SPINA BIFIDA
A DESCRIPTIVE RESEARCH STUDY

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FORMULATING THE PROBLEM

In society one is continually being made aware of the needs and demands of the physically or mentally handicapped child. This awareness is stimulated by the media, medical journals as well as the general public’s own increased interest in the children’s plight.

Whilst it would appear that more children are being born with some sort of disease or handicap now than a few decades ago, this can possibly be explained by the recent rapid increase in population. This raises the potential number of children likely to be born with or without a defect. Also, with the increased survival rate and lifespan as a result of advanced technology, it would appear that the actual number of afflicted children who are alive is higher in the twentieth century than it was before.

This is also true in the case of the incidence of spina bifida. Whilst the overall incidence in the general population is 1:1000, statistically it would appear that there are more live children with spina bifida now than a few years ago. However, to the person who is affected, or to his or her family, the handicaps are particularly severe. A normal, exciting and active childhood, where exploration of one’s environment is a daily adventure, is often physically impossible and the likelihood of a stimulating and successful school career is limited. There are thus fewer opportunities and abilities for the afflicted child to lead a productive and fulfilling life.

It was thus because of a stimulated awareness of the needs of children with Spina Bifida, having nursed and spoken to patients and their families with this defect, because of its relatively high incidence compared to other abnormalities, and a general desire to learn more about the condition itself, that the author, as a first year student nurse, undertook this descriptive research essay.

The purpose of the study was to describe and discuss spina bifida, the effects on the individual concerned — both physically and mentally, and to indicate methods of treatment, important aspects of prevention as well as the care which is available at present. The required information was gained by means of research, clinical interviews, and observations made at The Red Cross Spina Bifida Clinic.

DEFINITION

The spinal cord is normally well protected by its immediate coverings — the outer dural tube, the middle arachnoid mater and the inner pia mater which invests it closely. It is suspended by the dentate ligaments in the vertebral canal and cushioned by cerebrospinal fluid in the sub-arachnoid space. The secure bony canal is covered by muscle, facia and skin.

Spina bifida is a developmental defect of the spinal column, the fundamental defect occurring between the 21st and 28th day of embryonic life, during the early development of the neural tube. According to M.E. Marshall (1981, p. 30), it is manifested by a failure of fusion between the arches of the vertebras, with or without protrusion and dysplasia of the spinal cord or nerve roots and its meninges beyond the normal limits of the spinal canal.

HISTORICAL BACKGROUND

Spina bifida has been recognized in skeletons found in North-Eastern Morocco and estimated to have an age of almost 12 000 years. It was known to the ancient Greek and Arabian physicians who thought that the bony defect was due to a tumour. The term spina bifida was first used by Professor Nicolai Tulp of Amsterdam in 1652. Many other terms have been used to describe this defect, but spina bifida remains the most useful general term, as it describes the separation of the vertebral elements at the midline.

CLASSIFICATION

Spina bifida cannot be described adequately without further classification. There are two main varieties which are spina bifida occulta and spina bifida cystica.
FIG 1: Summary of classification of Spina Bifida

NORMAL VERTEBRAL AND SPINAL CORD DEVELOPMENT

DEFECTIVE CLOSURE OF NEURAL TUBE

Anterior defect (cephalic end) ↓
Incomplete brain development ↓
Anencephaly

Posterior defect (caudal end) ↓
SPINA BIFIDA

SPINA BIFIDA CYSTICA

SPINA BIFIDA OCCULTA

(incomplete vertebral development but normally formed spinal cord)

Myelomeningocele

Meningocele

MYELOMENINGOCELE

MENINGOCELE

Spina bifida occulta

Here there is failure of one or more vertebral lamina to fuse at the midline, but there is no cystic distention of the meninges. There may or may not be changes in the overlying skin, neurological signs or pathological changes in the spinal cord. However, in the majority of patients the split spine is indeed occult or hidden, but there are a few cases who have some external skin mark covering the lesion, in the form of a hairy patch or dimple.

