SPINAL DYSRAPHISM

A Study of Congenital Malformations of the Lower Back

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The marked tendency for a congenital malformation to be associated in the same patient with other errors of development (as for example the Sturge-Weber syndrome) is widely recognised; but it is only in recent years that a great variety of malformations involving the tissues of the lower back have been better understood and treated (James and Lassman 1960, 1964; Till 1965; Yashon and Beatty 1966; Anderson 1968).

The realisation that the spinal cord or the nerve roots may be involved in such malformations has added considerable therapeutic importance to conditions which previously were of only mild interest to the dermatologist or radiologist and occasionally to the orthopaedic surgeon. The initial error in tissue formation in the embryo occurs so early (probably before the seventh week after conception) that the resulting malformation is likely to involve tissues of both mesodermal and ectodermal origin. Because a complete diagnosis is never possible until surgical exploration has been undertaken, it has become convenient to group these disorders under the general title "spinal dysraphism." Not all the anomalies found cause disability. The findings are of great variety and it is important to differentiate as clearly as possible between developmental errors that should be dealt with by surgery and those that are innocuous and require no treatment.

It is convenient to consider the malformations in groups according to the type of error that occurred in the embryo.

Failure of fusion in the midline—Spina bifida is a term covering a variety of skeletal anomalies in which the neural arches fail to meet posteriorly. Unless the gap is wide its presence is only apparent on radiographs. There may be only a slit (often oblique rather than sagittal) or a wider separation of ill-formed and asymmetrical laminar processes. It has been estimated that about 20 per cent of all babies have spina bifida occulta at birth. Obviously only a small proportion of these have intraspinal lesions. However, such lesions are more likely to be present when the spina bifida involves more than one vertebra or when the gap is very wide.

Failure of normal separation of tissues—This may lead to adhesion between the tip of the conus and the dura.

Duplication—This is exemplified by diplomyelia, in which for part of its length the cord is represented by two structures side by side.

Overgrowth of normal tissue—An example of this is a lipoma, which may develop in abnormal situations such as within the spinal cord.

"Herniation" of tissue derived from one germinal layer through tissue derived from another—Examples are the intraspinal cyst lined with enteric epithelium or the dermoid cyst found far forwards in the mediastinum (see the description of "split notochord syndrome").

The abnormalities that are grouped under the heading "spinal dysraphism" include the following: diplomyelia, diastematomyelia (with or without a bony or fibrous structure passing between the two half cords), dermal sinus, dermoid cyst (which may be found in any of the tissue layers between skin and vertebral bodies), neurenteric cyst, fibrous bands between cord and dura, abnormal or supernumerary nerve roots, nerve roots which are abnormally adherent to dura, localised angioma of the cord, lipoma. The justification for grouping so many apparently unrelated findings under one heading is found in the observation that, in many patients, several of the malformations are found together. Moreover, for the most part they

are abnormalities that can be explained on embryological grounds as arising from errors in development during the first few weeks of embryonic life. Spinal dysraphism should be considered as separate from the much more common meningocele and myelomeningocele which are best grouped under the heading of spinal rachischisis. The considerable preponderance of females over males with spinal dysraphism compared with nearly equal sex incidence of myelomeningocele also points to the fundamental difference in origin between the two groups. Rarely the two conditions are found together—particularly diastematomyelia with myelomeningocele.

EMBRYOLOGICAL CONSIDERATIONS

The lower lumbar and sacral levels of the primitive spinal cord develop from a cell mass caudal to the posterior neuropore. Normally this tissue degenerates later and leaves only the filum terminale. Failure of this tissue to disappear leaves a prolongation of the conus attached

FIG. 1

To show partial duplication of the spinal canal.

at its caudal end, this being a common finding in children with dysraphism. Occasionally there is a "double conus" or cord prolongation, which represents the persistence of a normally short-lived stage in the early embryo. The so-called "ascent of the conus" in children is, in fact, slight, being only one segment—from the third to the second lumbar body—in the period from the twenty-sixth week of intra-uterine life to maturity.

The attachment of a "low" or prolonged conus in dysraphism, preventing the normal mobility of the conus during spinal extension and flexion, is probably the cause of neural damage in later life, especially when a lipoma is present.

Many of the forms of dysraphism appear to derive from a sagittal splitting of the notochord as described by Bentley and Smith (1960), in which there is an attempt at duplication of part of the spinal column (Fig. 1). An intermediate form is seen in diastematomyelia with a bony spur. Herniation of entoderm or ectoderm tissue through the midline gap may occur. Figure 2 shows a stoma of ileum which has passed posteriorly through a cleft or split of the spinal cord.

