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SOME PAEDIATRIC ASPECTS OF MYELOMENINGOCELE

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This review is written from the paediatric point of view and is concerned with:

(1) recent advances in the antenatal detection and the prevention of myelomeningocele,
(2) the changes in incidence and pattern of myelomeningocele,
(3) the selective approach to treatment of the newborn,
(4) the need for selective approach to aggressive orthopaedic policy and practice.

1. *Prevention of myelomeningocele*

We still do not know the cause or causes of spinal dysraphism but advances made since 1972 promise the eventual elimination of at least the more severe cases. Brock & Sutcliffe (1972) found that alpha fetoprotein, which is a normal constituent of foetal serum and of amniotic fluid, is present in excessive quantities in the amniotic fluid when the foetus has an open neural tube defect. Fortunately this excess is most marked between the 16th and 18th week of pregnancy, when it is easy to carry out amniocentesis and when it is still easy to terminate pregnancy, if need be, by an intraamniotic injection of prostaglandin F_2. Milder and closed cases of myelomeningocele may be missed by this technique, but false positive results have not been seen. Subsequently these findings have been confirmed by many groups (Allan et al. 1973, Lorber et al. 1973, Harris et al. 1974, Milunsky et al. 1974, Nevin et al. 1974, Stewart et al. 1975). In Britain it is now routine practice in major units to carry out amniocentesis at 16–18 weeks of pregnancy in high risk cases, that is, when a mother has a child with spinal dysraphism already or where there is a family history in close relatives.

The alpha-fetoprotein level also should be determined in amniotic fluid samples obtained for other purposes. An abnormal result is followed by termination of pregnancy. While this method is of great

value in high risk families, it will prevent the birth of only 5 per cent of all cases of myelomeningocele, because it is not practicable to carry out amniocentesis in all pregnancies.

It was therefore necessary to look for more universally applicable techniques. It was found that alpha-fetoprotein is normally present in maternal serum but is often raised if the foetus has a neural tube defect. Even so, as the values are measured in nanograms, its estimation requires a highly sensitive radio-immune assay. At present these are done individually in a few research centres, but mass testing techniques are being developed and soon we shall be able to monitor all pregnancies with a simple blood test, probably between the 14th and 18th weeks of pregnancy. A positive or doubtful result is followed by amniocentesis to confirm the diagnosis. From the relatively few data so far, it appears likely that approximately half the cases of spina bifida could be detected by this method. As these will be the more severe cases, the beneficial effects and the major implications for future reduced need for orthopaedic services are evident (Brock et al. 1973, 1974, Seller et al. 1974, Wald et al. 1974).

2. *Decreased incidence of myelomeningocele*

Antenatal detection and termination of pregnancy is not the only reason for a decrease in the number of babies born with myelomeningocele. The rapidly falling birth rate may well be responsible for a 30 per cent drop; this was the decrease of birth rate in Britain between 1968 and 1973 and the fall is continuing.

Table 1. Spina bifida cystica in Sheffield residents.

	Liveborn (treated)		Stillborn
1968–1970	46	(41)	7
1971–1973	20	(4)	10

In addition there are other, unknown factors which led to a dramatic decrease of spina bifida births in Sheffield. Up to 1968 an average of 18 such infants was born each year. Since then, fewer and fewer affected babies have been born; by 1973 the number was only 6 and in 1974 it seems to have been only 4 (Table 1). Neither the antenatal detection nor the fall in birth rate can account for such a large and consistent drop. Whether this will be a future pattern and whether

such a change may occur elsewhere, too, is not yet apparent. There is little doubt, however, that in future we shall have fewer such infants and their orthopaedic problems will be less complex.