The defect usually occurs in the lumbo-sacral region. Sometimes the gap may be palpable or it may be so narrow as only to be evident on radiological examination.

Spina bifida cystica

This is the term used to describe the cystic distension or protrusion of the spinal cord and/or its meninges through a congenital defect in the posterior neural arches, and it is therefore the more severe of the two varieties.

According to M.E. Marshall (1981, p. 31), there are two distinct and differing lesions, that is Myelomeningocele and Meningocele. MYELOMENINGOCELE is the most serious type of spina bifida which results in paralysis of the nerves from the point of the lesion down. There is a wide defect in the posterior neural arches over several vertebral segments, the normal spines are absent and the everted pedicles and laminae are found well away from the midline. Through this bony defect there is a protrusion of meninges and neural tissue which is situated outside the vertebral canal (see figure 2).

In the open myelomeningocele the defective spinal cord is situated on the surface of the sac, taking the form of a flat neural plate. The nerve roots stretch ventrally to the neural plate. Peripherally the lesion is joined by a skin which is frequently thin and vascular and may leak cerebrospinal fluid. At the margin of the lesion the dura fuses with the edge of the skin defect. This type of myelomeningocele is thus open as the neural tissue is uncovered and the neural tube is not closed.
FIG 3: DIAGRAMATICAL REPRESENTATION OF EMBRYOGENESIS IN THE 4TH WEEK OF PREGNANCY (Day 19-28).

Cross Section of Dorsal surface of human embryo:

Days

19
- Embryonic ectoderm
- paravertebral muscles
- somite
- intermediate mesoderm
- dorsal aortas

20
- neural groove
- neuro-ectodermal junction
- somite (2nd)
- dorsal aortas

21
- neural fold
- paravertebral muscles
- 2nd somite

22
- embryonic ectoderm
- neural tube
- somite

23
- embryonic ectoderm
- neural tube
- somite
- intraembryonic coelam

24
- embryonic ectoderm
- neural tube
- somite

25
- embryonic ectoderm
- neural tube
- somite

26
- Posterior root of ganglion
- neural tube
- myotome
- dermatome

Coronal Section of human embryo:

- neural plate
- cut edge of amnion
- 2nd somite

- closed neural tube
- posterior neuropore
- neural groove

- closed neural tube

Lateral section of human embryo
In the closed myelomeningocele, although the defective spinal cord and/or nerve roots are situated outside the vertebral canal, the neural tube is closed as the cord is situated within the wall of the sac or lesion which is covered with a combination of skin and membrane.

**Meningocele** In the simple meningocele, the unfused condition of vertebral arches is confined to a few vertebral segments with cystic distension of the meninges. The protruding meningeal sac has a narrow neck and usually contains cerebrospinal fluid. There is thus usually an absence of myelodysplasia of the spinal cord which developed normally. The meningocele is covered by a more or less intact skin.

**EMBRYOGENESIS OF THE SPINAL CORD, ITS COVERINGS AND VERTEBRAL BODIES**

**Normal embryogenesis** (see figure 3)  
There are three distinct and successive phases in normal spinal cord development.

**Closure of the neural tube**

On the nineteenth day after conception when the embryo is only 1.4 mm long, the skin over the middle of the back becomes thickened to form a neural plate which is the of the back becomes thickened to 

**Development of the coverings of the spinal cord**

Appearance of the primordial spinal cord is followed by a development of its coverings in a cranial-caudal direction. The pia mater develops from about forty days following by the duramater. The arachnoid-mater later separates from the duramater.

By the time closure of the neural tube and canalisation of the caudal cell mass are complete at 45 — 50 days gestation, the vertebral bodies are well advanced.

**EMBRYOGENESIS OF SPINA BIFIDA**

It is generally agreed that the lesion begins with failure of the embryonic neural plate to close in its caudal part, accompanied by an over-growth of neural tissue in the area of the defect.

