

# Overview of spina bifida and the nervous system

## Chapter 1: The spinal cord and brain in myelodysplasias

*This chapter defines various neurological abnormalities and terms specific to the management of spina bifida and related disorders.*

### Key issues for clinicians

- While spina bifida is a congenital condition, complications may occur in young people or adults.
- Neural tube defects can affect the entire length of the central nervous system. This includes the brain as well as the spinal cord.
- Hydrocephalus affects most people with spina bifida.
- All those with spina bifida and their relatives require genetic counselling prior to conception.
- High dose folate supplementation is given to all high risk cases.

Spina bifida is a complex birth defect that has been recognised for thousands of years. Until the middle of the last century, most babies born with spina bifida did not survive more than a year, but due to medical advances — especially in neurosurgery and urology — most infants will survive into adulthood.

### Definitions

Spina bifida is the collective term used to describe a group of multiple, complex congenital abnormalities and anomalies of the neural tube known as myelodysplasias.<sup>1,2</sup> As spina bifida affects the entire length of the neural tube, additional central nervous system abnormalities are very common, especially hydrocephalus and Arnold–Chiari malformations.

While spina bifida is a congenital condition, complications may occur in later life. The underlying pathology provides an understanding for the ongoing clinical management of those affected.



The commonest types of myelodysplasias are:

- spina bifida occulta
- meningocele
- myelomeningocele
- lipomatous malformations of the spinal cord and central nervous system
- other related conditions: sacral agenesis, Vater syndrome.

Spina bifida occurs when the spinal column does not form correctly at some point along its length. In reality, spina bifida refers to a complex of conditions. Of the 260 000 children born in Australia each year, 400 are affected by this condition.<sup>3</sup>

Spina bifida occulta refers to incomplete development of the vertebrae, but the spinal cord is intact with no obvious skin defects. Spina bifida occulta occurs in 5% of live births. The overlying skin may be normal, but may also be

associated with a dimple, hair patch or red discolouration — the so called herald or signature mark (*Figure 1*).<sup>4</sup> Signature marks may be associated with significant spinal abnormalities. Abnormalities in function can emerge at any time later in life.

A meningocele is a cystic lesion filled with cerebrospinal fluid (CSF) where the meninges protrude into an external sac, usually located in the lumbosacral region, due to failed closure of the vertebral arches. The amount of skin covering the lesion varies. However, there is no nerve involvement. The lesion can be associated with hydrocephalus and central nervous system abnormalities.

A myelomeningocele is one of the most important and severe types of spina bifida that occurs in about one in 1000 live births (*Figure 2*). It occurs when part of the spinal column is undeveloped, with incomplete formation of the overlying vertebrae and no overlying skin. Most frequently located in the lumbosacral region, myelomeningoceles are generally cystic and contain CSF that drains when the thin sac is disrupted. Both the meninges and spinal cord protrude into the sac, and the spinal cord is often abnormal. The level and severity of the lesion affects malformations and patterns of functional loss. Myelomeningoceles are often associated with Arnold–Chiari type II malformations. Approximately 80% of children with this lesion develop hydrocephalus.

### Other lesions

Diastematomyelia describes how the spinal cord is split into two hemicords with each having a set of dorsal and ventral nerve roots, each contained in a dural sheath. This belongs to the group of lesions called split cord malformations. Vertebral body abnormalities are often present, leading to scoliosis.

Syringohydromyelia occurs when a syrinx (a cystic cavity) forms within the spinal cord and may be found in the medulla or located anywhere from the cervical to the lumbar areas. Syringohydromyelia is associated with neural tube defects. The cystic cavity may or may not communicate with CSF flow pathways. The syrinx may progressively enlarge until pressure on the nervous tissue leads to serious, variable complications.

Lipomatous malformations, commonly called lipomeningocele, occur when excessive lipomatous tissue is within or attached to the spinal cord or filum terminale. This group of malformations are by far the most common form of closed spinal neural tube defects and vary from an enlarged filum terminale containing adipose tissue to a huge fatty mass occupying much of the dorsal lumbosacral region

which contains the spinal cord and CSF. This group of lesions includes the lipomeningocele, lipomyelocele, leptomyelolipoma, lumbosacral lipoma and lipoma of the filum terminale. They represent a continuum of embryologic maldevelopments with similar clinical findings and prognosis to the open neural tube defects.

### Spina bifida and the brain

As neural tube abnormalities affect the entire length of the spine and central nervous system, most individuals with myelodysplasia will have associated brain abnormalities. This often results in hydrocephalus.

The Arnold–Chiari malformation (type II) is the commonest brain abnormality in spina bifida (*Figure 3*). Affecting almost all people with spina bifida, this is an anatomical defect of the lower brain and cerebellar structures. This causes herniation of the cerebellar tonsils through the foramen magnum and also causes the medulla to kink and move downwards into the



Figure 1. Herald mark. Hairy patch over repaired lumbosacral spina defect.

