

# spina bifida

## Introduction

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### Spina bifida: key primary care issues

*\*Spina bifida is a static congenital neural tube disorder resulting in continuing evolving disease involving multiple organ systems. Those affected require lifelong surveillance with coordinated management involving the patient, parents, general practitioners, neurologists, physiotherapists, occupational therapists, social workers, nurses, neurosurgeons, urologists, orthopaedic surgeons, physicians and other health professionals.*

*\*The term spina bifida is used throughout this manual to refer to all myelodysplasias unless specifically stated. Similarly, issues specific to hydrocephalus alone will be highlighted, although many of the issues relating to spina bifida are relevant.*

#### Key issues for clinicians

- Spina bifida is one of the most severe congenital abnormalities compatible with a full and active life.
- Active surveillance to diagnose problems early is more likely to keep a person with spina bifida active and independent.
- Routine surveillance must involve regular review of the neurological, urological and musculoskeletal systems.
- Changes in neurological signs can indicate severe, treatable underlying pathology. Clinicians need to have a record of past neurological signs for a clear comparison in any ongoing neurological review.
- All new neurological signs require immediate referral to a specialist centre.
- Urological complications are a major cause of morbidity and mortality in spina bifida.
- Urological assessment is an integral part of ongoing management of spina bifida at all ages.
- Progressive loss of mobility profoundly decreases the quality of life for people with spina bifida. Attending to orthopaedic issues and encouraging appropriate physical activity helps to minimise the impact of progressive loss of mobility.
- Incontinence management is not just an aesthetic issue — it is a key barrier to achieving independence for young people and adults with spina bifida.
- Dealing with sexuality and reproductive issues is a routine part of managing spina bifida.
- Paediatric centres can help GPs locate adult spina bifida treatment centres. In the absence of adult centres, paediatric centres routinely offer support for adults with spina bifida.
- Cognitive dysfunction often adversely affects perception of symptoms and the ability of the patient to follow medical management instructions.

Spina bifida — a severe congenital condition compatible with independent life

Spina bifida is a term used to describe a group of multiple complex congenital anomalies and abnormalities affecting development of the neural tube and related structures during the third and fourth week of pregnancy (see *Chapter 1 The spinal cord and brain in myelodysplasias* for a more detailed overview of spina bifida and neural tube defects).

Spina bifida has been described as one of the most serious congenital conditions compatible with full life. Prior to antibiotic and surgical advances of the last half century, life expectancy was often less than a year. Improvements in the management of children, young people and adults with spina bifida, related myelodysplasias and hydrocephalus has meant that many are now not only surviving into adulthood, but live active and happy lives, which may involve working and having children.

### Independent living for young people and adults — overcoming barriers

As more children with spina bifida reach adulthood, the special needs required to achieve and maintain independent living in the community are becoming more obvious and pressing.

Generalist clinicians who take a proactive role in detecting and overcoming the multiple problems facing young people and adults with spina bifida maximise their patients' chances of achieving physical and social independence. The clinician achieves this by becoming familiar with the physical, psychological, sexual and social barriers that impact daily upon the lives of those with spina bifida, as well as their affected partners, families and friends.

### Assess now — don't wait until problems occur

Because the medical consequences of spina bifida are continually evolving, regular follow up is essential to prevent major problems before they occur.

Many people with spina bifida may not have attended a specialist centre for some time, and should be encouraged to attend for a routine assessment. Familiarity with the primary care management of young people and adults with spina bifida enables treating doctors to ensure that regular, thorough and appropriate health maintenance occurs.

### The role of GPs and generalist health care providers

General practitioners can take a central role in the daily management of the young person or adult with spina

bifida. As well as day to day treatment issues, the GP plays a major role in the early detection and referral of spina bifida related complications to treatment centres.

Paediatric centres can help general practitioners locate adult spina bifida treatment centres (see *Chapter 9*).

### Locating referral clinics for young people and adults with spina bifida

Specialist clinics managing adults with spina bifida are a relatively recent development due to the increasing survival of children with spina bifida.

Children with spina bifida are managed in paediatric centres until adulthood, but upon reaching maturity may not establish contact with specialist treatment centres.

Paediatric centres can identify the location of adult spina bifida treatment centres, but many paediatric centres will treat adults with spina bifida if no other services are available.

### Critical areas of primary care — physical and psychosocial

The management of young people and adults with spina bifida and related disabilities depends upon many factors, but is largely determined by the level of the spina bifida lesion and the presence of hydrocephalus, related central nervous system problems and urological dysfunction. The nature and extent of complications can profoundly affect many body systems.

There are many psychosocial implications of spina bifida including personal, psychological, social and sexual issues.

### Monitoring of physical conditions<sup>1,2</sup>

While spina bifida is a multisystem condition, there are three main areas of functioning that require special ongoing surveillance:

- the neurological system
- the urological system
- the musculoskeletal system.

### Neurological system

Ongoing assessment of the neurological system is mandatory. Any changes in neurological signs require immediate referral to a specialist centre.

