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
cencephalocele?
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
cencephalocele 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