

Introduction

This website has been written to help you understand more about lipomyelomeningocele, and to provide further information from the leaflet you may have received.

We hope it answers some of the questions you may have about its causes, diagnosis and treatment. It may also be useful for partners, friends, families and carers and to everyone who is concerned about how lipomyelomeningocele affects people and what can be done about it.

What Is It?

Lipomyelomeningocele is one of a number of conditions that are caused by irregularities in the development of the spinal cord. Conditions caused by disorganised growth of the spinal cord during pregnancy are called neural tube defects. The term spinal dysraphism may also be used.

This is a medical term used to describe when the spinal cord fails to form properly. In the case of lipomyelomeningocele, fatty cells can mistakenly attach to the spinal cord.

How Many People Have It?

Lipomyelomeningocele is classified as an 'occult' lesion. This can make them hard to detect. As a result, there may be many people who have lipomyelomeningocele but do not know that they have it (Finn and Walker, 2007). This makes accurately predicting the number of people who have lipomyelomeningocele difficult.

Rough estimates predict that between 1.6-2.5 births per 10 000 may be affected (McNeely and Howes, 2004, Kanev and Bierbrauer, 1995). The condition is also twice as common in women (Finn and Walker, 2007).

In Leeds, 5-10 babies a year are born with it.

What Causes It?

The spinal cord starts as a sheet of cells, which eventually form a tube (the neural tube), which becomes the spinal cord. If these sheets separate, fat cells can intrude into the spinal cord itself. This happens very early into a pregnancy, between days 24 to 28 (Drolet, 2004). Several complex mechanisms are thought to be involved.

The fatty mass causes tethering or pressure on the spinal cord. This can be harmful to the

spinal cord in the long term, and cause a variety of symptoms.

What Are The Symptoms?

The symptoms can be very variable and depend largely on the age of the patient. Young children often present with signs on the skin, whilst older patients present with other changes.

As many as 9 out of every 10 children with lipomyelomeningocele may present with an obvious abnormality on the skin (Pierre-Kahn et al., 1997). These can include changes in the colour of the skin, hairy patches or masses. These may be very obvious or quite hard to detect. Some examples are shown below (Finn and Walker, 2007):



Numerous other signs and symptoms may indicate a lipomyelomeningocele. Patients can have trouble with their bladder and urinary tract. This can present as difficulty passing urine or recurrent urinary infections. These sorts of symptoms can affect up to half of all patients (Sokol and Schwartz, 1973).

One in three patients may have problems with their legs (clubfoot), a curved spine (scoliosis) or hips (Finn and Walker, 2007). These are thought to arise from imbalance between different muscle groups.

Lower back pain, changes in sensation and a changed walking pattern in older patients may indicate a lipomyelomeningocele in an older patient (Pang and Wilberger, 1982).

Patients may have any combination of these signs and symptoms. Some are very subtle and can be very hard to detect outwardly. This may require specialist investigations. Some patients may be fortunate and not have any of these symptoms for many years, and in some cases may not ever have symptoms.

How Are They Investigated?

The evaluation of spinal lipomas has been improved by Magnetic Resonance Imaging (MRI) scans. The scan has a high sensitivity and specificity for fatty lesions. Scans play an important role in evaluating the anatomy of a lesion and detecting associated problems. Computed tomography (CT) scans can be used if MRI is contraindicated.

Other investigations may include an ultrasound of the spine, an X-Ray or specialised tests to assess bladder function, such as renal ultrasounds.

Your specialists will decide on a case-by-case basis which investigations are required.

What Are The Treatment Options?

Currently, the optimal treatment for lipomyelomeningocele remains unknown. Whilst there is agreement that symptomatic patients require treatment, a dilemma exists whether patients without symptoms should have surgery.

There are currently two options:

1. Watch and Wait, performing surgery when symptoms dictate
2. Early, or Prophylactic, Surgery, to try to prevent the symptoms occurring

Watch and Wait

A French study Pierre-Kahn and colleagues evaluated the outcomes of 53 who did not have surgery, but were closely monitored (Pierre-Kahn et al., 1997). From the patients studied, 25% showed signs of worsening symptoms over a 4 year period. When compared to results of early surgery at the same hospital, there was no significant difference in the risk of decline between patients who had surgery and those who did not. The rate of problems in patients opting for surgery appeared to be equal or slightly less than those who are managed conservatively.

Again, there are some weaknesses in these results. Lipomyelomeningocele can cause progressive deterioration throughout a patient's lifetime. This makes the follow up period of

Potential Investigations for

Lipomyelomeningocele

- Magnetic Resonance Imaging (MRI)
 - Spinal Ultrasonography
 - Plain Radiography
 - Urodynamic Studies
 - Serial Somatosensory-Evoked Potential Observation
 - Computed Tomography (CT) Myelography
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4.4 years relatively short term. It is hard to comment on the long term outcomes of patients without surgery after only 4 years.