By the 30th day of development no bones have as yet been formed and when they do form, plates of bone fail to develop over the area of the neural tube defect.

Although the complete length of the neural tube is vulnerable to a large variety of developmental abnormalities, they occur more commonly at the lower and upper ends of the neural tube. Spina bifida commonly occurs in the lower parts of the neural tube (lumbosacral region), although no portion is exempt from the defect.

Abnormalities in the upper end of the tube, which involve the brain, frequently result in the massive brain degeneration, anencephaly. In this anomaly, the infant is either stillborn or dies shortly after birth.

**INCIDENCE, AETEOLOGY AND RECURRENCE RISKS**

**Incidence in the Cape Peninsula**

A survey was done into the incidence of neural tube defects (any defect in the morphogenesis of the neural tube) amongst the different ethnic groups in selected Cape Town hospitals in the period 1975 to 1980 (S.A. Medical Journal, July 1983 pp 83 — 84). The following results were found.

During the period between 1975 — 1980, 105 infants out of a total of 116 859 delivered at the selected hospitals in the Cape Town area had a neural tube defect. There were 58 cases of spina bifida and 47 of anencephaly. The combined incidence of spina bifida and anencephaly in Whites was approximately 1 : 300 births which is similar to that generally encountered in the United Kingdom. However, the incidence in other ethnic groups was considerably lower at 1 : 1 250 births for the Coloured (mixed ancestry) and 1 : 2 000 for Black groups (see table 1).
The total number of births and the incidence of spina bifida (1/1000 births) in all population groups in the Cape Peninsula during 1975—1980 are shown in table 2 below.

To conclude therefore, there is a fairly consistent overall incidence of neural tube defects of 1:1100 births, but there is considerable variation geographically and ethnically. Spina bifida is more common than anencephaly in both the White and Coloured groups. However, in the Black group anencephaly is 50% more common than spina bifida. It is speculated that this might be a result of local factors as it is contrary to findings reported in Natal.

Incidence in other parts of the world

Various surveys (Elwood, 1973; Edwards, 1958) have shown that amongst the Celtic people (Irish, Welsh, West Scots) the incidence is 2 to 3 times higher than the average world incidence which is 1:1000. There is also a wide variation in incidence by maternal age (increasing with age); parity (higher in first births); fetal sex (there is a slight preponderance of females over males of about 1.3 to 1.0 in most cases). Even seasons seem to affect incidence — higher in spring conceptions. Spina bifida occurs more frequently in times of economic depression and in many areas, including Scotland, a significant difference in social class incidence has been noted. The highest rates occurring in the lower socio-economic classes (Edwards, 1958).

Aetiology and recurrence risks

Spina bifida is generally considered to be caused by an unknown environmental agent interacting with genetic factors.

It is not hereditary in the sense that it is transmitted in one of the definite ratios which were discovered by Gregor Mendel and referred to as Mendelian inheritance. Nevertheless, there is a genetic influence which is expressed by an increased risk of having a second child with spina bifida after having a first child with this defect.

Studies have shown (Smithers et al 1968; Laurence 1969) that once a woman has produced an affected infant, the risk of recurrence of the condition in each subsequent pregnancy is 1:20. If a second child with the disorder is born the risk is increased to 1:10 at the next pregnancy. However, 25% of affected infants are born to primiparous mothers.

The environmental factors were discussed briefly earlier, where it was seen that the incidence seems to vary greatly according to the different racial groups. However, striking regional variations within a small country seem to point rather to an extrinsic factor. For example, the incidence of spina bifida in the mining valleys of South Wales is almost twice that in the Coastal Plain around Cardiff.

Nevertheless, there is a genetic influence which is expressed by an increased risk of having a second child with spina bifida after having a first child with this defect. As the defect rarely reaches the level of the upper back, the muscle weakness is nearly always confined to the lower limbs and lower back. However, there is often some impairment of the dexterity of the upper limbs, but with exercise they usually become powerful and often enable the child to become physically independent in activities of daily living.