3. *Selective treatment*

The next major change which affects the orthopaedic case load is the selective approach to the treatment of myelomeningocele. During the 1960s practically all infants born with myelomeningocele were treated. This world-wide policy was largely the result of the example of the Sheffield school (Sharrard et al. 1963), where we treated, without selection, 1191 newborn infants between 1958 and 1969. The 4-year survival rate progressively increased to over 60 per cent in spite of tackling the most severe cases, but the long-term results showed that this increase in survival rate has led to a very poor quality of life in the majority. In most cases survival produced such immense personal, family, social, surgical, administrative and financial problems that one had to take stock afresh. One had to come to the conclusion that the policy of treating all cases is not ethical and not justified. Accordingly, one had to search for criteria at or soon after birth which would identify those who do not have a chance of an acceptable quality of life. At the same time one had to be sure that no infant who had a chance of living with moderate handicaps would be excluded from total, modern treatment. Finally it was essential to establish such criteria that untreated survivors would not live long, and so produce an even more serious situation than if they had been treated from birth (Lorber 1971).

In other publications I have outlined in detail the long-term results related to various clinical signs which are readily detectable at birth by an expert in this field, without need for elaborate or time-consuming investigations (Lorber 1972, 1974 a). I found that if any infant had one or more of the specified adverse clinical signs, then the survival rate was less than 50 per cent and the survivors had such severe multisystem defects that none had an acceptable quality of life. None could hope to earn their living in competitive employment and none could reasonably hope for marriage and normal family life. All would have to depend on charity and life in institutions once their parents were too old or no longer alive to look after them. Two-thirds of the survivors of this unfavourable group were moderately or severely retarded, but it was the more intelligent who suffered most, especially when they

reached adolescence and understood all that they missed and all the problems that lay ahead of them.

These adverse signs, which subsequently served as criteria against any form of active treatment, are as follows:

(1) Large thoraco-lumbar or thoraco-lumbo-sacral lesions related to vertebral level.
(2) Extensive paralysis with a motor level at L3.
(3) Kyphosis or scoliosis clinically present at birth.
(4) Gross hydrocephalus with a maximal head circumference which exceeds the 90th percentile by at least 2 cm.
(5) Other gross congenital malformations, e.g., cyanotic heart disease.
(6) Gross cerebral birth injury.

In addition, social factors have to be taken into account, especially in borderline, clinically more favourable cases. The moderately affected unwanted, abandoned infant has a very poor chance for an acceptable quality of life.

Finally, treatment is contraindicated if a badly handicapped infant develops meningitis in the newborn period following closure of the back or the surgical treatment of hydrocephalus (Lorber 1974 b).

I have not included among the adverse criteria urinary or faecal incontinence, although this is a profoundly serious handicap, medically and socially. If one did include this, hardly any spina befida baby would be treated today.

Some orthopaedic surgeons naturally question the validity of excluding infants if the neurological level of their lesion is at L3 and not higher. However, many such infants lose muscle power above this level in spite of early closure; all are incontinent, at least half develop hydronephrosis, all have hydrocephalus and will be at risk of multiple operations and often death from complications of shunt therapy.

Between July, 1971 and 1974, 50 infants with one or more of these criteria were not treated except by normal nursing care. The longest survival was 8 months, and half were dead by 1 month. None lived to need orthopaedic procedures.

Such a policy of selection is now almost universal in Britain and is followed by many countries around the world. The impact on orthopaedic case load will become progressively more evident as the years go by.

4. At present there are still large numbers who need orthopaedic

care, often on a massive scale. It is not within my competence to discuss the surgical aspects of orthopaedic care. No doubt, many procedures are of great value and a planned series of operations based on neurological principles can often improve the quality of life. Nevertheless, there are also many disappointments after initial successes and sometimes the situation is worse after major procedures. I would urge therefore that before any series of operations are planned, the whole child should be taken into consideration, especially his intellectual level. The success of many operative procedures depends on the patient's intellectual ability and his will to make the most of what the surgeon hoped to achieve. It is no use use putting hips back into joint if the child is so retarded that he would never walk even if he had much more muscle power. It is harmful and a waste of time to carry out heroic procedures on patients who lack a heroic will to maintain the structural improvement. It is also essential to consider the child's emotional background: taking him away from his family or school may do him permanent emotional harm and interfere with vital education. It is of no use trying to make a child walk who has not the will to struggle slowly with calipers, when he can get on faster and with less effort in a wheelchair. It may be immensely harmful to occupy his hands with weightbearing when he so desperately needs the finer hand skills to earn a living. Most badly paralysed adolescents will take to their wheelchair whatever we advise and it is important to recognise this. I would like to close my paper with an appeal to orthopaedic surgeons to concentrate their energies on the more promising cases, however hard it seems. A rational, selective approach is likely to benefit those who can truly benefit by your skills, and will save much time and false hope in the others.

