Review Article

Current Trends in Spinal Dysraphism

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Spina bifida cystica is the 'most complex, treatable congenital anomaly consistent with life'.¹ Substantial past writings have documented case experience and analysed outcomes, hydrocephalus treatments and attendant problems occurring in the child with myelomeningocele.^{16, 18, 20, 22, 24} Yet there has probably been no period other than the present which is characterised by such exciting events with regard to the analysis of spina bifida, its epidemiology, pathophysiology, the pre- and postnatal imaging details, early treatments and the delayed features. This paper will review some of the current issues applicable to both the 'open' and 'closed' forms of spina bifida.

Aetiology

Present clues suggest that neural tube malformations arise as the result of exposure of embryos 'genetically at risk to additional intra-uterine environmental triggers'.⁴ The search for a responsible teratogen has examined a number of alleged dietary causes, and Laurence has marshalled the evidence in support of poor maternal nutrition, in particular folic acid deficiency.¹⁷ Aminopterin, a folic acid antagonist has been considered for its possible aetiological role as it is a human teratogen and such agents have caused neural tube defects (NTD) in mice, cats and rats.²⁵ Smithells *et al* studied the recurrence risks of myelodysplasia in a non-randomised, non-controlled study of multi-vitamin supplementation.²⁵ The recurrence rates were substantially lower in the group protected with peri-conceptional multi-vitamin supplementation. More recently, Milunsky *et al* examined the relation of multi-vitamin intake in general and folic acid in particular, to the risk of neural tube defects.²⁵ The prevalence of such defects was 3.5 per 1000 among women who never used multi-vitamins before or after conception or who used multi-vitamins before conception only. The prevalence of neural tube defects for women who used

folic acid containing multi-vitamins during the first 6 weeks of pregnancy was substantially lower—0.9 per 1000.

Still, the aetiological answer may be a long time coming.² Meanwhile, anecdotal and reported experiences from many North American and European centres indicate that for whatever reason, the incidence of open neural tube defects presenting to major paediatric neurosurgical centers has declined by at least 40% during the past 5 years.^{3, 19, 29}

Pre-natal diagnosis

Pre-natal ultrasound examinations notwithstanding, most open neural tube defects in newborns are not declared until that moment in delivery when the lower half of the trunk and legs make their appearance. But sophisticated pre-natal screening tests can help identify affected fetuses especially in women with a family history of NTD or a known risk factor. Maternal serum alphafetoprotein (AFP) levels which are elevated in open NTD can be determined as early as the eighth week of gestation, and if elevated, the testing repeated and correlated with amniocentesis and amniotic fluid AFP levels. Simultaneously, high resolution ultrasound performed after the twentieth week will detect about 80% of fetuses with open spina bifida. Unlike AFP testing, the ultrasound may also uncover some cases of skin-covered meningocele or lipomyelomeningocele. Together, amniocentesis and level II ultrasound will detect 96 to 100% of cases of spina bifida aperta. But practically, the confirmation of this diagnosis and the associated hydrocephalus if present does not occur before 24 to 28 weeks of gestation at which point the pregnancy and fetal management decisions become most vexing.

Open neural tube defects

McLone has itemised the 'significant progress' which has occurred in the past 25 years with the management of myelomeningocele,²² viz.

- 'Mental retardation is acquired post-natally
- '75% of surviving infants will have normal intelligence
- 'Bladder and bowel control can be achieved by school age in almost 90% of surviving children
- 'Greater than 80% of myelomeningocele patients will be community ambulators by school age
- 'Between 10–15% of surviving children in an unselected population of myelomeningocele patients are likely *not* to be socially competitive.'

The optimism which he has expressed arises after years of study of this problem and serves to buttress his recommendation that 'all children born with a myelomeningocele should have the lesion repaired surgically within 24 hours of birth'. This traditional stance is also supported by others.^{18, 21}

Not much has changed during the past several years with regard to the early evaluation and technical care for the infant with myelodysplasia. Surgeons continue to fine-tune operative closure techniques, urological laboratory analyses of sphincter performance have become increasingly detailed and multi-discipline follow-up clinics ensure co-ordinated, continuing care. Rather, there is now much more concern with just what we are doing. The philosophic debates continue as we

are exhorted to pay attention to qualitative outcomes and the social and family impacts—'the rest of the story'.^{10, 11} Proving that there is no right solution for the early management of myelomeningocele, some authors have retreated from the need for urgent closure of the back^{6, 24} or have even suggested non-closure.⁷ When assessing all of these factors, we are left wondering whether we are too sophisticated if we can only measure outcomes in terms of paraplegia, ventriculitis, hydrocephalus and intellect?