CLINICAL PRESENTATION

The reasons for obtaining medical advice may be concern at visible abnormalities, such as birth marks, or at a later age the appearance of abnormalities of the lower limbs or disturbances of micturition. With improved understanding of these conditions it has become desirable to identify the spinal cord abnormality before neural damage has occurred. Of the patients seen, 24 per cent were referred from orthopaedic surgeons, 31 per cent from paediatricians and 10 per cent from paediatric surgeons.

The frequent occurrence of abnormality of the skin in spinal dysraphism may be manifest in several different ways, a patient commonly having a combination of such stigmata.

Abnormal hair-bearing skin, sometimes known as "faun's tail," which always occurs in the midline, may be a small patch a few centimetres in diameter, but commonly is a wide area roughly lozenge-shaped in the lumbar or lower thoracic levels (Fig. 3). The underlying skeletal

FIG. 2

FIG. 3

FIG. 4

Figure 2—An ileal herniation presenting in the midline posteriorly. Figure 3—An example of abnormal hair-bearing skin. Figure 4—A midline post-anal dimple.

abnormalities are not necessarily confined to this area, and during examination it is important to inspect the skin in the sacral region, where a dimple may be visible.

Skin dimples in dysraphism are always in the midline. They may be so small as to be nearly invisible but usually are several millimetres in diameter and are often attached to underlying bone. They are usually in the post-anal area where they may be found to continue as a fibrous band to become attached to dura in the sacral canal and thence to an elongated conus (Fig. 4). A dimple at a higher level than the sacrum is more likely to be a dermal sinus. It may end blindly or superficially or it may run through the cleft in spina bifida to a dermoid cyst within the spinal canal; it may also have hairs growing from the depth of the pit. Signs of this kind are extremely rare in the sacral area. It is important to realise that the post-anal dimple is not evidence of deeper abnormality but should be regarded as evidence supporting further investigation only when other clinical findings are present.

A congenital scar in the midline of the lumbar region may be present. It usually takes the form of a small irregular area of thin epidermis without pigment.

A flat capillary naevus, although common elsewhere on the body surface, is rare in the lumbar midline except as part of the condition of spinal dysraphism.

Lower limb abnormality—A limp may draw attention to unequal development of the lower limbs, but commonly long before the walking age has been reached a difference in the size of the feet can be detected. One foot may be shorter and narrower than the other or there may be a degree of talipes (often equinus but sometimes curvatus). Careful measurement of the calf or thigh circumference may show a difference which casual inspection does not reveal. On rare occasions the lengths of the lower limbs are different.

Abnormal curvature may be present not only in the lumbar spine but also as a compensatory curve in the thoracic region. Bony abnormality of the spinous processes or laminae (such as laminar fusion or spina bifida) may be obvious on inspection and can nearly always be found on firm palpation. Limitation of spinal movement is only rarely apparent on examination of the young child.

The early diagnosis of spinal dysraphism depends more upon the doctor's recognition of the possible significance of the stigmata than upon the development of any physical disability. Incontinence of urine in particular may not be diagnosed until the second or even the third year of life, and has strangely been accepted as normal in some patients until much later. This is particularly unfortunate because such patients often have no other complaint or abnormal sign. The patients tend to fall into two groups, one with disorders of micturition in whom there is little or no skeletal or muscular abnormality, and the other with abnormality of the lower limbs but with little or no disturbance of micturition.

The discovery of incontinence of urine without an abnormality of the genito-urinary tract to account for it should perhaps always be investigated by myelography, and should certainly be so investigated when midline abnormalities of the skin or vertebral column are known to be present.

Complaint of difficulty in walking or of limping usually cannot be made until after the first year of life. The onset of disturbance of gait may be delayed for several years, and is then of particular significance because it suggests that new neural damage is occurring.

In a small minority of cases the presence of abnormality of the lower back is only revealed by careful examination during an attack of meningitis. It is undoubtedly wise and sometimes very helpful to examine the midline of skull and back of any child suffering from meningitis, although of course the origin of the infection may be apparent through injury or middle ear disease.

The presence of a midline skin dimple or other birth mark without other abnormality on clinical examination is increasingly often the sole reason for referral to the neurosurgeon. If it is accepted that early diagnosis and operative treatment is an effective preventive measure then this early referral will be gratifying, and it usually allows a simpler operation to be done during the first year of the child's life.

Sensory changes, usually in the skin of the toes of one foot, are often not detectable even on careful examination, but there is often a history of slow healing of minor wounds. Chronic or recurrent ulceration of the skin of one toe has indeed been the reason for further investigation in some of the older patients.

