

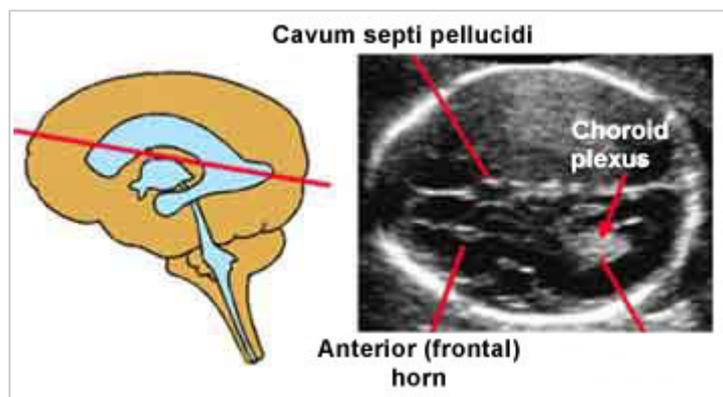
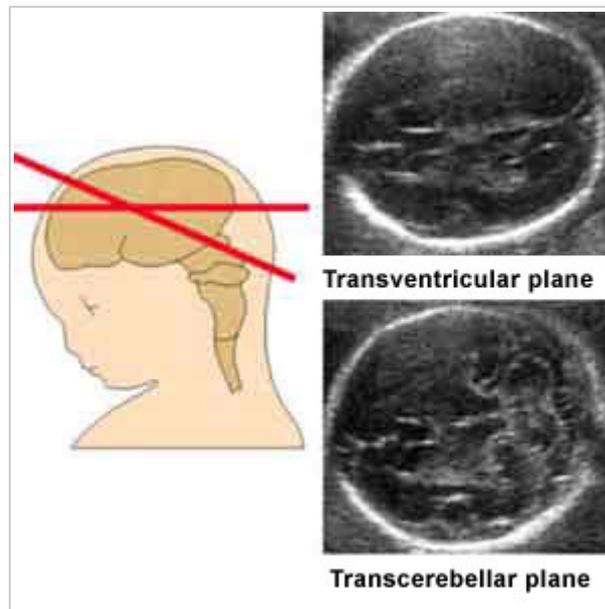
Chapter 2

Central nervous system

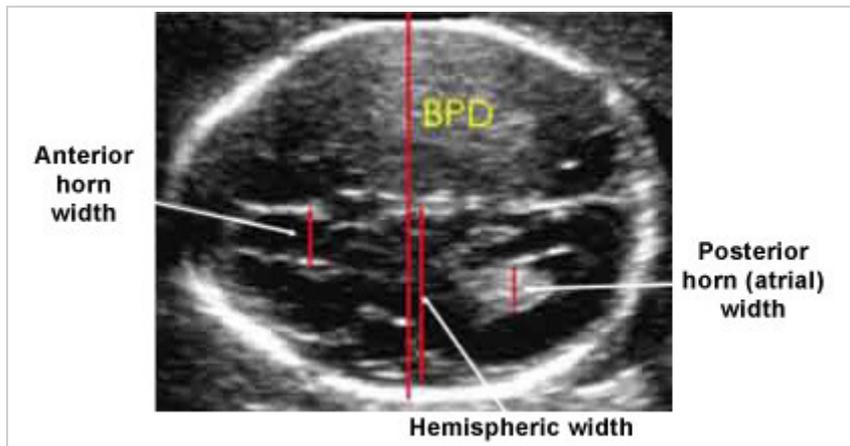
NORMAL SONOGRAPHIC ANATOMY

The fetal brain undergoes major developmental changes throughout pregnancy. At 7 weeks of gestation, a sonolucent area is seen in the cephalic pole, presumably representing the fluid-filled rhombencephalic vesicle. At 9 weeks, demonstration of the convoluted pattern of the three primary cerebral vesicles is feasible. From 11 weeks, the brightly echogenic choroid plexuses filling the large lateral ventricles are the most prominent intracranial structures. In the early second trimester, the lateral ventricles and choroid plexuses decrease in size relative to the brain mass.

Examination of the fetal brain can essentially be carried out by two transverse planes, commonly referred to as the transventricular and the transcerebellar plane.

