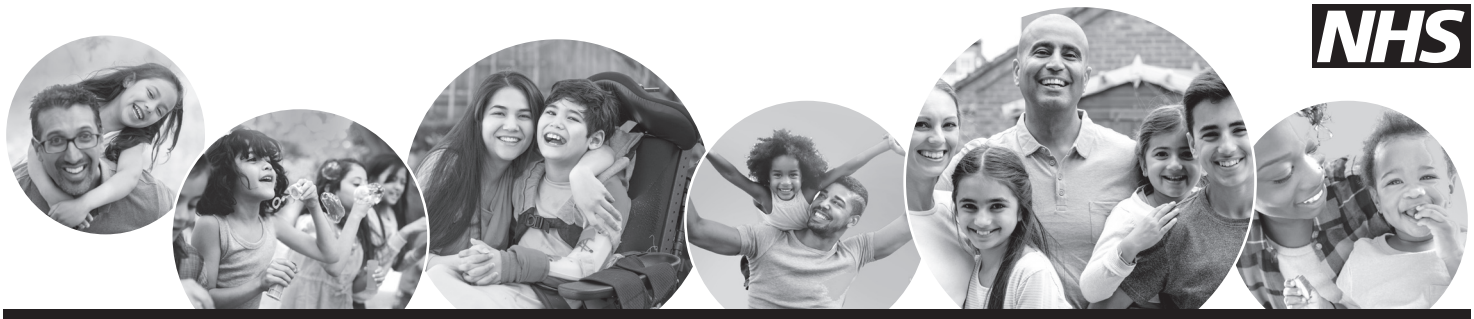
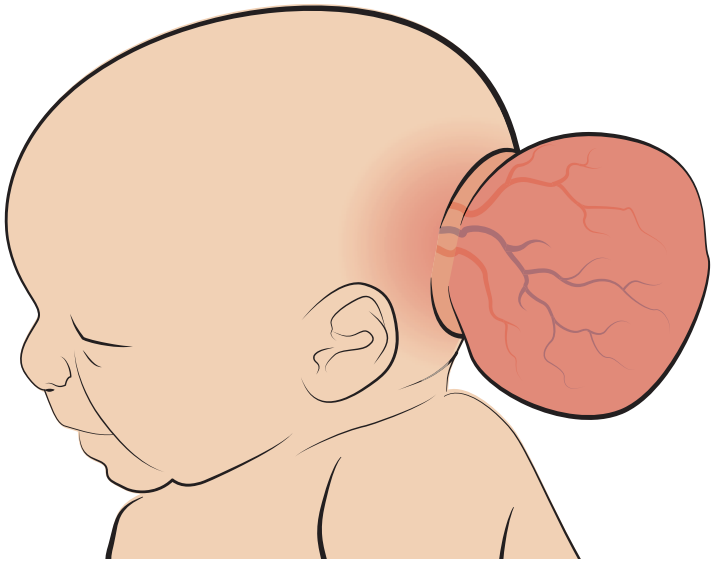


Encephalocele

An encephalocele is a rare congenital (present at birth) type of neural tube defect where part of the skull has not formed properly so a portion of brain tissue and associated structures are outside the skull. The protruding sac may be covered with skin or it may be covered with a thin membrane. This information sheet from Great Ormond Street Hospital (GOSH) explains the causes, symptoms and treatment of encephalocele – a type of neural tube defect – and where to get help.

If just the covering of the brain (meninges) are outside the skull, it tends to be called a meningocele. If both brain tissue and meninges are outside, this can be called an encephalomeningocele.

It can affect any area of the skull: the middle of the back of the head, the middle of the top of the head or the nose/forehead region. If it has formed in the nose/forehead region, this tends to be called a 'midfacial cleft', which is explained in a separate information sheet.



What causes an encephalocele?

In early development, the brain and spinal cord start as a tube-like structure called the 'neural tube' that is open at either end. These openings close within the first weeks of pregnancy, and the neural tube continues to grow and fold, eventually forming the brain and spinal cord. If the tube fails to close properly, this results in a group of problems called 'neural tube defects'. We do not really understand what causes neural tube defects in general but we do know that folic acid can reduce the risk of them happening in future pregnancies. More information about this is at the end of this information sheet.

In most cases, an encephalocele has developed sporadically (out of the blue) and is not passed on from parent to child. However, an encephalocele can be a feature of various syndromes (collection of symptoms often seen together), such as Dandy Walker syndrome, Chiari malformation or many others, which may have a genetic component. We do know that if there is a family history of neural tube defects, there is an increased risk of having a child with an encephalocele.