The conditions grouped under the heading spinal dysraphism are much more common in girls than in boys: there were seventy-nine girls and thirty-three boys in the author's series confirmed by operation. Stigmata such as post-anal dimples and even hairy patches of the back are probably more commonly unassociated with abnormality within the spinal canal in boys than in girls. It follows that the likelihood of finding abnormal myelographic appearances in the male must be less than in the female although the sex of the patient can never be used as a contra-indication to this investigation.

RADIOGRAPHIC FEATURES

Plain radiographs—It is improbable that tissues of mesodermal origin would remain normal in those areas where there is maldevelopment of neuro-ectodermal tissues. In none of the patients so far encountered has the spinal column been normal. The anomalies revealed by plain radiographs involve the neural arches primarily but also vertebral bodies and intervertebral discs. The neural arches may be divided or fused one with another, and are nearly always deformed. The pedicles commonly show increased separation, indicating a widening of the spinal canal. This latter finding is strongly suggestive of either a lipoma or a split cord, but myelography sometimes shows that the canal is widened merely as a growth abnormality without any other explanation. The vertebral bodies may be split sagittally, a condition which suggests the presence of a neurenteric cyst derived from entoderm.

The intervertebral disc may be narrowed or the vertebral bodies even fused; especially is this likely to be so at the level of diastematomyelia with bony spur (Fig. 5). The latter may be seen in the postero-anterior radiograph but is sometimes so small as to be difficult to distinguish. In the cases so far verified at operation spina bifida was present in 72 per cent, widened spinal canal in 63 per cent, midline bony spur in 36 per cent, abnormal lamina in 45 per cent and fused vertebral bodies or narrow disc space in 25 per cent. Burrows (1968) discussed in detail the radiological findings.

Myelography—It is unwise to draw definite conclusions from the plain radiographs because they commonly reveal only part of the spinal cord or root abnormality. Myelography is necessary if a more complete diagnosis is to be obtained. A clear indication for myelography is a history of progressive neural involvement such as lower limb weakness and the development of urinary incontinence or recurrent urinary infection. When abnormality of lower limb or micturition is present without definite evidence of deterioration, myelography is perhaps less clearly indicated. If plain radiographs show skeletal abnormalities of the types already described myelography should be done; a further indication is when the plain radiographs indicate the presence of an expanding lesion as shown by a widened spinal canal or thinned pedicles. Unfortunately an operation usually has very little effect upon neural or developmental abnormalities already present, or upon dysuria. It has therefore become the author's policy to perform myelography whenever clinical or radiological stigmata of dysraphism are present even in the absence of lower limb or sphincter disturbance. In this way an increasing number of young children with spinal dysraphism have been detected and operated upon in the belief that later deterioration and more difficult operation are thus prevented.

Technique of myelography—Lumbar puncture is contra-indicated in these children because the conus is low and a lipoma or other lesion may prevent the finding of cerebrospinal fluid.

Cisternal puncture is therefore used for the injection of air with the anaesthetised child placed at an angle of 15 degrees with the head down. Films are exposed in lateral projections in both supine and prone positions. Further exposures are made as necessary.

FIG. 5
Radiograph of a spine showing wide lumbar canal, hemivertebrae and thin bony spur in the midline.

INDICATIONS FOR OPERATION

Myelography, done for the indications outlined above, may not have shown any abnormality. There may then be little or no justification for spinal exploration, but this is not always so: the point will be discussed later.

When myelography has revealed an abnormality to which the clinical condition may well be attributed then spinal exploration is warranted. This is not merely because the lesion has been demonstrated but also because experience shows that further anomalies which need correction are often found although not shown by myelography. The clear-cut indications for operation therefore include the demonstration of an expanding lesion such as dermoid cyst, of a conus lying at a level below the normal whether or not the reason for this is demonstrated, the attachment of cord or conus to a mass which may be presumed to be lipoma and the fixation of a split cord with conus at a lower level.

A history of recurrent meningitis in a child with a midline skin dimple is sufficient indication for spinal exploration even though the myelograph is normal. The dissection must be carried to the dura and it is then sometimes evident that the sinus continues to the subarachnoid space and that an expansion of it may have formed a dermoid cyst. The normal myelograph should not dissuade the surgeon from operating because it is essential to prevent what would otherwise be almost certain further infection.

If there is an undoubted history of neural deterioration, and if there are stigmata—either of the skin or on plain radiographs—then exploration of the lumbar and upper sacral canal is probably justified on occasion even though the myelograph is normal. However, in the absence of evidence of clinical deterioration it is probably best to observe this small group carefully at intervals of not more than three months during the first few years of life, although it has not been possible so far to be certain that this is correct. Only by careful follow-up of these children will the matter be resolved.