While this may appear to be obvious, many individuals with spina bifida lose contact with specialist services on reaching adolescence and adulthood and may accept

changing neurological function as an inevitable part of spina bifida. Always examine the nervous system as part of the general patient review and look for changes in neurological signs.

Changes in neurological signs can indicate severe, treatable, underlying pathology. The clinician's task is easier when there is a record of past neurological signs for a clear comparison in ongoing neurological review. All new neurological signs require immediate referral to a specialist centre.

Changes in lower limb neurological signs or continence can indicate the presence of a serious condition called tethered cord which can occur at any time of life (see *Chapter 3 Spinal cord tethering*). Clinicians managing patients with spina bifida need to be familiar with this condition.

Clinicians managing individuals with spina bifida need to be familiar with the tethered spinal cord syndrome.

The manifestations of a blocked ventriculoperitoneal shunt extends beyond headache and includes visual symptoms, confusion, somnolence, behavioural disturbances and incontinence.

Clinicians managing patients with spina bifida need to be familiar with the symptoms and signs of a blocked ventriculoperitoneal shunt.

### Urological system

Although neural tube defects are neurological disorders, their impact on the urological system is responsible for a significant proportion of life threatening complications. As a consequence, an understanding of the detection and screening for urological abnormalities is central to an understanding of the management of spina bifida and related disorders.

In line with the urological management of spina bifida, attention to incontinence issues is central to the quality of life for all those with spina bifida.

Urological complications are a major cause of morbidity and mortality in those with spina bifida.

Due to neurological involvement of the bladder, bladder control is commonly impaired. Not only does this greatly impact upon quality of life, but lack of bladder control can lead to major kidney damage through reflux and recurrent infection.

Urological complications are responsible for a large part of morbidity and mortality from spina bifida. These

issues are fully covered in *Chapter 4 Urological management of spina bifida*.

Urological assessment is an integral part of ongoing management of spina bifida at all ages.

### Musculoskeletal system<sup>3</sup>

Normal nerve development and function do not occur at or below the spinal cord level of the spina bifida lesion. Therefore, the higher the lesion, the higher the level of the paralysis. However, the pattern of muscle weakness will vary with each individual, depending upon the site and extent of the spinal defect.

Muscle groups on opposite sides of the body may not be equally affected, leading to orthopaedic abnormalities, ranging from scoliosis and dislocations to leg and foot problems. Lack of weight bearing can also result in an increased risk of fractures and progressive deformity.

Ongoing orthopaedic assessment and review helps to maximise function and minimise the impact of the disability.

Progressive loss of mobility profoundly decreases the quality of life of people with spina bifida. Attention to orthopaedic issues and encouraging appropriate physical activity helps to minimise the impact of this progressive loss.

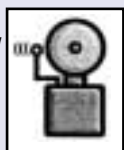
### Alarm bells for clinicians — some conditions require urgent referral

Clinicians also need to be aware of a number of important conditions that require more urgent referral to specialist centres. These conditions include:

- spinal cord tethering (back pain and loss of lower limb, sexual and/or sphincter function (see *Chapter 3*)
- headaches or cognitive impairment, especially as a sign of suspected ventriculoperitoneal shunt problems (see *Chapter 2*)
- any change in neurological function or signs, regardless of how slow or minor
- recurrent urinary tract infections (see *Chapter 4*)
- renal impairment (see *Chapter 4*)
- renal calculi.

These conditions are more fully explained in each section as indicated, and clinicians involved in the care of people with spina bifida should familiarise themselves with these possible serious complications.

When you see the 'alarm bell' icon, this means the point is important and may require urgent referral to a specialist centre.



## Dealing with sexuality and reproductive issues is a routine part of managing spina bifida

Concern about sexuality and reproductive issues may be symbolic of the increasing need for independence in a person who has been, and often still is, highly dependent upon others in their daily living needs.

### Incontinence — a major barrier to independence

A major barrier to total independence is urinary and faecal incontinence.

Effective management of incontinence enables access to education, work and financial independence, socialisation and fulfilling relationships.

As well as preventing independence, poor incontinence management contributes to poor self esteem, which can discourage people from being active and social. Commonly reported feelings illustrate the personal impact of incontinence on self esteem: worry; dismay; embarrassment; guilt; fear; isolation; sadness; and frustration.

### Monitoring psychosocial impact of spina bifida

Spina bifida can have a profound impact on body image and self esteem, and this can adversely affect the ability to form healthy relationships with family, friends, and partners. In addition, the common problem of incontinence can cause a further loss of self esteem.

This loss of self esteem not only interferes with a person's independence, but can erode their ability to participate fully in education, employment and other aspects of daily life that come with living a modern, active life.

A supportive primary care clinician aware of these issues can help a patient overcome these issues and gain full participation in society.

Spina bifida affects the spinal cord and inevitably affects sexual functioning. Dealing with these issues is a routine part of the daily management of spina bifida.

As for those without spina bifida, sexuality and reproductive issues become increasingly important for young people as they reach adolescence and adulthood. However, children with spina bifida have often grown up being especially dependent upon their parents and the emergence of sexuality issues can be a source of conflict for the person, their family and carers. This is dealt with in *Chapter 8 Sexuality and reproductive issues*.

### References

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