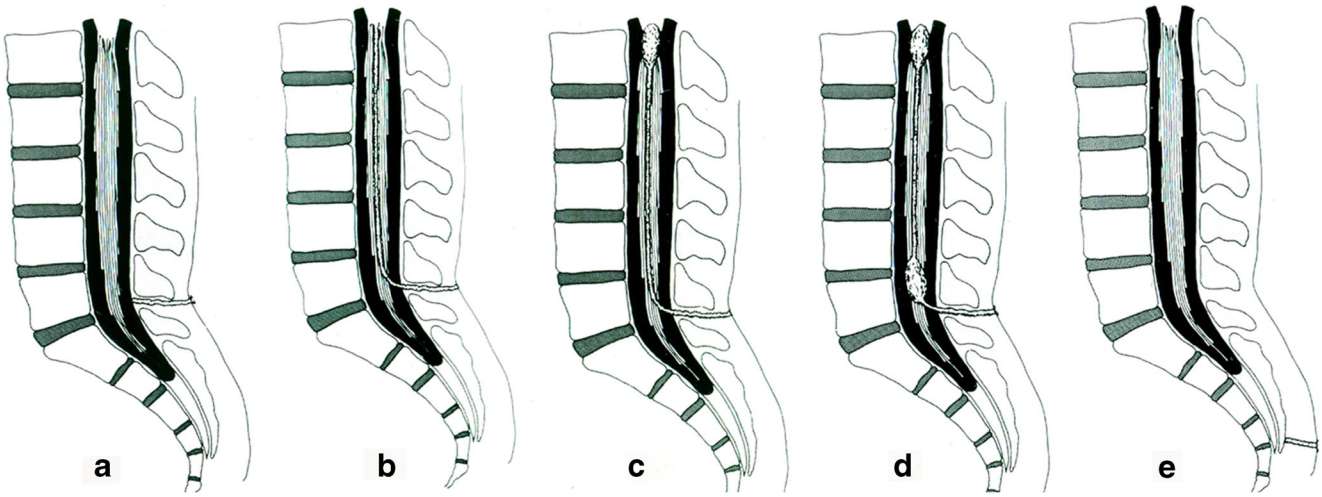



**Fig. 25** **a**–**e** Diagrams of congenital dermal sinus. **a** Congenital dermal sinus extending to the dura matter. Because there is no way of knowing whether the tract extends intradurally and can be a portal of entry for infection, it should be explored and excised. **b** Congenital dermal sinus that extends to the conus medullaris. No dermal inclusion cyst is present. **c** Congenital dermal sinus that expands into a dermoid cyst at the conus medullaris. **d** Congenital dermal sinus that expands into two dermoid cysts. The tract between the two cysts may be small. **e** Dimple in the coccygeal region. A tract in this location ends in the fascia and has no chance of extending to the subarachnoid space. Dimples in this location are fairly common and need not be excised. From McComb [21] (reproduced with permission)

distortion of the adjacent supporting structures of the spine is insignificant unless associated with another NTD (Fig. 26).

A dermal dimple in the coccygeal area (Fig. 27) appears to form by a different embryologic process with the final point of termination of these dimples being the coccygeal fascia, and thus, the tract does not extend to the subarachnoid space (Fig. 25e).

## Split cord malformations

Diastematomyelia is the term that previously has been used to describe the malformation in which the spinal cord is split into two hemicords with each having a single set of laterally located dorsal and ventral nerve roots contained within two distinct dural sheaths (Figs. 28 and 29a, b). Diplomyelia is used to indicate two spinal cord segments that are completely duplicated with a set of four dorsal and ventral nerve roots present within the single dural sleeve (Fig. 30a, b). Pang et al. [16] make a good argument that diastematomyelia and diplomyelia represent the opposite ends of a continuum with few if any "true" examples at either end. Because this group of malformations appears to have a common embryologic disorder, Pang et al. have referred to these lesions collectively as split cord malformations. The error in embryologic development needed to produce a split cord malformation is a ventral disruption in the normal spinal cord preneurulation process. One theory that unifies the spectrum of this group of NTD indicates that the disordered mechanism occurs during gastrulation when all three germ layers are in proximity. It is