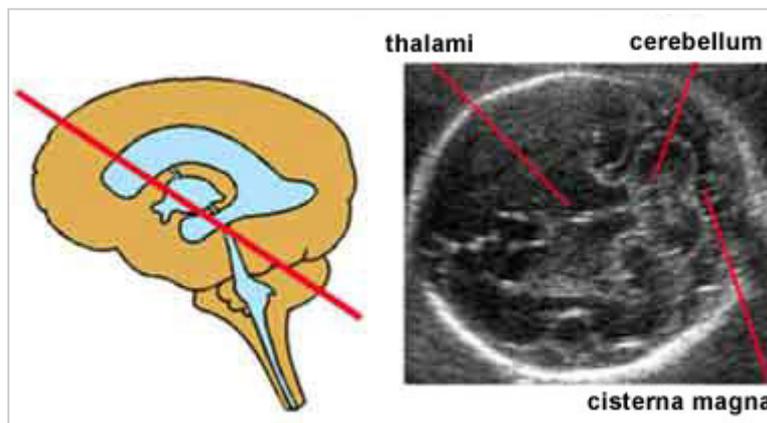


The transventricular plane, obtained by a transverse scan at the level of the cavum septum pellucidum will demonstrate the lateral borders of the anterior (or frontal) horns, the medial and lateral borders of the posterior horns (or atria) of the lateral ventricles, the choroid plexuses and the Sylvian fissures.

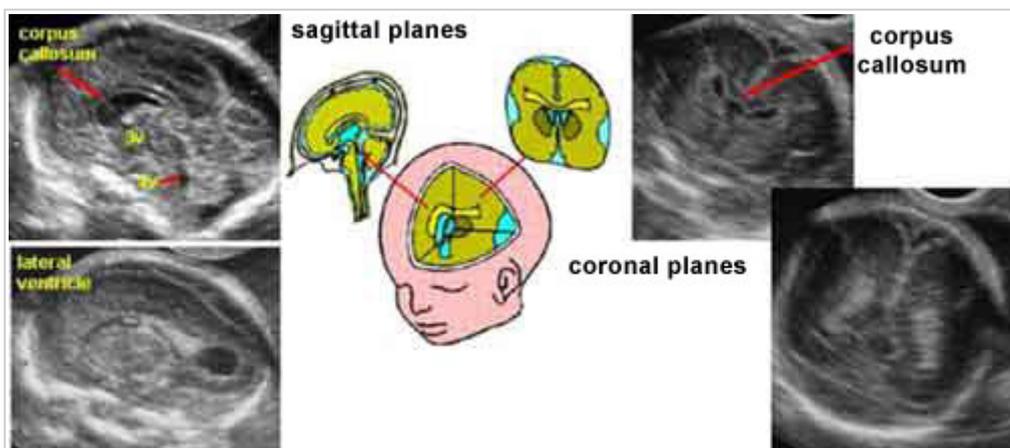


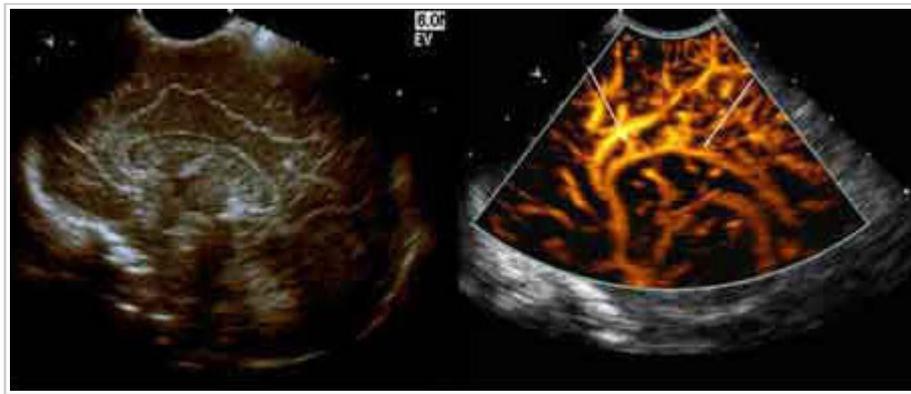
The transventricular view is used for measurement of the biparietal diameter (BPD), head circumference (HC), and width of the ventricles.

The transcerebellar (or suboccipitobregmatic) view allows examination of the mid-brain and posterior fossa; this view is used for measurement of the transverse cerebellar diameter (TCD) and cisterna magna (CM).