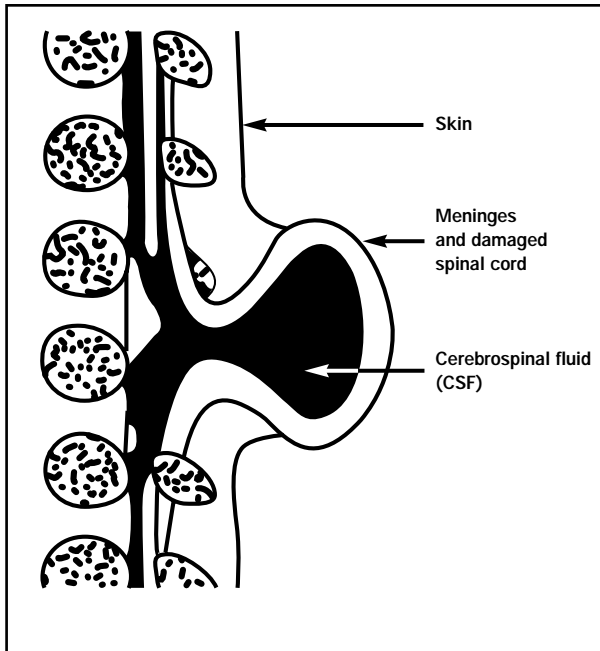


Figure 2. Myelomeningocele. Myelomeningoceles are the commonest cause of serious spina bifida related disability. The extent of the disability largely depends on the level of the lesion.

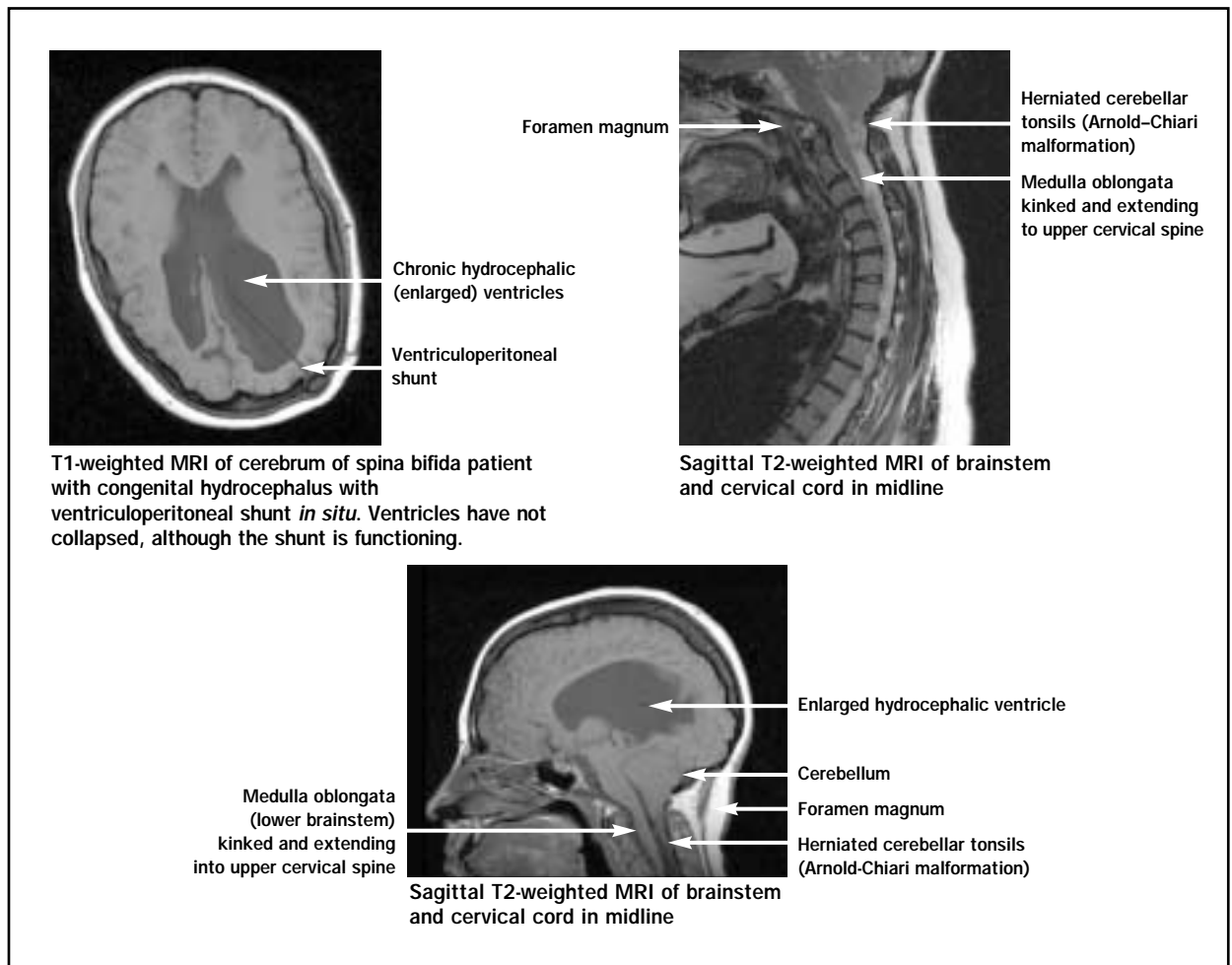


Figure 3. Hydrocephalus and Arnold-Chiari malformation. (Courtesy of Department of Diagnostic Imaging, Monash Medical Centre, Victoria.)

cervical spinal canal, also displacing cranial nerves. Around a quarter of patients with Arnold-Chiari malformations develop brainstem dysfunction, with symptoms often appearing in the first months of life.

Other central nervous system malformations can occur, including encephaloceles, syrinxes above the level of the lesion, callosal agenesis and other brain stem abnormalities.