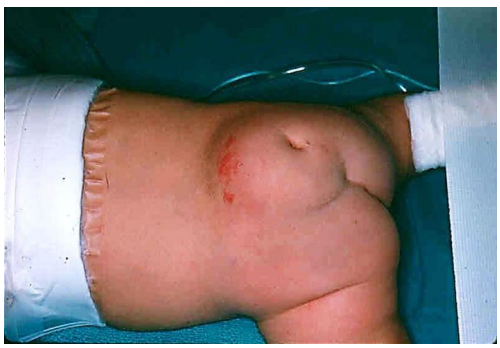


**Fig. 26** Closed NTD—multiple. This infant had a congenital dermal sinus associated with a lipomatous malformation with a low lying and tethered spinal cord. Also present is cutaneous capillary telangiectasia, a common association

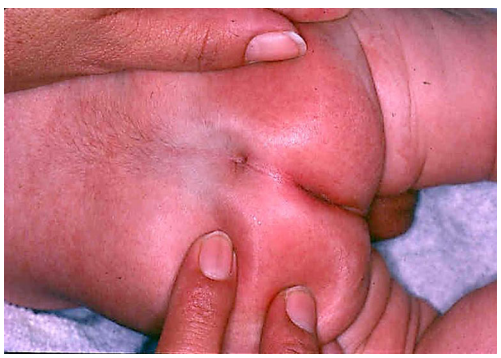


**Fig. 27** Non-NTD. Typical dimple in the coccygeal region. No additional neurocutaneous stigmata are present in this case; however, capillary telangiectasia and skin tags can also be seen infrequently in similar cases. The dimple is approximately within a centimeter of the tip of the coccyx. The point of termination of the dimple is the coccygeal fascia. Thus, the tract does not extend to the subarachnoid space

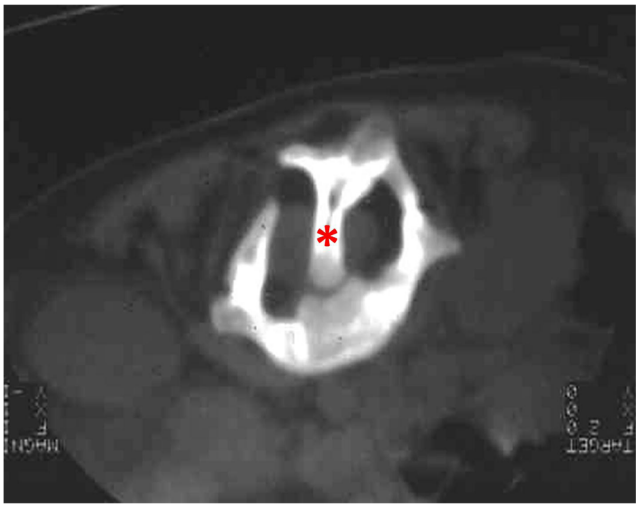


**Fig. 28** Closed NTD—split cord malformation. CT scan of a patient with a split cord malformation. The bony spur originates posteriorly (*asterisk*). Separate dural sheaths also enclose the two portions of the spinal cord

presumed that a second neuroenteric canal becomes invested with mesenchyme to form an endomesenchymal tract that splits the notochord and neural plate, and that the timing and severity determine the extent of the resultant malformation.

Although often the two hemicords are approximately of the same size (Fig. 28), significant discrepancies can occur (Fig. 29a, b). The spinal cord often reunites distal to the cleft (Figs. 31 and 32). When two dural sheaths are present, a bony or fibrocartilaginous spur exists at the caudal end of the cleft and may be attached to the surrounding vertebral bone dorsally (Fig. 28), ventrally (Fig. 29b), or both. The spinal cord is tethered to the spicule as well as medially to the dura mater surrounding the spur (Fig. 31). Frequently, the conus medullaris is low lying and tethered by a thickened filum terminale (Fig. 32). Other neural tube defects, especially myelomeningoceles (Fig. 33) and neurenteric cysts, can be seen associated with the split cord malformations. This embryologic disorder results in a high instance of bony vertebral column abnormalities that include failure of vertebral body segmentation, hemivertebra, butterfly vertebra, a widened spinal canal diameter, and fusion of pedicles, transverse processes, and laminae that can produce kyphoscoliosis (Fig. 34).