In addition, the results of the early surgery at this centre are particularly disappointing when compared to other hospitals. The large number of complex cases or the time of surgery may explain this (from 1972 onwards). Since then surgical techniques have evolved considerably (Cohen, 2004).

Early Surgery

Surgical treatment aims to improve or stabilise symptoms in symptomatic patients and prevent future problems in patients without any symptoms (Bui et al., 2007). This is achieved through:

- preserving spinal cord tissue;
- untethering the cord;
- debulking the fatty mass;
- re-establishing normal anatomy in the region.

It is argued that early surgery offers improved outcomes compared to how the condition would progress naturally (Kanev and Bierbrauer, 1995).

Analysis of 270 paediatric cases from Chicago by LaMarca and colleagues (La Marca et al., 1997) found 16.7% of patients in the early surgery group had symptoms at 5 years.

However, there are some weaknesses in this study. Firstly, the rate of symptom development with an untreated lipomyelomeningocele remains unknown. Some patients will develop symptoms rapidly, whilst others may never develop them. The reasons for this remain unknown. With no knowledge of the number of older patients without symptoms, little of value can be said regarding the efficacy of early surgery when compared to the unknown progression of lipomyelomeningoceles (Kulkarni et al., 2004).

These arguments are summarised in the table below.

	Arguments For	Arguments Against
Early Surgery	<ul style="list-style-type: none"> • Some studies have shown that early surgery can prevent symptoms occurring in later life. • Some studies have shown that infants can develop symptoms by 2-4 years of age. • Some problems, such as bladder problems, may not be reversible with surgery. • Studies have shown that neurological problems following surgery are uncommon. 	<ul style="list-style-type: none"> • We currently do not know what happens to patients who do not have surgery in the long term. As such, we cannot say surgery gives a better outcome. • There is a risk that after surgery the spinal cord will re-tether, potentially producing symptoms in the future. • The surgery is not risk free.
Delayed Surgery	<ul style="list-style-type: none"> • Some children reach their teenage years without any symptoms developing. • In rare cases, symptoms may present in adulthood, or not at all. • Some studies have shown that there is no difference in outcome between patients who waited for surgery compared to those who had it early. 	<ul style="list-style-type: none"> • The numbers of older children/adults with the condition isn't accurately known. There may be very few patients who reach adulthood without any symptoms. • The follow up period for patients who opt against surgery is relatively short. This makes it difficult to assess the long-term outcome of not having surgery.

Other Considerations

Other factors also need to be taken into consideration. Patients with large skin masses are likely to request cosmetic surgery for debulking of the mass. Purely aesthetic procedures have been performed, yet cosmetic treatment of a lesion has been associated with future deterioration (Hoffman et al., 1985).

Also, the orthopaedic and urological dysfunction associated with lipomyelomeningoceles are normally irreversible once present. The only reports of patients with normal bladder function after long-term follow up are those who were operated on early (Atala et al., 1992).

Finally, the surgery itself is not without risk, although these risks appear to be declining with new techniques. Common complications include cerebrospinal fluid leakage, wound infection, meningitis and bladder or neurological injury. Non-neurological complications, whilst frequently minor and with no effect on long-term outcome, have been reported to affect 20% of cases. The incidence of neurological injury is between 0.1-1% (Byrne et al., 1995, La Marca et al., 1997).

What Are The Results of Surgery?

The outcomes of surgery are generally favourable. Most importantly, retethering of the spinal cord requiring further operative intervention has been reported in 15-20% of cases (Hoffman et al., 1985, La Marca et al., 1997, Pierre-Kahn et al., 1997).

The degree of symptom improvement after surgery varies according to the nature of the preoperative symptoms. Pain and spasticity respond well to surgical treatment, whereas improvements in sensation and bladder function are related to the length of time they have been present. Long-term bladder problems are less likely to be completely cured than those that have been relatively short (Huttmann et al., 2001). That said, many surgeons report marked improvements in urological function following surgery, although complete recovery is rare (Bui et al., 2007).

How Do Things Work In Leeds?

In Leeds and across Europe, the 'watch and wait' approach is favoured. Your specialist will be able to explain the benefits of this method in more depth.

A specialist clinic is held once a month at Leeds General Infirmary. Here, doctors experienced in treating this condition will provide further information, perform examinations and discuss potential investigations or treatments. You are likely to be given an appointment

for every 3-6 months to closely monitor your child's development. The length of time between appointments may be extended when certain milestones, such as bladder and bowel control are met.

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