Symptomatic decompensation secondary to anatomical abnormalities can also occur in adults.

### Hydrocephalus

Hydrocephalus is found in most infants during prenatal ultrasonography. Most individuals with spina bifida and hydrocephalus require a ventriculoperitoneal shunt to relieve intracranial pressure.

If symptomatic hydrocephalus is present at birth, back surgery and shunt insertion may be performed at the same time.

Clinicians need to be aware of the signs and symptoms of hydrocephalus and the related signs and symptoms of ventriculoperitoneal shunt dysfunction (see *Chapter 2* for further information).



### Skin abnormalities may herald serious signs

Skin abnormalities can be associated with neural tube defects. As the skin and nervous system share a common ectodermal origin, this may explain the simultaneous presence of malformation of the skin and nervous system (*Figure 1*).

Skin lesions can be indicators of occult spina bifida, and are important as they may connect to the spine and be a source of infection.

The presence of a 'herald mark' is an important sign in young people with lower lumbar pain or sphincter dysfunction.

Commonly associated lesions include: dimples, tufts and patches of hair, pigmented areas, achordons (pseudotails), lipomas, haemangiomas, dermoid cysts or sinuses.<sup>3</sup>

## Development of myelodysplasias — mechanisms and risk factors

Myelodysplasias are also called neural tube defects, which refer to their origin as developmental abnormalities of the central nervous system. These developmental abnormalities can occur at any site along the length of the brain and spinal cord (*Figure 4*).

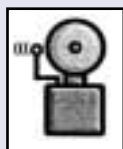
The critical development of the nervous system occurs in the first month after conception. If this process is disrupted, spina bifida may occur. When the brain does not develop, this results in anencephaly.

### Spina bifida occurs early in pregnancy

As spina bifida occurs during the first month of pregnancy, most cases will develop before most women know they are pregnant.

### Role of low folate

Low folate at the time of conception is one factor with an increased risk of myelodysplasias. Prevalence has decreased in areas where folate has been added to food supplies, such as bread.



### Genetic factors

There is a strong familial tendency for myelodysplasias, although the exact mechanisms are unknown. A family with one child with spina bifida has a 4% chance of a second child being born with spina bifida and a 10% risk after two children with the condition. Siblings of a person with spina bifida and adults with spina bifida have a 2% chance of having a child affected with spina bifida. All relatives of people with spina bifida require genetic counselling. Genetic counselling is recommended:

- for mothers older than 35 years
- for all mothers with previously affected children
- when either prospective parent has myelodysplasia. This includes all potential fathers with spina bifida.
- when any family history of myelodysplasia is present.

### Drug induced myelodysplasias

The antiepileptic drug valproic acid (Epilim, Valpro) causes spina bifida. Alcohol and some antipsychotic drugs have also been implicated as causes.

**Valproic acid can cause spina bifida.**

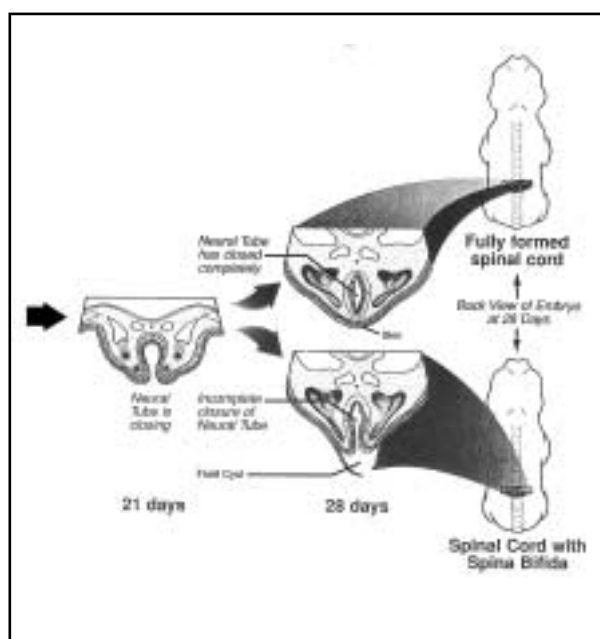


Figure 4. Development of spina bifida compared to normal development of the spinal vertebrae and central nervous system.

### Prenatal detection and management of spina bifida

Currently about one in 1000 pregnancies are affected with myelodysplasia although the number of new myelodysplasias is decreasing due to the increased use of routine folate and early detection of myelodysplasia through ultrasound scanning and serum alphafoeto-protein testing during pregnancy. Current management practice consists of:

- preconception genetic counselling usually by a geneticist, neurologist and obstetrician on the risks and benefits to enable patients to make informed decisions
- preconception folate
- antenatal diagnosis.

### Preconception genetic counselling<sup>4-7</sup>

Those at increased risk of having children with spina bifida are offered genetic counselling (see *Chapter 9 Organisations and further resources* and also *Table 1*). Some young people and adults may not be aware of these increased risks and clinicians should always assess whether their patients are familiar with the risks of pregnancy and refer accordingly.

### Preconception folate

High dose preconception folate supplementation is the cornerstone of reducing the risk of spina bifida in families at risk.