## Neurenteric cysts

The embryologic disorder responsible for neurenteric cysts appears to occur during gastrulation, similar to that for a split cord malformation, and in fact, these two types of NTD can occur together (Fig. 35). At the time of gastrulation, the neurenteric canal connects the yolk sac and the amniotic cavity through the blastopore; this connection temporarily links the precursors of the gastrointestinal or respiratory tract, the spine, and the spinal cord. Depending upon the severity, timing, and healing processes, a host of malformations can occur. At the ventral end of the neurenteric canal, the abnormalities include intestinal duplication and malrotation; in the midportion of the neurenteric canal, the malformations that occur are mediastinal enteric cysts, abdominal mesenteric cysts, and neurenteric cysts. The dorsally located neurenteric canal anomalies involve the spinal column and can vary from a minimal disruption to a massive combined anterior and posterior NTD in which the vertebral bodies are divided over a number of segments allowing the intestinal contents to herniate through the defect. The NTD could be open, closed, or both. Such combined anterior-posterior lesions are rare and often not compatible with viability.

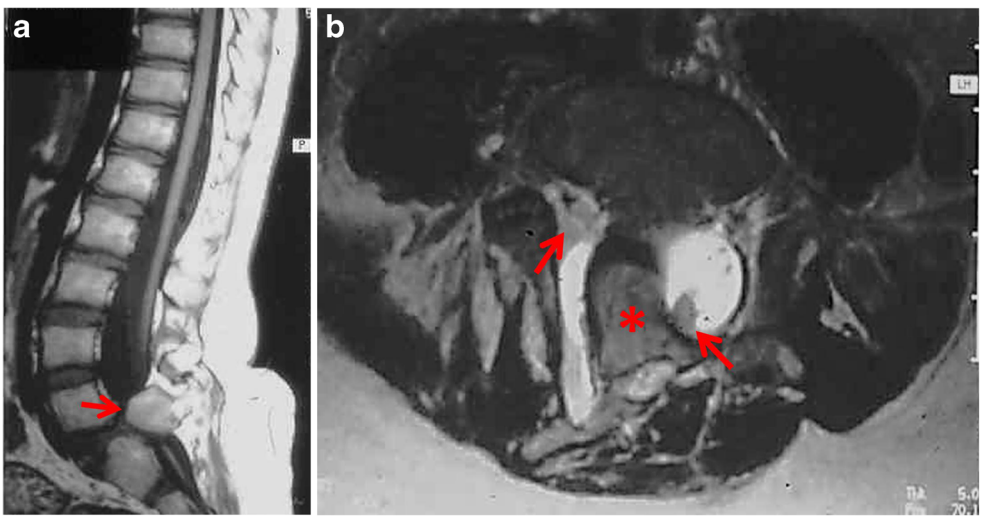



**Fig. 29** **a**, **b** Closed NTD—split cord malformation. **a** Sagittal T1 MRI scan of the lumbosacral spine. The *arrow* points to the bony spur that splits the spinal cord with each hemicord encased within its own dural sheath. **b** Axial T2 MRI scan of the same patient as shown in **a**. The *asterisk* marks the bony spur. The configuration and size of the two hemicords and dural sheaths are unusually asymmetric in this patient. The *arrows* indicate the two hemicords. The *lighter gray* is the CSF within the dural sheath

