Spina Bifida and Hydrocephalus

Aims

Condition specific aims

RCPCH Level	Learning outcome	
1	4	Recognises and initiates investigations of the common causes of hydrocephalus.
1	4	Recognises and manages blocked ventricular shunts.
2	4 (17)	Recognises and manages the complications of neural tube defects.
2	4 (16)	Investigates and manages other voiding disorders according to clinical guidelines.

Generic aims

RCPCH Level	Learning outcome	
1	4 (19)	Recognises the impact of mental health problems on physical health and the impact of physical health problems on mental health.
1	4 (19)	Manages the CYP presenting with deliberate self-harm.
2	4 (14)	Develops strategies and skills to support and engage parents of children with emotional or mental health difficulties.
2	4 (14)	Assesses and manages acute presentations of psychological distress (self-harm) and refers on when appropriate.

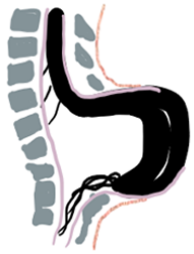
What are neural tube defects?

Neural tube defects (NTDs) are amongst the most common birth defects. The prevalence has dropped in the UK from around 4.5 per 1000 live births to less than 1 per 1000 live births. This is attributed to a combination of screening and preconception folate supplementation.

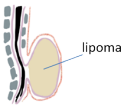
NTD is a broad term encompassing a heterogeneous group of congenital spinal anomalies that result from defective closure of the neural tube early in foetal life. The upper and lower ends of the neural tube close last and these tend to be common sites of the anomalies. There are 2 broad categories: open lesions when the skin fails to close in the midline, and closed where there is skin coverage. The presentation varies with both the site and the extent of the lesion.

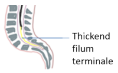
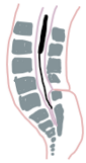
Open neural tube defects These describe lesions when there has been failure to close the neural tube (either brain or spinal cord) leading to herniation of neural tissue through a bony and skin defect where there has also been a failure of midline fusion in utero.

	Malformation	**Clinical course**
	**Anencephaly**: Forebrain fails to form	A lethal condition. Any infants born alive do not survive beyond a few hours.
	**Encephalocele**: Herniation of meninges and brain	If there is significant brain herniation then infants will die in the new born period.

	**Spina bifida manifesta** Meninges plusmn spinal cord protruding through a defect in the posterior spine (meningocele/ myelomeningocele). This is an example of open spinal dysraphism as vertebra and failed to close.	The spinal cord defect will require surgical closure at birth and there will be long-term neurological deficits

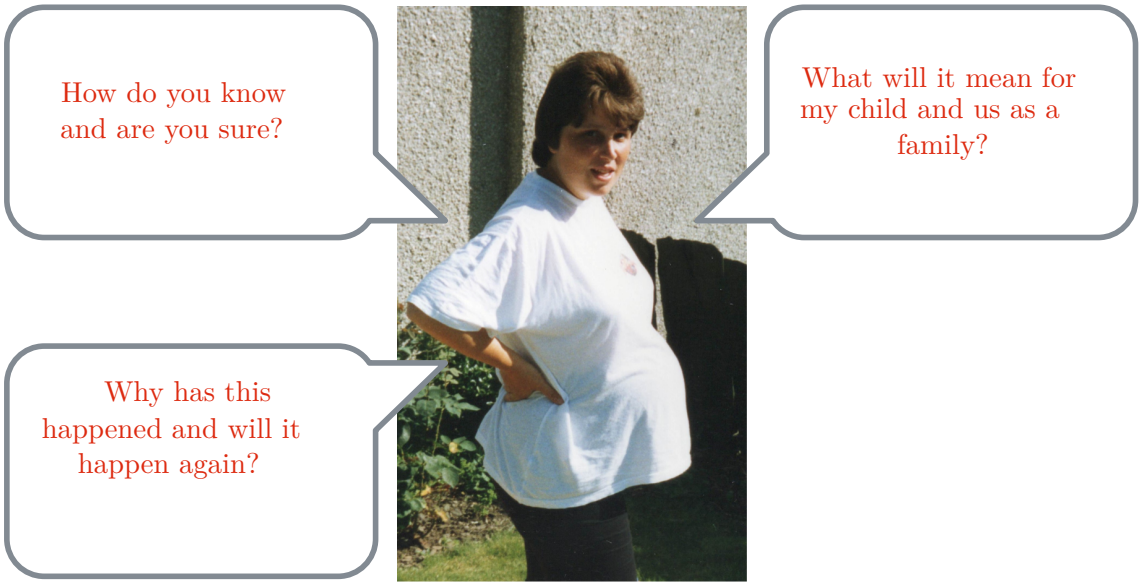
Closed neural tube defects

	Malformation	**Clinical course**
	**Spina bifida occulta** : Failure of spinal arch to close	These can be seen clinically by a tuft of hair, or deviated natal cleft. The incidence of spina bifida occulta is high (~10% of the population) and most are asymptomatic and incidentally recognized on routine radiography.
	**Lipomeningocele**: A meningocele with an associated fatty mass	This is presents sent as fatty mass over the spine – requires imaging to clarify if dysraphism or cord involvement.