Weakness of the muscles in the wall of the bladder and bowel

The bladder and bowel innervation problems usually result in urinary and bowel incontinence. These difficulties arise from either a weakness of the muscles in the walls of these organs which normally contract to expel their contents or a weakness, tightness or failure of relaxation of the sphincters muscles which are normally tightly closed around the urethra and anus, and relaxed during emptying of the bladder and bowel. Problems are related to the functional inter-relationships of the detrusor (emptying muscles) and sphincters (retaining circular muscles).

The structural and functional defects associated with myelomeningocele include the following.

Hydrocephalus

Hydrocephalus is a complication occurring in approximately 95% of children with myelomeningocele. Hydrocephalus may cause a rapid enlargement of the head resulting from the abnormal accumulation of cerebro-spinal fluid within the ventricles of the brain. Checking the head circumference is thus vitally important in all cases of myelomeningocele.

Hydrocephalus it is a potentially life threatening problem which requires the prompt attention of a paediatric neurosurgeon. Serial checking will indicate whether the hydrocephalus is arrested or progressive. If it is arrested, no surgery is required but if it is progressive operative intervention is necessary.

Paralysis of the lower limbs

There may be moderate degrees of muscle weakness or bowel incontinence associated with a meningocoele. However, these symptoms are far more severe in the myelomeningocele variety and associated with it are a number of fundamental structural and functional defects which will be discussed later.

Loss of skin sensation

There is a loss of awareness of touch, pain, pressure and heat or cold in those areas of the skin normally innervated by nerves which are involved in the spinal cord defect. Skin which is not sensitive blisters easily and pressure from tight clothing or braces can break the skin down into a weeping sore or pressure ulceration.

Muscle weakness

Although there is some variability of weakness in specific muscles, the extent of the muscle weakness is related to the level of the spinal cord defect. As the defect rarely reaches the level of the upper back, the muscle weakness is nearly always confined to the lower limbs and lower back. However, there is often some impairment of the dexterity of the upper limbs, but with exercise they usually become powerful and often enable the child to become physically independent in activities of daily living.
Secondary paralysis is due to the onset or extension of paralysis after birth due to traction pressure or other effects on the cord. According to Marshall (March 1981, p 34) deformities as a result of spina bifida can be divided into three main types.

Firstly, congenital deformities occurring prenatally in the process of embryogenesis due to the spinal lesion.

Secondly, primary deformities caused by intra-uterine paralysis of the muscles resulting in imbalance and consequent deformities. Thirdly, secondary deformities which develop after birth and are due to an imbalance of muscle power.

**Spina Bifida Occulta**

The most important early evidence of spina bifida occulta is the cavovarus action of one foot without fixed deformity. It must be distinguished from the same foot action occurring for non-neurological reasons, the difference being that in the latter the foot action is transient and disappears very quickly with barefoot walking. In spina bifida occulta, the foot action persists although it can be consciously controlled in the early stages before deformity occurs. As the foot deformity becomes established, the shoe may become distorted accordingly.

Overlying the lesion the skin is generally normal, but external cutaneous manifestations have been found covering the lesion in the midline of the back, from the mid-thoracic region to the end of the sacrum. Table 3 shows the various types of manifestations which may occur as well as those which predominated in a study of 200 cases of spina bifida occulta.

The other symptoms which may be present are similar to those of spina bifida cystica with meningocele, but are far less severe.

**TREATMENT**

The actual sac-like lesion may or may not be surgically closed at birth depending on the extent and type of the lesion.