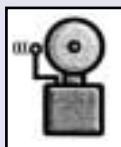
**Table 1. Preconception genetic counselling issues for those at increased risk of children with spina bifida**

- Discussion of risks of newly affected child
  - approximately 2% risk where one parent affected
  - approximately 4% risk where previous child is affected
- Attitudes of couples toward having a baby with neural tube defects including exploring their personal experience and understanding of these conditions.
- Discussion of possibility, risks and impact of anencephaly and spina bifida
- Role of preconception folic acid supplementation in reducing risk
- Availability and use of screening tests during pregnancy

While routine folate administration has been demonstrated to reduce the risk of neural tube defects on a population basis (such as 0.4–0.5 mg folate once daily), higher dosages of folate are routinely used in high risk patients (such as 5 mg once daily).

High folate supplementation is given in ALL high risk cases — not just to high risk mothers, but also to fathers in a group at high risk of developing spina bifida.

High dose folate supplementation is given to ALL high risk cases.



### Antenatal diagnosis

Ultrasonography can detect around 95% of cases of spina bifida by the age of 18–20 weeks. High risk pregnancies need to be screened by ultrasonographers experienced in the detection of neural tube defects. The first child with spina bifida in a family is often missed on ultrasound, as the pregnancy may be considered at low risk due to a lack of previous family history of spina bifida.

Expert ultrasound can provide an approximate guide regarding the severity and level of the neural tube defect to assist in determining the prognosis for the fetus.

Alphafetoprotein serum levels can be raised at the 16th week of pregnancy. Spina bifida is not the only cause

of a raised serum alphafetoprotein, but this is an indication for further testing. This especially applies to at risk pregnancies, such as in older women and individuals with, and relatives of those with, spina bifida. A typical screening schedule would include:

- 12 week expert ultrasound examination
- 18 week expert ultrasound examination
- option of maternal serum screening (for trisomy 21 and neural tube defects).

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# spina bifida

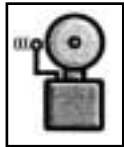
## Overview of spina bifida and the nervous system

### Chapter 2: The impact of hydrocephalus and other CNS conditions on case management

*Hydrocephalus affects most people with spina bifida and can impact upon continence management adversely in two main areas. Firstly, increased intracranial pressure can cause a deterioration of continence and mental status, which sometimes can be insidious and at other times dramatic. Secondly, the effect of cognitive deficits secondary to central nervous system abnormalities can be subtle, but can be a profound barrier to achieving successful case management, including continence control and independent living.*

#### Key issues for clinicians

- If there is any suspicion of raised intracranial pressure at all, patients with spina bifida need immediate referral to specialist centres for a full assessment. Any suspicion, no matter how small, is an indication for referral.
- Cognitive deficits secondary to hydrocephalus and other central nervous system abnormalities have a major impact on compliance with diagnosis, attending for investigations and following through with treatment and management plans.
- Clinicians can adopt strategies to help improve patient communication by understanding the types of cognitive problems that are common in people with spina bifida.



Hydrocephalus — an almost inevitable consequence of spina bifida

Hydrocephalus is not a specific disease, but rather a consequence of a diverse group of conditions resulting from impaired flow of cerebrospinal fluid (CSF).

Around 90% of infants born with spina bifida have hydrocephalus.

#### Raised intracranial pressure from hydrocephalus — a life threatening complication

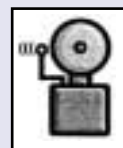
As neural tube defects affect the entire length of the brain and spinal cord, central nervous malformations are very common in people with spina bifida. One of the commonest types, the Arnold–Chiari malformation, often results in raised intracranial pressure early in childhood, requiring the surgical insertion of a ventriculoperitoneal shunt to divert CSF flow. Shunts can block at any time, causing a life threatening condition, as well as long term central nervous system damage which has the potential to severely interfere with independent living.

#### Impact of hydrocephalus on physical and cognitive deficits

##### Impact of cognitive deficits on incontinence management.

As most people with spina bifida have some central nervous system abnormalities, cognitive deficits secondary to brain dysfunction and other physical complications, especially hydrocephalus, are common.

Cognitive deficits secondary to complications have a major impact on compliance with diagnosis, attending for treatment investigations and following through with treatment and management plans. Clinicians need to be aware of the extent of these often subtle effects to ensure maximal adherence to management plans.



Good continence control usually involves fairly complex procedures and, to be effective, depends upon good planning. The clinician needs to understand any potential cognitive barriers to effective continence management.

### Detecting raised intracranial pressure<sup>1,2</sup>

The diagnosis of raised intracranial pressure can be difficult, but a high degree of suspicion is necessary to avoid the possible severe adverse effects of complications secondary to central nervous system damage.

Raised intracranial pressure can be of gradual onset, increasing over a few months, and can be an important cause of change in overall continence status.

A clear understanding of the pathophysiology of the condition helps the clinician in diagnosis.

### Hydrocephalus and mechanisms of raised intracranial pressure

Where is CSF formed?

Cerebrospinal fluid is primarily formed in the ventricular system of the brain by the choroid plexus, which is situated in the lateral third and fourth ventricles, although 25% of the CSF originates from extrachoroidal sources.

The total volume of the CSF is about 50 mL in infants and about 150 mL in adults. Most CSF is extraventricular.

CSF flow mechanisms

Cerebrospinal fluid flow results from a pressure gradient that exists between the ventricular system and venous channels. The fluid flows from the lateral ventricles through foramina (foramina of Monro) into the third ventricle, and then passes through a narrow aqueduct (aqueduct of Sylvius) which is only 3 mm in length and 2 mm in diameter in children. The CSF then exits the fourth ventricle through three foramina (two foramina of Luschka and the midline foramen of Magendie) into cisterns at the base of the brain.