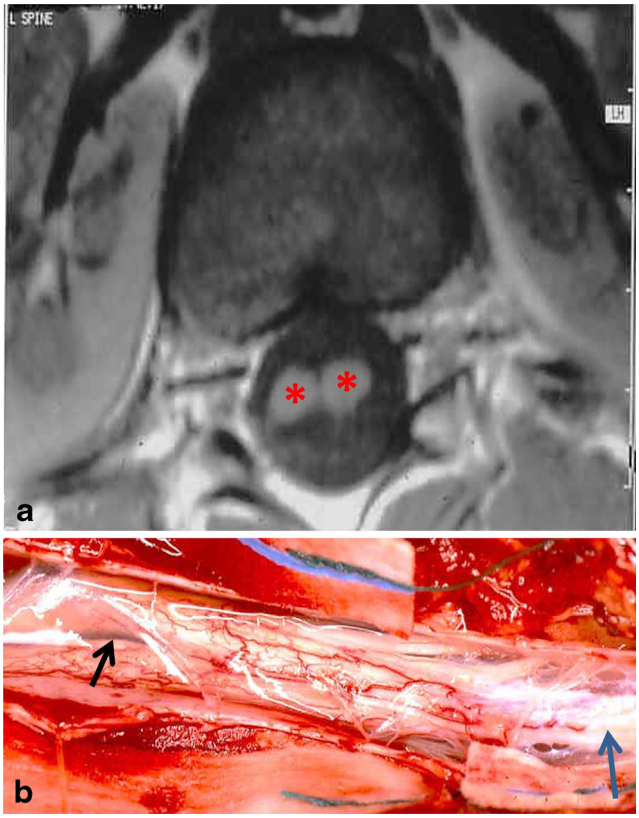



**Fig. 30** **a**, **b** Closed NTD—split cord malformation. **a** T1- weighted axial MRI study depicts the presence of two spinal cord segments (*asterisk*) within a single dural sheath. Fibrous bands can tether the spinal cord in this form of NTD. **b** The *black arrow* points to a cleft between the two hemicords that were tethered by fibrous bands. The *blue arrow* points to the enlarged filum terminale containing fat. The filum was divided and a segment of the abnormal filum removed to reduce the chance of retethering

Neurenteric cysts occur most frequently at the lower cervical or upper thoracic regions and at the conus medullaris. The neurenteric cysts can be extra- or intradural and if intradural, can be anterior to, within or posterior to the spinal cord, or even between, in the case of a split cord malformation (Fig. 35).

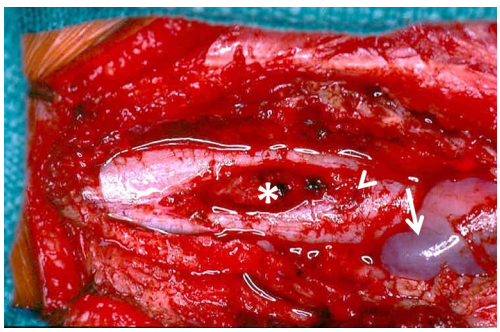


**Fig. 31** Closed NTD—split cord malformation. Split cord malformation with a bony spur (*asterisk*) extending between two segments of the spinal cord that are circumferentially covered with dura mater. Distal to the bony spur, the hemicords reunite and reside within one dural sheath (*arrowhead*). The *arrow* points to an extradural arachnoid cyst

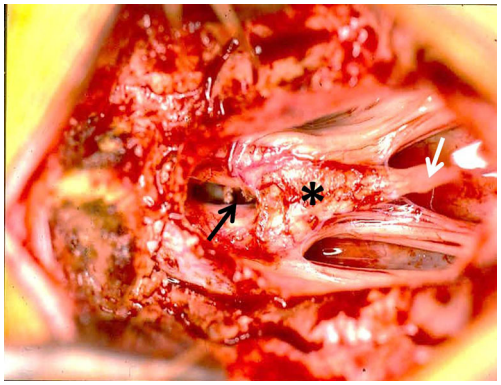


**Fig. 32** Closed NTD—Multiple. An infant with a split cord malformation in which both segments of the cord were within the same dural tube. The *black arrow* points to the space between the two cords and fibrous tissue previously extending between the cord segments. A lipomatous malformation can be seen at the conus medullaris (*asterisk*), and an enlarged filum terminale (*white arrow*) contains adipose tissue. The nerve roots of the cauda equina are divided into two bundles. Split cord malformations can be seen alone or in combination with other NTD

## Neural tube defects associated with caudal regression

Caudal regression, also known as caudal agenesis and sacral agenesis, is a nonsyndromic malformation that results in partial to complete absence of the coccygeal, sacral, lumbar, and lower thoracic vertebrae. In varying degrees, caudal