	**Thickened filum terminale**	This presents during childhood due to thethering of the cord with problems with leg movement, bladder and bowel control, lower back pain and spinal deformity. The presentation may be delayed until adulthood. It is important to diagnose and intervene as early as possible to minimize functional loss.
	**Dermal sinus**	A pit at the base of spine, for which the base cannot be seen. If missed on neonatal examination it presents as meningitis in infancy.

Life story – antenatal

Fiona is a 28 year old young woman in her first pregnancy. Although this was unplanned she is delighted with the news and attends for routine antenatal screening which shows that there are signs of spina bifida. You have been asked to meet her and she has written down the questions she would most like to ask you.



How do you know and are you sure?

All pregnancies in the UK are offered the 'triple test', which is a blood test for alpha fetoprotein (AFP), beta human chorionic gonadotrophin and oestriol. Many places add in a nuchal translucency test which helps to identify foetus's with Downs syndrome and thus becomes a quadruple test. A raised AFP can be associated with incomplete closure of the spinal canal. In addition all women have an antenatal ultrasound. Together, these tests offer a 70% sensitivity marker of Down's syndrome, Edwards Syndrome and Patau's syndrome. Foetal ultrasonography looks particularly at the development of the spinal cord and canal, which in the hands of an expert radiographer will demonstrate the lesion which can then be confirmed non-invasively with a foetal MRI scan. Some babies may be offered antenatal surgery to close the neural tube defect and the evidence suggests that this decreases the likelihood of developing hydrocephalus.

Why has this happened and will it happen again?

Factors associated with spina bifida include:

- Spina bifida is more common among Hispanic and white people of Northern European descent. Genetic studies have identified 42 candidate genes and more are likely to be found as technology advances.
- Folic acid deficiency – taking folate supplements decreases the risk of spina bifida.
- Diabetes – particularly if poor control of diabetes in early pregnancy.
- Maternal obesity.
- Medications – in particular sodium valproate (Epilim) increases the risk of spina bifida by around 4-5 times. Young women should not be treated with sodium valproate for epilepsy unless all other treatments have been tried. The advice is that if on sodium valproate they should also be using contraception and they are aware of the risk birth malformation in particular spina bifida and cleft palate. To ensure women are aware there is now a pregnancy prevention programme for women on valproate and they need to sign a form on an annual basis to make sure clinical teams have counselled them.
- https://www.gov.uk/drug-safety-update/valproate-pregnancy-prevention-programme-actions-required-now
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- Family history – Couples who have had one child with a neural tube defect have a slightly higher chance of having another baby with the same defect. That risk increases if two previous children have been affected by the condition. In addition, a woman who was born with a neural tube defect, or who has a close relative with one, has a greater chance of giving birth to a child with spina bifida.

Prevention:

- All women should take folate (400mcg) if planning a pregnancy, and for the first 12 weeks of pregnancy. If they or a close family member has previously had an

affected child in the family then they should take a high dose folate supplement of 4mg again for one to three months prior to conception and for the first three months of pregnancy.

- If diabetic optimise diabetic control prior to conception.

- If on sodium valproate young women should be changed to a suitable alternative anti epileptic in consultation with the team managing their epilepsy prior to planning a pregnancy

What will it mean for my child and us as a family?

- **Will my baby survive the pregnancy and delivery?**

  Almost certainly your baby will be born alive. It can be helpful to see pictures of babies with previous lesions in order to prepare themselves for delivery. It is also important to explain that this is an issue about the way the spinal cord was formed and hat whilst the baby will have specific needs that will have to be promptly addressed after birth they are not otherwise ill or fragile.

- **What will happen after my baby is born?**

  When the baby is born there will be a lot of people in the room to make sure the baby is immediately appropriately cared for. That the baby will need to go to special care baby unit and if this is an open lesion will require immediate surgery. Some babies may develop hydrocephalus, or increased fluid in the central spaces within the brain that may also require treatment with a tube or shunt. We will also need to help you baby empty their bladder by putting a tube into their bladder. It is helpful for parents to have a leaflet about the neonatal unit so they know what this will look like and what to expect.