After exiting the ventricular system of the brain, the CSF then circulates over the cerebral hemispheres and spinal cord, and is absorbed by the arachnoid villi and to a lesser extent, by the lymphatic channels of the paranasal sinuses.

Types of hydrocephalus — obstructive versus nonobstructive

Hydrocephalus resulting from obstruction in the ventricular system is called obstructive or

noncommunicating hydrocephalus. Hydrocephalus resulting from obliteration of the subarachnoid cisterns or abnormalities in functioning of the arachnoid villi is called nonobstructive or communicating hydrocephalus.

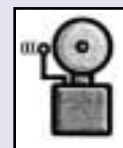
### Hydrocephalus in spina bifida

Hydrocephalus in spina bifida is usually due to the existence of the hindbrain malformation called the Arnold–Chiari malformation (type II) (see *Chapter 1*). Around a quarter of those with Arnold–Chiari malformations develop brainstem dysfunction, with symptoms often appearing in the first months of life.

The symptoms of raised intracranial pressure can mimic many other conditions, making diagnosis by even the most experienced specialist clinicians difficult. In young people and adults, raised intracranial pressure can be indolent, insidious and slow in onset, but can dramatically escalate over hours to a life threatening condition.

To avoid possible misdiagnosis, general practitioners need to have a low threshold for communication with specialist centres for assessment for advice.

If there is any suspicion of raised intracranial pressure at all, patients with spina bifida need to be referred to specialist centres for a full assessment. Any suspicion, no matter how small, is an indication for urgent referral.



### Causes of raised intracranial pressure in spina bifida

Many young people and adults with spina bifida will have ventriculoperitoneal shunts inserted within the first few months of life. In these individuals, the concern is that the shunt may become blocked, resulting in increased intracranial pressure.

Raised intracranial pressure secondary to Arnold–Chiari malformations can occasionally occur in later life resulting in spasticity, and abnormalities in gait and coordination during childhood.

### Presentation of increased intracranial pressure

Clinicians need to familiarise themselves with the presentation of this life threatening situation.

Raised intracranial pressure can be a cause of change in continence patterns. Any change should be examined closely for the possibility of raised intracranial pressure or other neurological causes such as spinal tethering.

## Treatment — ventriculoperitoneal shunt

Hydrocephalus is treated with the insertion of a ventriculoperitoneal (VP) shunt, usually within the first few years of life, to enable circulation of CSF and to reduce the intracranial pressure.

### Signs of raised intracranial pressure and VP shunt problems

Although this resource is aimed at health maintenance for young people and adults with spina bifida, the symptoms of raised intracranial pressure in children and infants are included, as it is such an important presentation (*Table 2*).

### Cognitive impact of hydrocephalus and other central nervous system conditions on patient management<sup>3</sup>

Hydrocephalus can result in a series of physical complications that can adversely affect cognition. These include memory abnormalities, attention problems, visual problems, behavioural problems including aggressive and delinquent behaviour, which all affect comprehension and adherence to any medical management plan.

While around 80% of people with spina bifida will have normal intellectual functioning, many will have subtle executive and cognitive problems that may affect the outcome of any medical management.

### Other causes of cognitive problems

In addition, there may be other structural central nervous system abnormalities contributing to cognitive deficits. Commonly prescribed agents such as antiepileptic and anticholinergic drugs can also interfere with cognition. A spina bifida specialist centre can help to address these issues, and contact with the centre will provide assistance in overcoming problems.

To help in GP consultations, a list of common problems and some concrete strategies follows.

### Common cognitive problems encountered in people with spina bifida

Organisational difficulties interfere with the ability to think or perform activities in a logical and planned way. This may manifest as difficulties in written language, learning sequences for procedures, keeping items and equipment in order, locating belongings or remembering to complete tasks.

Short attention span and distractability interferes with the ability to pay attention to important details of a new task. Brief attention spans mean the person may not learn all of the necessary information, or may forget or hear only

**Table 2. Signs of raised intracranial pressure and VP shunt problems**

This is a life threatening situation. Symptoms can occur over weeks but can escalate over hours. Always refer urgently to a specialist centre if any suspicious symptoms occur.

#### Adults, young people and children

- headache
- nausea and vomiting
- lack of appetite, refusal to eat
- increased irritability, lethargy, drowsiness
- personality changes
- disorientation
- pseudodementia
- visual problems: nystagmus, double or blurred vision; setting sun sign
- decreased motor and sensory function
- fits and seizures
- lower extremity hypertonia with generalised hyperreflexia.
- incontinence, especially a change in continence patterns

#### Infants

- bulging fontanelle
- increased head circumference
- irritability
- poor feeding
- impaired cognitive development
- respiratory stridor and/or high pitched cry in an infant

part of any instructions given, as well as taking longer to complete tasks. Distractability can be internal, from the person's own thoughts, as well as from the environment.

Ringling patients with a gentle reminder about appointment times may help them to remember to attend, although this has the potential to increase patient dependence. Encourage patients to remember their appointments by using a diary or the health planner diary in the companion volume to this supplement called *Passport to success*.