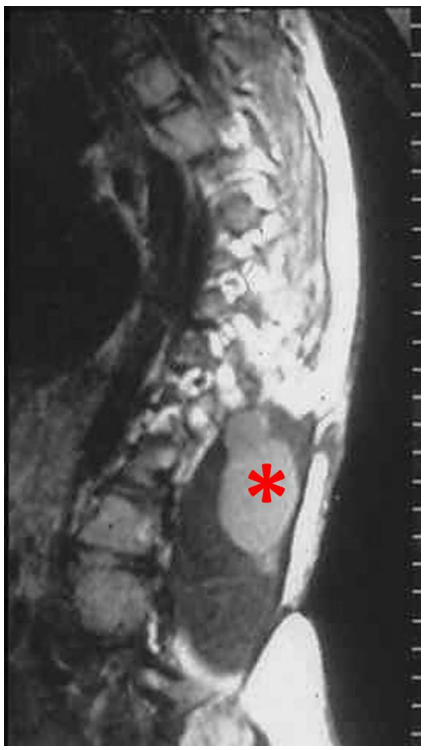


**Fig. 33** Open and closed NTD. T1-weighted sagittal MRIs showing a neurenteric cyst in the lumbar region (*asterisk*). The patient had a previously repaired open NTD (myelomeningocele). A split cord malformation was also present. This example emphasizes the diversity of NTD

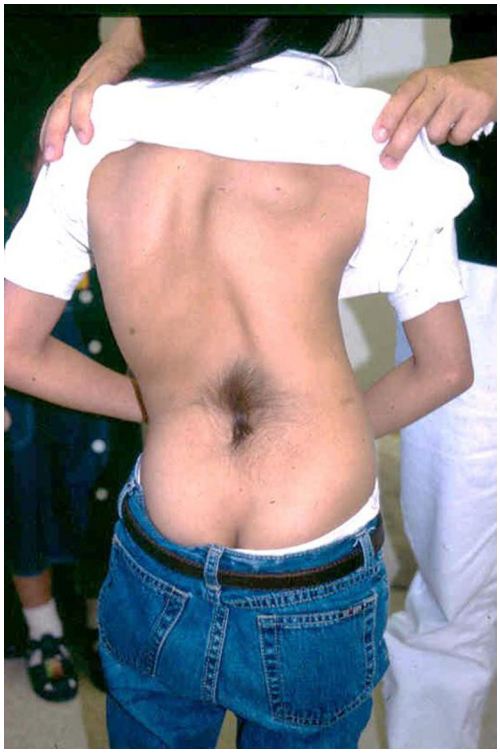


**Fig. 34** Closed NTD—split cord malformation. Hairy patch associated with a split cord malformation. Hypertrichosis of this degree seems to be uniquely associated with split cord malformations especially when each hemicord has its own dural sheath split by a bony spur. Vertebral segmentation abnormalities often produce kyphoscoliosis

regression may be associated with urogenital, hindgut, and neural tube anomalies to produce vertebral anomalies, anal defects, cardiac malformations, tracheoesophageal fistulae, renal anomalies, radial bone anomalies, and limb defects (VACTERL) (Fig. 36), and omphalocele, exstrophy of the bladder, imperforate anus, and sacral agenesis (OEIS) (Fig. 20) groupings. The caudal regression spectrum ranges from asymptomatic and missing lower coccygeal segments

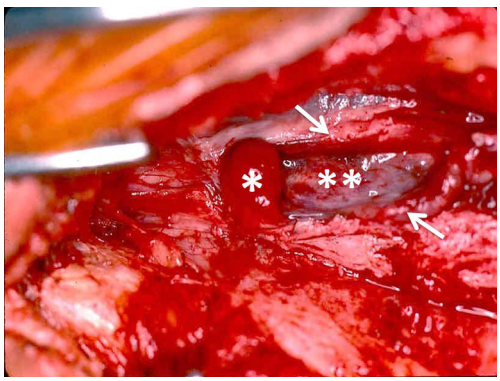


**Fig. 35** Closed NTD—multiple. Neurenteric cyst associated with a split cord malformation (*arrows*). The *white asterisk* is on the solid component of the neurenteric cyst, while the *double asterisk* is on the wall of the cystic component