Additional scanning planes along different orientations may be required from time to time to better define subtle details of intracranial anatomy in selected cases. Reverberation artifacts usually obscure the cerebral hemisphere close to the transducer. Visualization of both cerebral hemispheres would require sagittal and coronal planes that are often difficult to obtain and may require vaginal sonography.





Transvaginal Scan + Color Doppler (Sagittal plane) Vascularization of Brain (arrow - Pericallosal Artery)

Luckily unilateral cerebral lesions are rare and are often associated with a shift in the midline echo. Therefore, we adhere to the approach that in standard examination only one hemisphere is seen, and symmetry is assumed unless otherwise proven.

A sagittal and/or coronal view of the entire fetal spine should be obtained in each case. In the sagittal plane the normal spine has a 'double railway' appearance and it is possible to appreciate the intact soft tissues above it. In the coronal plane, the three ossification centers of the vertebra form three regular lines that tether down into the sacrum. These views are used to assess the integrity of the vertebrae (to rule out spina bifida) and the presence and regularity of the whole spine (to rule out sacral agenesis and scoliosis).



Whether a systematic examination of each neural arch from the cervical to the sacral region in the transverse plane is necessary is debatable. This is certainly required for patients at high-risk of neural tube defects. In low-risk patients, intact cerebral anatomy rules out more than 90% of cases of spina bifida and we believe that the longitudinal / coronal scan may suffice.

NEURAL TUBE DEFECTS

These include anencephaly, spina bifida and encephalocele. In anencephaly there is absence of the cranial vault (acrania) with secondary degeneration of the brain. Encephaloceles are cranial defects, usually occipital, with herniated fluid-filled or brain-filled cysts. In spina bifida the neural arch, usually in the lumbosacral region, is incomplete with secondary damage to the exposed nerves.

Prevalence

This is subject to large geographical and temporal variations; in the UK the prevalence is about 5 per 1,000 births. Anencephaly and spina bifida, with an approximately equal prevalence, account for 95% of the cases and encephalocele for the remaining 5%.

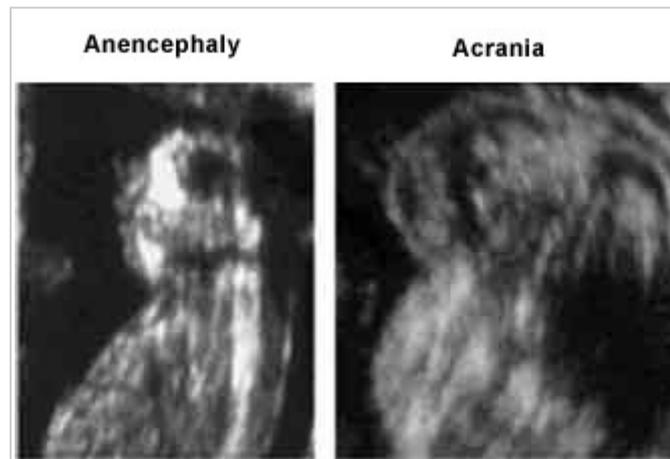
Etiology

Chromosomal abnormalities, single mutant genes, and maternal diabetes mellitus or ingestion of teratogens, such as antiepileptic drugs, are implicated in about 10% of the cases. However, the precise etiology for the majority of these defects is unknown. When a parent or previous sibling has had a neural tube defect, the risk of recurrence is 5-10%.

Periconceptual supplementation of the maternal diet with folate reduces by about half the risk of developing these defects.

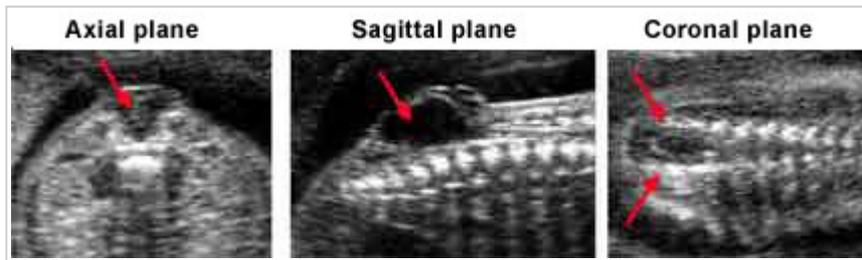
Diagnosis

The diagnosis of anencephaly during the second trimester of pregnancy is based on the demonstration of absent cranial vault and cerebral hemispheres. However, the facial bones, brain stem and portions of the occipital bones and mid-brain are usually present. Associated spinal lesions are found in up to 50% of cases. In the first trimester the diagnosis can be made after 11 weeks, when ossification of the skull normally occurs. Ultrasound reports have demonstrated that there is progression from acrania to exencephaly and finally anencephaly. Not everyone agrees however, since acrania are defects of the mesenchymal layer and there is no evidence in the literature of recurrence rate. In the first trimester the pathognomonic feature is acrania, the brain being either entirely normal or at varying degrees of distortion and disruption.