Encephaloceles affect between 1 and 2 in every 10,000 births. Racial background seems to have an effect on the location of the encephalocele: in Western babies it is more common to have a

'posterior' encephalocele affecting the back of the head, whereas in Eastern babies an 'anterior' encephalocele affecting the front of the head is more common. We also know that females are more likely to have a posterior encephalocele whereas males tend to have an anterior encephalocele.

What are the signs and symptoms of an encephalocele?

The location of the encephalocele is an important factor in the plan for treatment and outlook, as is the type and amount of brain tissue outside the skull. Very large encephaloceles may be incompatible with life so affected babies may not survive pregnancy. Anterior encephaloceles are less likely to contain brain tissue so tend to produce fewer symptoms.

The symptoms present with an encephalocele are very variable. Hydrocephalus occurs when cerebro-spinal fluid (CSF) builds up within the ventricles (cavities) of the brain resulting in increased pressure on the brain. The portion of the brain tissue being outside the skull reduces the CSF flow. CSF is a watery liquid that surrounds the brain and spinal cord, acting as a 'cushion'. It also supplies nutrients to the brain.

Some children show some signs of developmental delay, that is, they reach their 'milestones' such as sitting, crawling or walking, later than other children of a similar age. They may be smaller than children of a similar age as well. They may have learning disabilities which continue throughout life. Other children have seizures (fits or convulsions) or visual impairment.

Despite this, many children with encephaloceles have no symptoms at all other than the lump itself. In these cases, many parents opt to have the encephalocele removed for cosmetic reasons and due to worry about injuries later in life, for instance, when playing sports.

How is an encephalocele diagnosed?

If the encephalocele is large, it may be seen on routine prenatal ultrasound, which may allow planning of a caesarean section if it could be risky to have the baby vaginally. Otherwise, in most cases an encephalocele will be visible at birth so is easily diagnosed. Very small encephaloceles, especially those in the nose/forehead area may not be so visible.

Once an encephalocele is suspected, the diagnosis will usually be confirmed with imaging scans, such as magnetic resonance imaging (MRI) scans. This will allow doctors to see exactly how much of the skull is affected and whether the sac contains meninges or brain tissue or both. As encephaloceles can be associated with other problems, so the doctors will examine your child closely to check if this is the case.

How is an encephalocele treated?

Encephaloceles always require correction with an operation under general anaesthetic. The timing of surgery will vary depending on whether the sac is covered with skin or a thin membrane. If it is only covered with a thin membrane, surgery soon after birth will be needed to prevent infection and damage/drying of the exposed tissue. If there is a covering of skin, surgery can be delayed for a month or two to allow the baby to grow and develop.

The operation to correct an encephalocele involves the surgeon making an incision near the encephalocele, cutting through the tough covering of the brain (dura) and removing a portion of skull bone to enable them to reposition the meninges and/or brain tissue back inside the skull. If hydrocephalus is present, they will insert a shunt to drain the excess CSF into the abdomen. They can then repair the dura and replace the skull bone, either with the portion removed or with a patch of artificial material if there is not enough bone to close the opening. Finally, they will close the incision and apply a dressing.

Close monitoring will be needed for the first few hours after the operation to ensure that the CSF is not leaking and the shunt (if used) is working properly. This will involve taking regular neurological observations, at least for the first night after the operation.

What happens next?

The outlook is variable depending on the location of the encephalocele and the amount of meninges/brain tissue outside the skull. It will also depend on any other problems present and if the encephalocele is part of a syndrome.

The outlook for a proportion of children with an encephalocele is good with few long term effects on bodily functions such as breathing, vision and hearing. Children with learning disabilities will benefit from support in education and day to day life although a degree of independence may be possible. The majority of children with no learning disabilities grow up to lead a normal life, working and raising a family.

There is now evidence that an adequate intake of folic acid can dramatically reduce the risk of encephalocele occurring in future pregnancies. If you are planning a further pregnancy, we recommend that you take 4mg of folic acid each day for at least three months before conception and for the first three months of pregnancy. This dose is higher than the standard recommendation for women who have not previously had a child with a neural tube defect. If you have any concerns about future pregnancies, please talk to us as we can arrange a consultation with a genetic specialist for you.

Further information and support

As an encephalocele is a type of neural tube defect like spina bifida, the support group SHINE can offer support and advice. Call them on 01733 555 988 or visit their website at www.shinecharity.org.uk

Compiled by the Neurosurgery department in collaboration with the Child and Family Information Group
Great Ormond Street Hospital for Children NHS Foundation Trust, Great Ormond Street, London WC1N 3JH
www.gosh.nhs.uk