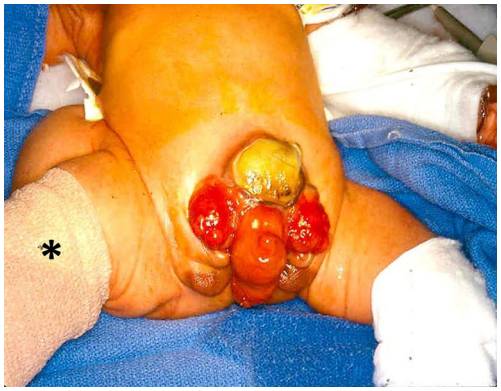


**Fig. 36** Closed NTD associated with caudal regression. This infant has the clustering of anomalies consistent with VACTERL designation. In these cases, the conus medullaris is often blunted, low lying, and tethered. It may also be associated with lipomatous tissue. Visible is exstrophy of the bladder and ambiguous genitalia. The motor deficits in the lower extremities present in this patient were more profound than the sensory, which is often the case. The right lower extremity is splinted (*asterisk*), secondary to fracture during delivery

that go unnoticed to severe malformation resulting in spontaneous abortion or stillbirth.

The embryology is not well understood but appears to stem from the failure of normal cell migration in the primitive streak during gastrulation wherein the three embryonic germ cell layers are being formed. This perturbation produces the multisystem abnormalities that often include the neural tube. The NTD associated with caudal regression most frequently appear to be an abnormal conus medullaris with a thickened filum terminale. This is followed by lipomatous malformations, meningocele, myelocystocele, and split cord malformations [17]. In one series, none of these patients had any cutaneous stigmata often seen with a closed NTD, the presence of such easily overlooked in the face of serious multisystem abnormalities that require prompt surgical attention [17].

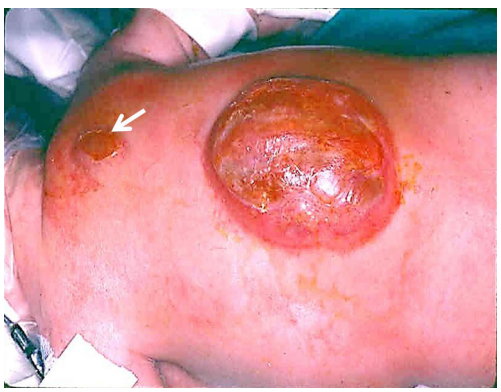


**Fig. 37** Open NTD—multiple. This neonate has two open NTD with the smaller (*arrow*) being high thoracic and the larger thoraco-lumbar. There were obviously two separate areas of disruption in the neurulation process. No function was present in the lower extremities along with a neurogenic bladder and bowel. Present were a Chiari II malformation and hydrocephalus

## Multiple neural tube defects

Although not frequent, multiple NTD can be seen in the same patient. The NTD can be multiple open (Fig. 37), multiple closed (Figs. 32 and 35), or a combination of either (Fig. 33). Their presence would indicate more than one disruption in the neurulation process and may relate in part to the multiclosure site theory. As to the extent of CNS malformations, it is the open component that dominates. It appears that certain populations have a higher incidence of multiple NTD and may reflect genetic factors although the environmental contribution is not known [18–20].

## Conclusions

Current commonly used terminology to describe NTD is inconsistent, overlapping, contradictory and, at times, inaccurate making it difficult to convey the nature of the malformation and what needs to be done to optimally treat patients with these congenital abnormalities.

NTD can be broadly divided into those that are open with exposed neural tissue and leaking CSF and those that are closed with no exposed neural tissue nor loss of CSF. It appears that the loss of CSF during development is the underlying factor that leads to the entire CNS involvement with an open NTD and lack thereof with a closed NTD, wherein only the spinal cord is malformed. There are, however, rare transitional cases that bridge the gap between the two forms.

Agreeing on a nomenclature that is used in a standard fashion would be of help in addressing this group of congenital anomalies that have a great deal of variability and, at times, can be quite complex.

Undoubtedly, exceptions to the classification presented exist; however, it is anticipated that it covers the vast majority of spinal NTD.

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