Language skills may be deceptive. There may be a stronger ability to say words than to comprehend their meaning. Despite what appears to be normal verbal skills, there may be a lack of comprehension of the words said. This may reflect use of rote memory of sounds rather than their meaning, as some people with spina bifida have very good auditory memories, but poor comprehension.

Perseveration, or repeating information over and over, can occur and the clinician may mistakenly perceive that they are being understood.

The cocktail party syndrome describes a speech pattern characterised by the habit of repeating back phrases used, saying memorised common phrases (such as 'How are you? Hi!') and talking about topics not always meaningful or appropriate to the situation. Cocktail party speech may be due to difficulty with inhibiting the flow of thoughts going through the mind or difficulty focusing on and comprehending relevant aspects of a situation

Difficulty in answering questions, following instructions, participating in back and forth conversation, or misinterpreting information or responding inappropriately to situations may indicate difficulties in actual comprehension.

Problems with abstract reasoning may cause difficulties in analysing and synthesising information and distinguishing between relevant and irrelevant information. This can also cause problems of generalisation and understanding complex information that involves words and concepts that cannot be seen or touched.

Visual-spatial difficulties can make simple tasks difficult. Tasks requiring judgments about visual and/or spatial information such as tying shoelaces, doing up buttons and zippers without looking can become difficult. Technical procedures, such as teaching self catheterisation, need to take these difficulties into account.

Lack of persistence causes difficulty in focusing on tasks requiring internal motivation or have a reward that is delayed. Tasks with immediate rewards are more likely to encourage persistence.

Lack of time management skills may result in difficulties organising daily tasks, doing things at the last minute, or not keeping to deadlines and appointment times. This behaviour can be interpreted as a lack of responsibility or caring about the needs of others, but may be a result of a deficit related to understanding the concept of time.

Passivity, avoidance and withdrawal may result when difficult situations are encountered, especially in a new context. Losing motivation to try new things may be a strategy to avoid failure. People who interact with the person may then focus on the behaviour rather than the learning difficulties that can result in unrealistic expectations or inappropriate programming.

## Strategies for maximising adherence to medical management

Learning to adapt treatment instructions to each individual's circumstances is an everyday skill for all clinicians. For people with spina bifida, there is no magic formula, but the following is a list of strategies for dealing with the commonest problems.

Work in conjunction with multidisciplinary teams, if possible, and be creative in finding methods that work for that individual person. Previous health personnel may have already identified the patient's learning strengths and weaknesses. Learn what you can about that person's pattern of learning. Also, if teaching a specific procedure, there may be specially qualified health personnel to assist, such as continence nurses.

Encourage tasks that are possible. Some people may believe that the tasks are too hard and may not have the belief that they are achievable. Helping to motivate a person's belief in themselves may be the first step to them achieving independence. Acknowledge all successes honestly and sensitively. Even if an outcome was not good, acknowledge the effort and attention involved.

Help improve comprehension by engaging eye contact whenever possible and have the person repeat back what was said.

Keep verbal explanations simple. There may be a tendency to over-explain tasks in an attempt to achieve understanding, but keeping explanations simple with specific, concrete language is likely to be more effective.

Adapt the methods used in the consultation to the person's strengths. For example, a person with problems comprehending language may have a better understanding by the use of diagrams and pictures, rather than repeating the same words over and over again. This will also help with any visual-spatial problems.

Ask the person to explain to you previous instructions to establish the level of comprehension. This may not necessarily occur at the time of the consultation, but at follow up visits, when the person has had time to process information.

Reward and promote persistence to help lessen frustration and avoidance when learning new procedures.

Reward success not only as an outcome, but also as an attempt. Empathise with any frustration felt during the attempt, even when the desired outcome has not been achieved.

Avoid interpreting unsuccessful tasks as behavioural problems as they may be due to cognitive deficits. This helps to focus the clinician's attention on overcoming

cognitive problems and reduces any frustration on the part of the patient.

Break tasks down into smaller steps and provide time and structured instruction for each step, and rewards for success as each stage is completed. Learn to do one step at a time.

Reduce demands to realistic levels. Rather than trying to achieve a whole series of tasks at once, try to spread tasks over time, so that goals are realistically achievable. This helps to lessen frustration.

Teach organisational skills such as 'Everything has a place and a place for everything'. A highly structured environment may make task completion easier.

Establish routines and structures as much as possible.

Use checklists, calendars and diaries.

Tape recorders are useful for some people and remove the need to take notes and may help them to remain attentive during listening.

Be conscious of the attention span. With experience, the clinician may be able to judge how much information can be taken on board at each session, and then tailor the consultation to the person's needs.

*Passport to success* includes a planning diary and has been produced in a user friendly way to help overcome the above barriers. The guide can be downloaded and provided to your patients.

[www.sbav.org.au](http://www.sbav.org.au) or [www.racgp.org.au](http://www.racgp.org.au)

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# Overview of spina bifida and the nervous system

## Chapter 3: Spinal cord tethering

*Tethering of the spinal cord is an insidious cause of worsening incontinence that requires urgent medical assessment and possibly surgical intervention to prevent any further deterioration. Clinicians monitoring young people and adults with spina bifida need to maintain a high degree of suspicion to ensure the early detection of tethered cord syndrome to help promote and maintain independent living.*

### Key issues for clinicians

- Clinicians need to consider spinal cord tethering as a possible cause of change in continence patterns.
- Any suspicion of spinal cord tethering should be referred to a specialist centre for urgent assessment.
- Earlier surgical intervention in clinically demonstrated spinal cord tethering is more likely to result in an improved long term outcome.