Active surgical treatment of the child with myelomeningocele was of little avail until the development of the Spitz-Holter valve and ventriculo-caval shunt because of the crippling complication of hydrocephalus. As mentioned earlier, if the hydrocephalus is progressive, operative intervention is necessary. The operation is in effect a bypass or shunt, the aim being to bypass the obstruction and allow for the reabsorption of cerebro-spinal fluid into the blood stream. A thin tube is inserted into the dilated ventricle of the brain and is then attached to a pump which in turn is attached to another catheter which may be placed into the vena cava (ventriculo-caval shunt) or peritoneal cavity (ventriculo-peritoneal shunt).

<table>
<thead>
<tr>
<th>Table 3 Cutaneous manifestations found in 200 cases of spina bifida occulta</th>
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<tbody>
<tr>
<td>Lumbosacral lipoma (30%)</td>
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<tr>
<td>None (27%)</td>
</tr>
<tr>
<td>Hypertrichosis (19%)</td>
</tr>
<tr>
<td>Sinus or dimple (10%)</td>
</tr>
<tr>
<td>Scarred area (7%)</td>
</tr>
<tr>
<td>Naevus (4%)</td>
</tr>
<tr>
<td>Atrophied fatty meningocele (3%)</td>
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**Fig 4: Ventriculo-caval and ventriculo-peritoneal shunts**

In the **ventriculo-caval shunt** (A) fluid is redirected into the superior vena cava by means of a flexible tube. In the **ventriculo-peritoneal shunt** (B) fluid is redirected into the peritoneal cavity. This extra length allows for the child’s growth.
Often the shunt becomes blocked by a blood clot or common brain damage. Should a hydrocephalic child be detected early and provided with a shunt, there should be no brain damage.

The treatment of paralysis of the lower limbs is aimed at correcting and preventing the deformities associated with paralysis, maintaining the correction obtained and improving the function of the part involved. Most orthopaedic operations are carried out between 6 months and 3 years of age and in general surgical correction takes place in a proximal, distal direction — hips first and feet last.

The bladder incontinence is treated according to the type of bladder dysfunction of which there are two main types: the inert bladder and the reflex bladder.

In the inert bladder there is no evidence of sensation and small volume dribbling occurs after a variable degree of filling. Manual expression of urine is usually possible, carried out by the mother and later by the child himself.

With the reflex bladder, a certain amount of filling occurs. Intermittent reflexive contractions result in voiding of urine in a stream or may be almost continuous with constant dribbling. Should the external sphincter be very tense, stagnation of urine will occur in the bladder resulting in the constant threat of infection. As soon as possible after birth, a child is placed on intermittent catheterisation primarily to reduce the danger of upper urinary tract infection.

The main aim of treatment of bladder dysfunction is to prevent renal deterioration which could result in renal failure and premature death. From approximately 3 — 9 years the aim is to gain continence by conservative means. The majority of children continue with intermittent catheterisation together with drug manipulation to increase the bladder capacity.

Other alternatives are penile appliances for males, continuous catheter drainage for females or, as a last resort, urinary diversion or ileal conduit.

The neurogenic bowel is slightly easier to treat than the neurogenic bladder. The child is encouraged to develop a bowel regimen and constipation, which is inevitable as there are no effective contractions in the descending colon and rectum, is prevented by the administration of drugs.

From the above it is evident that no single doctor can cope adequately with all the clinical problems which may arise in the fields of paediatrics, neurology, neurosurgery, urology, orthopaedics, plastic surgery and ophthalmology. In addition, other serious problems require nursing, social work, physiotherapy and psychology. Thus, for treatment to be successful all these disciplines need to be co-ordinated. This has resulted in the recent trend to set up spina bifida clinics in paediatric and general hospitals so that optimum treatment can be initiated with minimal delay. An example of one such clinic is the Red Cross Memorial Children’s Hospital Spina Bifida Clinic.

The optimal management of children with spina bifida also makes heavy demands on medical, surgical, social and educational resources, not to mention financial resources, all of which are usually limited.