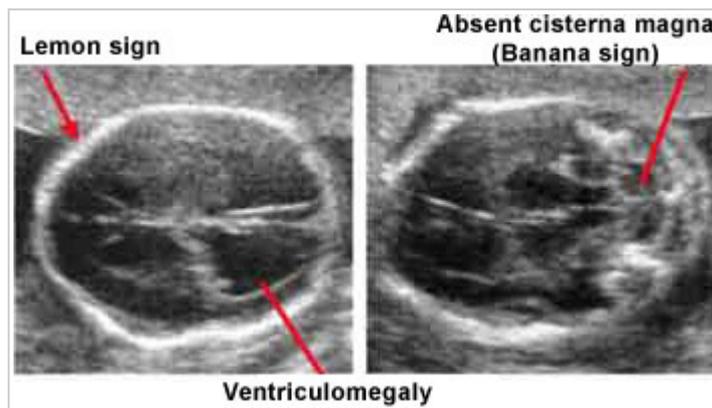
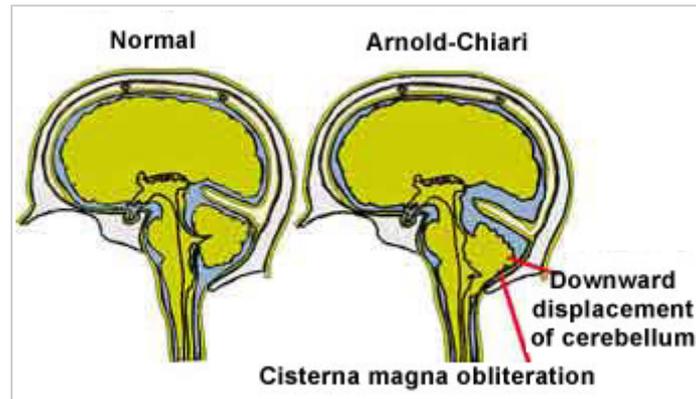


Anencephaly (3D view)

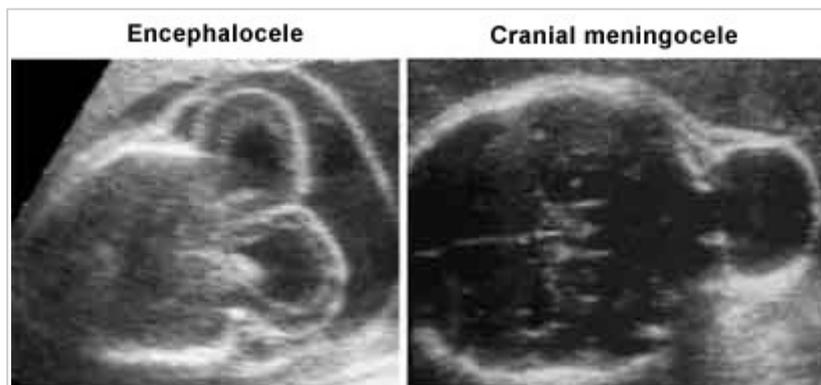
Diagnosis of spina bifida requires the systematic examination of each neural arch from the cervical to the sacral region both transversely and longitudinally. In the transverse scan the normal neural arch appears as a closed circle with an intact skin covering, whereas in spina bifida the arch is 'U'-shaped and there is an associated bulging meningocele (thin-walled cyst) or myelomeningocele. The extent of the defect and any associated kyphoscoliosis are best assessed in the longitudinal scan.



The diagnosis of spina bifida has been greatly enhanced by the recognition of associated abnormalities in the skull and brain. These abnormalities are secondary to the Arnold-Chiari malformation and include frontal bone scalloping (lemon sign), and obliteration of the cisterna magna with either an "absent" cerebellum or abnormal anterior curvature of the cerebellar hemispheres (banana sign).