Tethered cord syndrome — an insidious condition

Tethering or stretching of the spinal cord in young people and adults results from fixation of the spinal cord to inelastic structures.<sup>1</sup> Spinal cord tethering is a major source of morbidity in spina bifida and clinicians need to be familiar with its presentation and maintain a high degree of suspicion when monitoring patients with spina bifida.

In the past, spinal cord tethering was thought of as mainly a condition affecting only children, especially during growth spurts, but the condition can occur in people with spina bifida at any age.

Spinal cord tethering is a particularly insidious complication of spina bifida that can have a major adverse impact on independence through its effect on mobility and continence.

Many causes but consistent presentation

The fixation of the spinal cord in spinal tethering is due to fibrous or lipomatous tissue. In spina bifida this can be due to a myelomeningocele, lipomyelomeningocele, scar tissue, a fibroadenomatous filum terminale or many other conditions.<sup>2</sup>

Clinical presentation of tethered cord syndrome

### Symptoms and signs

Tethered cord syndrome typically causes a progressive loss of function at or below the level of the spinal cord defect, with or without lower lumbar pain.

Regardless of the particular mechanical cause of spinal tethering, the spectrum of clinical presentations of tethered cord syndrome are consistent and should alert the clinician to the need for IMMEDIATE neurological and neurosurgical referral.



**Table 3. Characteristics of pain in spinal cord tethering**

- Pain in lower back and legs exacerbated by physical activity, especially any which involves flexion and extension of the lumbosacral area
- Leg pain is often in medial, lateral, anterior, or posterior aspects of thighs or legs
- Groin pain or pain in the genitorectal area is common
- Pain is not less when lying supine (as opposed to disc disease)
- Straight leg raising causes no difference to pain
- In adults, pain governed by three 'B' signs (usefulness will depend upon any pre-existing neurological and orthopaedic disabilities):
  - inability to sit with legs crossed (like Buddha)
  - difficulty in bending slightly at the waist with activities such as washing dishes
  - holding a baby or light material (< 2.5 kg) at the waist level while standing

**Table 4. Common findings in tethered cord syndrome**

- Pain (*Table 3*)
- Gait problems, often quite subtle
- Progressive urinary incontinence; symptoms include urgency, frequency and enuresis
- Progressive faecal incontinence including urgency, frequency, and encopresis
- Any motor or sensory deficits in lower extremities
- Altered sensation in genital regions and during sex
- Impotence
- Muscle weakness
- Muscle atrophy
- Hyporeflexia, especially any change in pre-existing signs
- Faecal incontinence
- Scoliosis/lordosis
- Foot deformities
- Skin abnormalities — herald marks

Tethering may occur in young adults at times of growth spurts, when lengthening of the spinal column can increase spinal cord tension, but it can occur at any adult age.

#### Key diagnostic issue — progressive loss and change

The key diagnostic issue in spinal cord tethering in young people and adults with spina bifida is a progressive deterioration in neurological function at or below the level of the defect. This includes urinary and faecal incontinence.

Spinal tethering needs to be excluded in the presence of progressive deterioration of urinary or faecal incontinence.

Neurological deficits in spina bifida are usually not progressive. Any change in signs requires immediate specialist assessment.

Spinal cord tethering can also occur in adults with no known past history of spina bifida.

#### Patchy distribution of clinical findings of tethered cord syndrome

The pattern of clinical findings of spinal cord tethering often fails to follow strict dermatomal patterns like those due to compression of one or two nerve roots or a particular level of spinal cord injury. Weakness, pain and

other signs of spinal cord tethering may have a patchy distribution below the level of the lesion, rather than a strict neurotomal pattern.

#### Pain in spinal cord tethering

Regardless of the cause, the characteristics of the associated pain are often suggestive of spinal tethering (*Table 3*).

While other causes of back pain, such as disc herniation, need to be excluded, clinicians still need to organise urgent neurosurgical referral and assessment to ensure that spinal cord tethering is not missed.

#### Other common findings in tethered cord syndrome

As highlighted above, the findings need to be considered in the context of any pre-existing neurological abnormalities, but clinicians should have a low threshold for specialist referral when there are any changes in pre-existing clinical findings (*Table 4*).

#### Management

##### Diagnostic imaging

Magnetic resonance imaging

Magnetic resonance imaging (MRI) is the best currently available technique for viewing the spinal