Therefore selective treatment seems to be the only possible solution at present. Reports and surveys are continually in progress and the quality of life, both physically and intellectually, of survivors has improved markedly. However, there is always the possibility of survival of children who were not treated and might be more handicapped as a result. Thus, although selective treatment is an unhappy compromise, it is not a final solution but rather at present the only acceptable approach where resources are limited. In the future when prevention is a reality, selection for treatment will become irrelevant.

PREVENTION

It is important that patients who are at risk for this condition be referred to their local clinical genetic department for counselling.

The other very important aspect of prevention is the method of prenatal detection. The technique involves determination of minute quantities of alfa-feto-protein which is formed in the liver of all persons, including the liver of the developing fetus.

It has recently been shown that the level of alfa-feto-protein in the amniotic fluid is raised if the fetus has either anencephaly or an open spina bifida. The biochemical estimation of the substance is now used as an antenatal diagnostic test.

Amniocentesis is the procedure used to describe the penetration of the amniotic sac with a needle and the withdrawal of a small quantity of amniotic fluid for alfa-feto-protein examination. It is therefore available for all mothers with a previous affected child or a family history of a neural tube defect. Most cases of spina bifida are sporadic and as amniocentesis is highly technical and expensive, it would be impossible to perform this test on all pregnant women as a means of screening for raised alfa-feto-protein levels.

However, it has recently been demonstrated that the alfa-feto-protein level is raised in the maternal serum as well as in the amniotic fluid of a woman carrying a fetus with spina bifida. Therefore, it is now possible for the maternal serum of a pregnant woman to be screened routinely for raised alfa-feto-protein levels. This test has an 80% accuracy. If a raised serum alfa-feto-protein is found an ultrasound and amniocentesis will subsequently be done. These methods can also miss an affected fetus and as with amniocentesis, there is about a 1% risk that the mother may abort.

Thus, although steady progress has been made in refining techniques and improving the accuracy of a method of prenatal detection, antenatal screening is still not 100% effective.
CONCLUSION

The two first months in the life of an infant born with a severe handicap, such as spina bifida, are going to be a time of great stress and crisis for the family. Not only will the child have to spend considerable time in hospital, with all its attendant problems, but the handicaps associated with the deformity are severe.

The discrepancy between the idealised perfect child and the handicapped child will affect the parents. The mother and family will therefore require much help and support to enable them to work through their grief and come to terms with the fact that their child is going to have many problems. This has led to the establishment of Spina Bifida Clinics which play a vital role in helping the child to overcome his problems as well as in assisting with the emotional needs of the parents, encouraging them to have positive inter-actions with their child and overcoming the negative feelings they might have towards the child.

Furthermore, the socio-economic hardships associated with frequent hospitalisation are great, not to mention the financial burden these children place on the economy of the state, the resources of which are usually limited.

Limitations of the project

Being a first year student nurse, the author's knowledge on the subject of spina bifida was limited and she therefore had to do a considerable amount of research.

The time involved was a limiting factor regarding the detail of individual case studies.

No monetary costs were involved as no questionnaire was drawn up. The author did not have the experience or knowhow regarding the formulation of such a questionnaire and it was also found to be unnecessary in this project as the aim was at producing a purely descriptive study on the condition of spina bifida.

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Die toegewyde en roepingsbe­ wuste verpleegkundige het die in­ dersaad die geleiendheid om deel te hê in die geestelijke groei van die ly­ dende mens. Hiërdaardie verpleegkundig­ genee ervaar arbeidsvreugde, 'n siepe tevredenheid en roepingsver­ vulling, by die aanskou van die vrugte op pyn en lyding in haar me­ demens, van haar moeitevolle arbeid. Op dié wyse verkry sy 'n sterk bevestiging van die diepste be­ tekenis van lyding in die mens se lewe, en vind ook die essensië van haar hulpverlening. In dié sin is hulp aan die lydende mens voor­ waar 'n werk van geloof — van ewige waarde.