MRI examinations of the craniovertebral junction and spinal cord of patients with treated myelomeningocele reveal what the pathologist has previously recognised.⁸ There may be rostral areas of cord atrophy, or hydromyelic cavitation or ventral compression from arachnoid cysts in addition to the features of the repaired (and 'tethered') placode, and the Chiari malformation. This new information emendates the earlier belief that gradual deterioration in neurological function was the inevitable natural history for children with myelomeningocele. The delayed complex clinical situation should it occur usually appears after age 5 years and requires a 'top down' investigative and therapeutic approach.

Closed Spinal Dysraphism

Unintended as an oxymoron, 'closed' spinal dysraphism refers specifically to those occult forms of spina bifida which are associated with a hallmark on the otherwise intact surface ectoderm. The characteristic cutaneous feature—a nevus, hemangioma, hairy patch, subcutaneous lipoma or dermal sinus—is present at birth, usually located over the midline of the lumbar spine. The cutaneous feature signals a dysraphic spinal lesion which should be subject to early documentation and prophylactic treatment.

Lipomyelomeningocele

The lipomyelomeningocele, of all the spinal dysraphic conditions has been until recently, mired in confusion. The terminology and classifications were imprecise, embryologic anatomy very disordered and the recommendations for therapy controversial.⁵ However, there is now consensus amongst paediatric neurosurgeons that the subcutaneous spinal lipoma associated with spina bifida is not harmless and that it must be properly defined and corrected before inevitable neurologic deterioration occurs.^{5, 9, 14, 23, 27} For, the subcutaneous lumbar fatty mass extends through the dysraphic posterior elements and dura (which it may partly replace) and 'tethers' the spinal cord.

A neurosurgical opinion is initially sought in many instances for cosmetic reasons, but delayed neurological deterioration due to cord tethering with growth seems inevitable. There is a 56% risk of spontaneous deterioration without surgery and currently detailed urological investigation is confirming that many infants already have sphincter compromise at first testing.²⁷ A meticulous releasing and debulking operation is recommended for patients with established neurological findings (eg. calf, foot, ankle, motor weakness or urinary incontinence), as well as in those infants who appear normal. The results of operation are age dependent and at least 40% of patients will show improvement in their neurological performance.²³

Diastematomyelia

It is paradoxical that one of the more intriguing dysraphic anomalies can at this time of precise neuro-imaging be readily diagnosed with routine plain spine X-ray films. The clinical and radiological features of this abnormality, characterised by a split spinal cord with each segment residing in its own dural tube, are well established.^{12 15} Not so clear are the pathogenetic factors. In addition to those customarily listed, Pang *et al* argue that all split cord malformations probably result from dorsal migration of an entomesenchymal tract through the notochord and the neuroectoderm, so that a split spinal cord and bifid vertebral body are features common to all variants.²⁶ They suggest elimination of the early confusing terminology ('diastematomyelia' and 'diplomylelia') in favour of the 'split cord syndrome'. They recommend that all patients with splitting of the cord be operatively explored to rule out tethering of the cord.

Tethered spinal cord

Intended as a very specific term, the 'tethered spinal cord' when associated with spinal dysraphism has taken on a more generic meaning.¹³ Initial reports referred to traction on the conus medullaris caused by a thickened and tight filum terminale. In the developing fetus, the vertebral column lengthens more rapidly in a caudal direction and the spinal cord and conus ascend the canal to reach approximately the L3 level at about 30 weeks gestation. With further growth, the portion of the conus formed by the caudally placed cell mass regresses to become the filum terminale, which normally should remain thin and slightly relaxed to allow for further ascent of the cord and conus.

If the filum develops abnormally into a short, thick anchor, then the conus may be trapped at an abnormal level. Of interest is that the neurological and/or orthopaedic disturbances associated with a tethered conus, usually appear during a rapid growth period in early childhood and adolescence. Given the presence of the features of the neurological and/or orthopaedic 'syndromes',¹² in a child who is proven to have posterior spinal dysraphism, then CT metrizamide myelography or MRI examinations are required.

If the filum can be identified and is shortened and thick, and the conus appropriately tethered below its anticipated high lumbar level, then operative division of the filum is recommended to prevent inevitable, further neurological deterioration with growth.

Conclusion

Lorber and Ward have examined the 'vanishing nightmare' of spina bifida.¹⁹ To the extent that such refers to myelomeningocele, there is no doubt that open neural tube defects in newborns are less prevalent now than just a decade ago. But, one's individual case encounters with children suffering from either the 'open' or 'closed' forms of spina bifida provoke substantial medical, surgical, rehabilitation, ethical, social and resource issues. Fortunately, the health care team has never before been so informed and better equipped to manage the challenge of a child's spinal dysraphism.

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