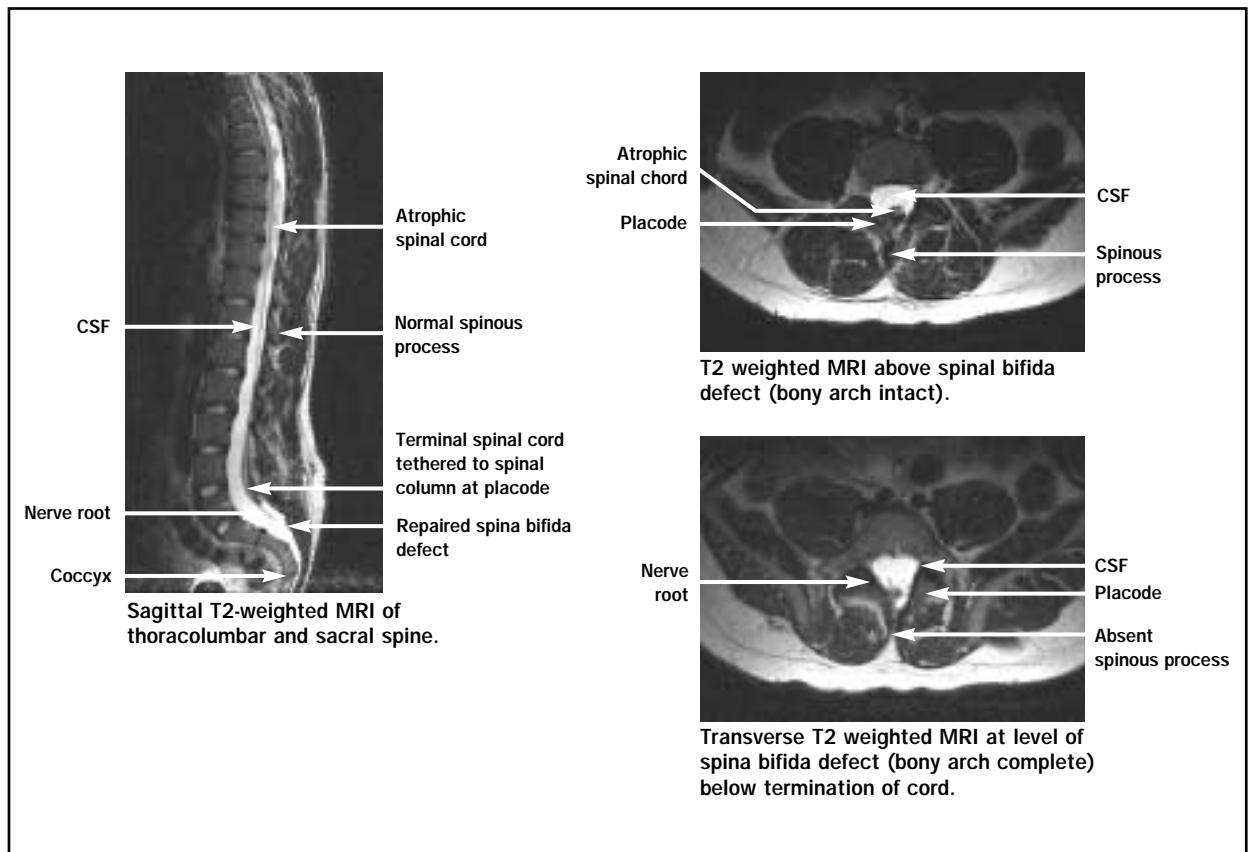


Figure 5. Surgically repaired spina bifida defect with absent spinous process and posterior spinal arch at low sacral level. Atrophic cord tethered to placode and spinal column. (Courtesy of Department of Diagnostic Imaging, Monash Medical Centre, Melbourne.)

cord. Most people with spina bifida will have some MRI findings suggestive of spinal tethering, but the decision to treat is based upon a combination of MRI and clinical findings. Ideally, a baseline MRI should be taken on all young people and adults with spina bifida to compare any changes with subsequent imaging, should symptoms of tethering arise (*Figure 5*).

Typical MRI findings in spinal cord tethering

Typical MRI findings in spinal cord tethering include

- thick filum terminale (>2 mm in diameter)
- presence of structures such as fibrolipomatous filum terminale
- obliteration of subarachnoid space suggesting caudal spinal cord or nerve root adhesion
- changes in the structure of the spina bifida lesion such as dermoid, epidermoid cyst, myelomeningocele, lipomyelomeningocele or other problem
- elongation of spinal cord
- posterior displacement of conus medullaris with the filum pressing against the thecal lining at or near L5, or when compared with previous films.

### Surgical intervention

Indications for detethering the cord

Once the diagnosis of spinal cord tethering is made, the decision for surgical intervention is based on clinical evidence. Treatment is especially indicated in the presence of new or worsening symptoms.

In studies of adults, when performed by experienced neurosurgeons, spinal cord tethering has been demonstrated to be a well tolerated, effective intervention.

Ultimately, the neurosurgeon can only confirm the presence of spinal tethering at operation. The surgery performed depends upon the intraoperative findings.

Effect of surgical intervention on spinal cord tethering<sup>3-5</sup>

Timely surgical intervention of tethered cord syndrome can arrest, and in some cases improve neurological signs. The more long standing the neurological signs, the less the chance of resolution of symptoms and signs.

Pain improves in many people and is usually relieved within three months of the detethering, but improvements in neurological signs can take many more months.

Patients need constant monitoring after the operation to assess recovery and help to adjust to any lifestyle changes.

**Case history: a 27 year old woman with worsening incontinence.**

M is a 27 year woman with spina bifida. She is independently mobile, has never used calipers, has no hydrocephalus, is fully employed, and is in a steady relationship

M presents with 12 months of progressively worsening faecal incontinence. She is now freely incontinent of faeces and manually evacuates herself before going out to avoid humiliation.

On examination, the anal tone is grossly reduced and at the time of assessment, anal sphincter reconstruction was being considered.

She has also had mild urinary frequency and urgency for six months, as well as eight months of reduced vaginal sensation during intercourse.

Her MRI demonstrated the presence of a tethered cord with a large neural placode. M was referred to a neurosurgeon and detethering is now planned.

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