REFERENCES

Allan, L. D., Ferguson-Smith, M. A., Donald, I., Sweet, E. M. & Gibson, A. A. M. (1973) Amniotic-fluid alpha-fetoprotein in the antenatal diagnosis of spina bifida. *Lancet* ii, 522–525.

Brock, D. J. H. & Sutcliffe, R. G. (1972) Alpha-fetoprotein in the antenatal diagnosis of anencephaly and spina bifida. *Lancet* ii, 197–199.

Brock, D. J. H., Bolton, A. E. & Monaghan, J. M. (1973) Prenatal diagnosis of anencephaly through maternal serum-alphafetoprotein measurement. *Lancet* i, 923–924.

Brock, D. J. H., Bolton, A. E. & Scrimgeour, J. B. (1974) Prenatal diagnosis of spina bifida and anencephaly through maternal plasma alpha-fetoprotein measurement. *Lancet* i, 767–769.

Harris, R., Jennison, R. F., Barson, A. J., Laurence, K. M., Ruoslahti, E. & Seppälä, M. (1974) Comparison of amniotic-fluid and maternal serum-alpha-fetoprotein levels in the early antenatal diagnosis of spina bifida and anencephaly. *Lancet* **i**, 429.

Lorber, J. (1971) Results of treatment of myelomeningocele. *Develop. Med. Child Neurol.* **13**, 279–303.

Lorber, J. (1972) Spina bifida cystica. Results of treatment of 270 consecutive cases with criteria for selection for the future. *Arch. Dis. childh.* **47**, 854–873.

Lorber, J. (1974 a) Long term results of treatment of myelomeningocele in infancy. In: *Progress in paediatric neurosurgery*, ed. Bushe, K. A., Spoerri, O. & Shaw, p. 104–110. Hippokrates-Verlag, Stuttgart.

Lorber, J. (1974 b) Selective treatment of myelomeningocele: to treat or not to treat? *Pediatrics* **53**, 307–308.

Lorber, J., Stewart, C. R. & Milford-Ward, A. (1973) Alpha-fetoprotein in antenatal diagnosis of spina befida. *Lancet* **i**, 1187.

Milunsky, A. & Alpert, E. (1974) The value of alpha-fetoprotein in the prenatal diagnosis of neural tube defects. *J. Pediat.* **84**, 889–893.

Nevin, N. C., Thompson, W. & Nesbitt, S. (1974) Amniotic fluid alpha-fetoprotein in the antenatal diagnosis of neural tube defects. *J. Obstet. Gynaec. Brit. Cwth.* **81**, 757–760.

Sharrard, W. J. W., Zachary, R. B., Lorber, J. & Bruce, A. M. (1963) A controlled trial of immediate and delayed closure of spina bifida cystica. *Arch. Dis. Childh.* **38**, 18–22.

Seller, M. J., Singer, J. D., Coltart, T. M. & Campbell, S. (1974) Maternal serum-alpha-fetoprotein levels and prenatal diagnosis of neural-tube defects. *Lancet* **i**, 428–429.

Stewart, C. R., Milford-Ward, A. & Lorber, J. (1975) Amniotic fluid α_1 fetoprotein in the diagnosis of neural tract malformations. In press.

Wald, N. J., Brock, D. J. H. & Bonnar, J. (1974) Prenatal diagnosis of spina bifida and anencephaly by maternal serum-alpha-fetoprotein measurement. *Lancet* **i**, 765–767.

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