OPERATIVE PROCEDURE

All the patients have been operated upon under endotracheal inhalation anaesthesia with the child in the prone position. Because most of the operations are at the lower lumbar and sacral levels particular care must be taken in skin preparation to avoid contamination from the peri-anal region. Contamination after operation is prevented by the use of waterproof adhesive tape over the lower part of the dressing. The dissection required is, in many patients, made difficult by the abnormal anatomy. It is, however, not often necessary to remove bone to expose the spinal canal. In extensive spina bifida the ligament which roofs the canal may be cut without difficulty and retracted. If an extradural lipoma is present—which may extend to the cord—then particular care has to be taken. Cutting with diathermy, which is so satisfactory in the more superficial part of the lipoma, becomes potentially more dangerous as the neural tissue (often of unknown extent) is approached.

The main purpose of operation is to free the cord from any immobilising structure. A spur of bone and its enveloping dura are removed; tethering bands are cut; an elongated or attenuated conus attached to dura is freed; and an intraspinal lipoma with an attachment to an extraspinal extension needs incision—often in an arbitrary manner because the conus and sometimes roots are attached in such a way that dissection would be harmful. Further aims of operation will sometimes include the removal of a dermal sinus or of a dermoid cyst. Total excision of a cyst may be impossible if it is attached to the conus or if it is intramedullary. Operation at two levels (usually undertaken at different times) may therefore be necessary when, for example, there is conus attachment in the sacrum and diastematomyelia at a higher level.

FINDINGS AT OPERATION

It will be seen from Table I, which lists the principal lesions found, that many patients had a combination of abnormalities. The sixty-one patients with “tethered conus” were those in whom the spinal cord extended to an abnormally low level—usually the upper sacrum—with no filum terminale. The group with a split spinal cord included several where

the half cords did not rejoin caudally and continued to the sacrum as a split conus. Diastematomyelia should no longer be regarded as a complete diagnosis because most of the patients had an abnormal attachment of the conus at a low level. Figures 6 and 7 illustrate some of the findings at operation.

TABLE I
FINDINGS AT OPERATION

Split spinal cord . . .	45
Tethered conus . . .	61
Tethering bands to conus	26
Intraspinal lipoma . . .	24
Dermoid cyst . . .	9
Hamartoma . . .	3
Angioma . . .	1
Neurenteric cyst . . .	2

RESULTS OF TREATMENT

The purpose of spinal exploration in patients with dysraphism is essentially to reduce the possibility of later deterioration either of sphincter function or of lower limb development. It is unlikely that any neural deficit present before operation will be reduced. The assessment

FIG. 6

FIG. 7

Figure 6—Photograph at operation showing the gap between two portions of split cord after removal of a bony spur. Figure 7—Photograph at operation showing a double conus. Note absence of emerging nerve roots.

of the effect of surgery of a preventive or prophylactic type is particularly difficult. It has not been thought proper even in the earlier stages of this work to attempt random selection for the formation of a control group, and a study of untreated patients has not been found in the literature. During the first few years investigation by myelography was undertaken only if there was clinical evidence of deterioration. Despite examination of the patient every three months there were some who showed deterioration during the waiting period. Thus it gradually became evident that it was wiser to investigate by myelography all those children who had evidence of dysraphism. Therefore the age at operation became younger. In two patients there was definite operative damage with weakness of a limb. One of the later patients recovered to the state before operation, but in the other the limb weakness remained. In no other patient has there been evidence of deterioration because of operation. In no patient has deterioration continued after operation. In several patients with weakness of one lower limb there was improvement in power and muscle bulk during the year after operation. In three children whose habits of micturition had previously been thought to be normal the mothers reported a reduction in frequency which appeared to be in fact a change towards normal. From these remarks it will be clear that the parents must be informed of the preventive nature of the treatment and must not be led to expect any improvement of disability that already exists. Many of the children who were first seen near puberty have been particularly difficult to treat surgically, and there is a strong suspicion that if the diagnosis had been made during early childhood—which would have been possible in all—it would have led to much simpler, and probably far more effective, operative management. There can be no reasonable doubt that excision of expanding lesions, such as a dermoid cyst or a neurenteric cyst, is likely to be far easier in infancy than after the passage of a few years. In none of the children with a history of meningitis or a dermal sinus or cyst has there been infection during the follow-up period after operation.

SUMMARY AND CONCLUSIONS

1. A group of children with congenital malformations of the lower back involving the spinal cord or nerve roots is described. The malformations have little or no resemblance to spinal rachischisis, either embryologically or clinically. The lesions found are often complex.
2. It is considered from clinical and operative findings that the malformations are, in many patients, responsible for increasing neural damage in childhood and even in later life.
3. Early investigation and operative treatment are recommended.

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