These easily recognizable alterations in skull and brain morphology are often more readily attainable than detailed spinal views. A variable degree of ventricular enlargement is present in virtually all cases of open spina bifida at birth, but in only about 70% of cases in the mid-trimester.



Encephaloceles are recognized as cranial defects with herniated fluid-filled or brain-filled cysts. They are most commonly found in an occipital location (75% of the cases) but alternative sites include the frontoethmoidal and parietal regions.

Prognosis

Anencephaly is fatal at or within hours of birth. In encephalocele the prognosis is inversely related to the amount of herniated cerebral tissue; overall the neonatal mortality is about 40% and more than 80% of survivors are intellectually and neurologically handicapped. In spina bifida the surviving infants are often severely handicapped, with paralysis in the lower limbs and double incontinence; despite the associated hydrocephalus requiring surgery, intelligence may be normal.

Fetal therapy

There is some experimental evidence that *in utero* closure of spina bifida may reduce the risk of handicap because the amniotic fluid in the third trimester is thought to be neurotoxic.

HYDROCEPHALUS AND VENTRICULOMEGALY

In hydrocephalus there is pathological increase in the size of the cerebral ventricles.

Prevalence

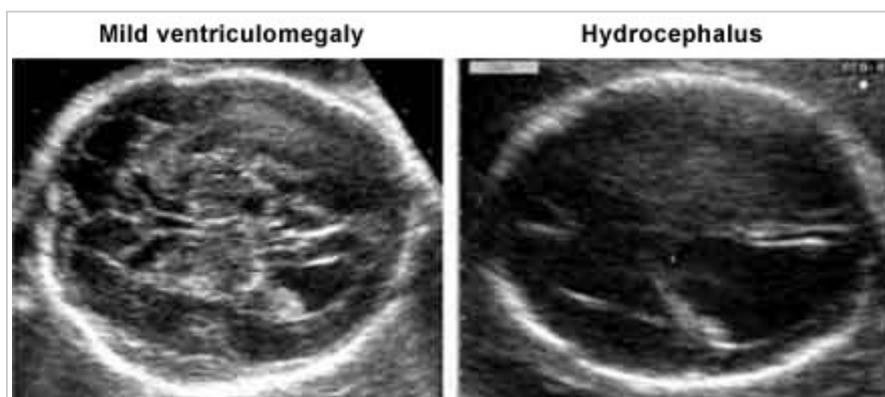
Hydrocephalus is found in about 2 per 1,000 births. Ventriculomegaly (lateral ventricle diameter of 10 mm or more) is found in 1% of pregnancies at the 18-23 week scan. Therefore the majority of fetuses with ventriculomegaly do not develop hydrocephalus.

Etiology

This may result from chromosomal and genetic abnormalities, intrauterine hemorrhage or congenital infection, although many cases have as yet no clear-cut etiology.

Diagnosis

Fetal hydrocephalus is diagnosed sonographically, by the demonstration of abnormally dilated lateral cerebral ventricles. Certainly before 24 weeks and particularly in cases of associated spina bifida, the head circumference may be small rather than large for gestation. A transverse scan of the fetal head at the level of the cavum septum pellucidum will demonstrate the dilated lateral ventricles, defined by a diameter of 10 mm or more. The choroid plexuses, which normally fill the lateral ventricles are surrounded by fluid. A distinction is usually made between mild, or borderline, ventriculomegaly (diameter of the posterior horn 10-15 mm) and overt ventriculomegaly or hydrocephalus (diameter greater than 15 mm).



Prognosis

Fetal or perinatal death and neurodevelopment in survivors are strongly related to the presence of other malformations and chromosomal defects. Although mild, also referred to as borderline, ventriculomegaly is generally associated with a good prognosis, affected fetuses form the group with the highest incidence of chromosomal abnormalities (often trisomy 21). In addition in a few cases with apparently isolated mild ventriculomegaly there may be an underlying cerebral maldevelopment (such as lissencephaly) or destructive lesion (such as periventricular leukomalacia). Recent evidence suggests that in about 10% of cases there is mild to moderate neurodevelopmental

delay.

Fetal therapy

There is some experimental evidence that *in utero* cerebrospinal fluid diversion may be beneficial. However, attempts in the 1980's to treat hydrocephalic fetuses by ventriculo-amniotic shunting have now been abandoned because of poor results mainly because of inappropriate selection of patients. It is possible that intrauterine drainage may be beneficial if all intra- and extra- cerebral malformations and chromosomal defects are excluded, and if serial ultrasound scans demonstrate progressive ventriculomegaly.