- **In what ways will I have to adapt my parenting to meet my child needs?**

  You will be helped to learn skills to keep your child well which will include learning to use a catheter to empty their bladder, signs to look out for suggesting problems

with their shunt and techniques to help with their mobility. There are ways for an older child to be socially continent and ways to optimise their independent mobility.

- **Will one of us have to stop working to meet our child's needs?**

  All babies need a lot of care and your baby is no different, but the way you care for them may be different; you may need to learn to catheterise rather than change a nappy. Therapy and nursing staff will be happy to teach these skills to anyone caring for your child, so it should be possible to keep working.

- **Will my child have a happy life?**

  Most children lead very happy and fulfilled lives, and lots live independently as adults. Shine https://www.shinecharity.org.uk is a charity that supports people with spina bifida and their families. They have an excellent website that will help you to see how people have lived inspiring lives.

- **Will my child be able to talk? How will it affect their learning?**

  Most children with spina bifida and/or hydrocephalus are able communicators, have intellectual abilities within the typical range for their age and will go to mainstream schools. Some children with spina bifida and/or hydrocephalus will have learning difficulties, some of whom will go to more specialist schools, depending on their individual needs and choices. With appropriate support, based on assessment of individual needs, all children with spina bifida and/or hydrocephalus should be able to participate in most activities and enjoy good quality of life.

- **Will it affect the length of my child's life?**

  Most children with spina bifida will have medical issues and it is important that these are managed well as they impact on both the quality of the child's life and also its length. There are also other medical issues that can crop up, such as hip problems, but these are not because parents have missed caring in an appropriate way. In recent years, with improved understanding of long term needs and the

importance of specialist follow-up into adulthood, instances have greatly reduced of secondary issues such as renal impairment which used to be seen in adults with spina bifida. This has been associated with an increase in lifespan.

More information on how to approach antenatal counselling #.

Medical issues associated with spina bifida

Hydrocephalus (present in 80-90%).

Coordination problems, attention problems, seizure (23% have 1 seizure).

If open lesion this will require closure at birth. Can develop tethered cord (32%). Later life children can develop scoliosis (43% require spinal fusion).

Respiratory difficulties - rare that brainstem is affected.

Neuropathic bowel and bladder

(present in 95% - but 80% will develop social bladder continence).

When older, sexual dysfunction.

Weakness and low tone below the level of the lesion. This can be associated with hip dysplasia and talipes.

Approximately 10% of children with spina bifida develop latex allergy and therefore latex free products should be used

Skin will lack sensation below the level of the lesion leading to increased risk of pressure sores.

Weakness and low tone below the level of the lesion

The symptoms and signs in spinal cord lesions can be predicted from the anatomy. Important factors to consider are:

1. The spinal level.

2. The site in the spine – before or after the last cell body will determine if it is an upper or lower motor neurone sign.

3. Whether it is affecting the whole cross-section of the cord or only a part of it.

4. The underlying cause – in this case we are thinking about spina bifida. Acute injuries to the spine such as trauma present with features of spinal shock.

There can be orthopaedic complications as a longer term consequence including hip dysplasia and talipes. It is helpful to warn families about these so they do not feel it is their fault.

Sexual dysfunction

Sexual dysfunction can be common in young people with spina bifida. It can affect their relationships, sex lives and fertility.

Early referral to appropriate experts in sexual health including urology and gynaecology can be discussed with young people, based on their individual needs and choices.

Hydrocephalus

Video playlist

Neuropathic bladder

Video playlist

Insensate skin

Pressure sores can bechallenging and are best prevented. It is important to treat early and involve tissue viability experts about management. Pressures sores are more likely if the young person is obese or has a spinal curvature as this increases specific areas of pressure on the skin. For this group other skin problems include latex allergy from repeated exposure are common and it is advisable for these children to avoid all latex products from birth.

Tips on pressure sore prevention

Lift or shift bottom from chair every 20 minutes and change position of legs at the same time. Check skin all over at least once a day (twice is better). If wet or soiled, the quicker the clean up and change, the better.

Wheelchair users should take care with transfers to avoid skin damage.

Statistics from: *Spina bifida outcome: a 25-year prospective study.* R M Bowman RM, D G McLone D G , J A Grant JA et al . Pediatr Neurosurg. 2001 Mar;34(3):114-20.