HOLOPROSENCEPHALY

This is a spectrum of cerebral abnormalities resulting from incomplete cleavage of the forebrain. There are three types according to the degree of forebrain cleavage. The alobar type, which is the most severe, is characterized by a monoventricular cavity and fusion of the thalami. In the semilobar type there is partial segmentation of the ventricles and cerebral hemispheres posteriorly with incomplete fusion of the thalami. In lobar holoprosencephaly there is normal separation of the ventricles and thalami but absence of the septum pellucidum. The first two types are often accompanied by microcephaly and facial abnormalities.

Prevalence

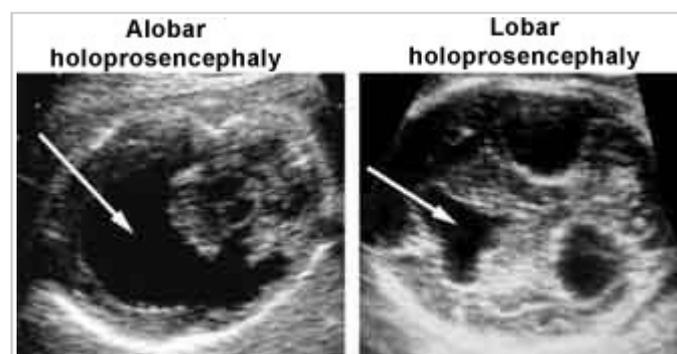
Holoprosencephaly is found in about 1 per 10,000 births.

Etiology

Although in many cases the cause is a chromosomal abnormality (usually trisomy 13) or a genetic disorder with an autosomal dominant or recessive mode of transmission, in many cases the etiology is unknown. For sporadic, non-chromosomal holoprosencephaly, the empirical recurrence risk is 6%.

Diagnosis

In the standard transverse view of the fetal head for measurement of the biparietal diameter there is a single dilated midline ventricle replacing the two lateral ventricles or partial segmentation of the ventricles. The alobar and semilobar types are often associated with facial defects, such as hypotelorism or cycloopia, facial cleft and nasal hypoplasia or proboscis



Prognosis

Alobar and semilobar holoprosencephaly are lethal. Lobar holoprosencephaly is associated with mental retardation.

AGENESIS OF THE CORPUS CALLOSUM

The corpus callosum is a bundle of fibers that connects the two cerebral hemispheres. It develops at 12-18 weeks of gestation. Agenesis of the corpus callosum may be either complete or partial (usually affecting the posterior part).

Prevalence

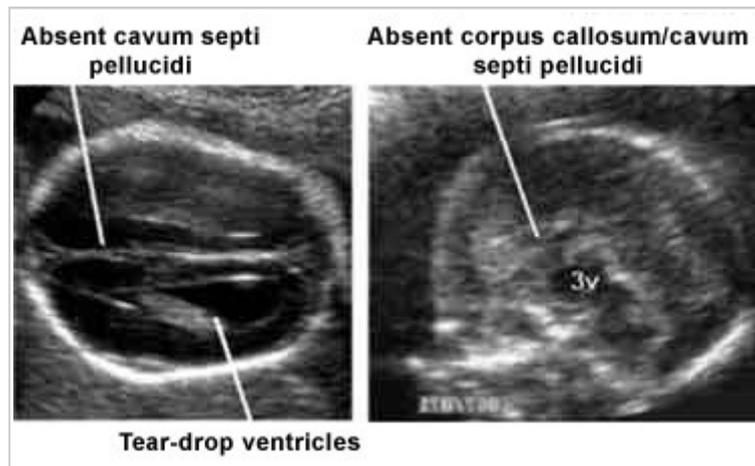
Agenesis of the corpus callosum is found in about 5 per 1,000 births.

Etiology

Agenesis of the corpus callosum may be due to maldevelopment or secondary to a destructive lesion. It is commonly associated with chromosomal abnormalities (usually trisomies 18, 13 and 8) and more than 100 genetic syndromes.

Diagnosis

The corpus callosum is not visible in the standard transverse views of the brain but agenesis of the corpus callosum may be suspected by the absence of the cavum septum pellucidum and the 'teardrop' configuration of the lateral ventricles (enlargement of the posterior horns). Agenesis of the corpus callosum is demonstrated in the mid-coronal and mid-sagittal views, which may require vaginal sonography.



Prognosis

This depends on the underlying cause. In about 90% of those with apparently isolated agenesis of the corpus callosum development is normal.

DANDY-WALKER COMPLEX

The Dandy-Walker complex refers to a spectrum of abnormalities of the cerebellar vermis, cystic dilation of the fourth ventricle and enlargement of the cisterna magna. The condition is classified into (a) Dandy-Walker malformation (complete or partial agenesis of the cerebellar vermis and enlarged posterior fossa), (b) Dandy-Walker variant (partial agenesis of the cerebellar vermis without enlargement of the posterior fossa), and (c) mega-cisterna magna (normal vermis and fourth ventricle).

Prevalence

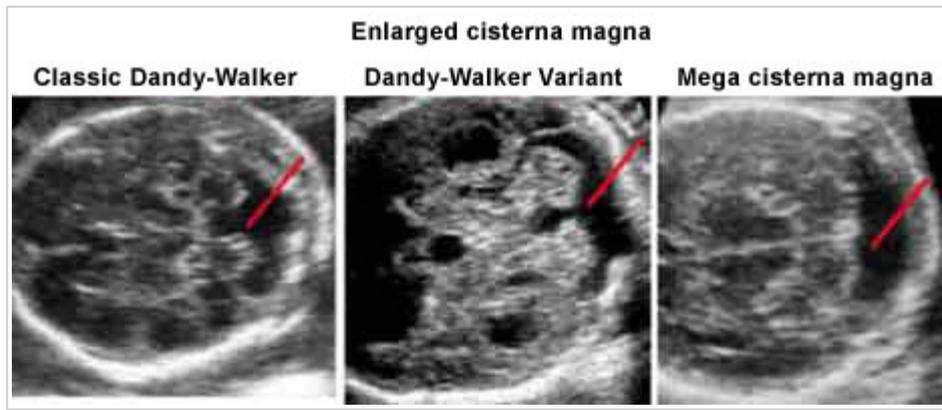
Dandy-Walker malformation is found in about 1 per 30,000 births.

Etiology

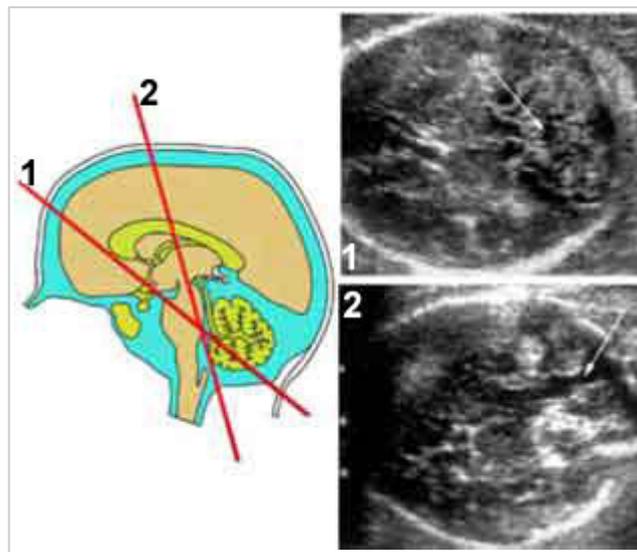
The Dandy-Walker complex is a non-specific end-point of chromosomal abnormalities (usually trisomy 18 or 13 and triploidy), more than 50 genetic syndromes, congenital infection or teratogens such as warfarin, but it can also be an isolated finding.

Diagnosis

Ultrasonographically, the contents of the posterior fossa are visualized through a transverse suboccipito-bregmatic section of the fetal head. In the Dandy-Walker malformation there is cystic dilatation of the fourth ventricle with partial or complete agenesis of the vermis; in more than 50% of the cases there is associated hydrocephalus and other extracranial defects. Enlarged cisterna magna is diagnosed if the vertical distance from the vermis to the inner border of the skull is more than 10 mm.



Prenatal diagnosis of isolated partial agenesis of the vermis is difficult and a false diagnosis can be made prior to 18 weeks gestation, when the formation of the vermis is incomplete and anytime in gestation if the angle of insonation is too steep.



Prognosis

Dandy-Walker malformation is associated with a high postnatal mortality (about 20%) and a high incidence (more than 50%) of impaired intellectual and neurological development. Experience with apparently isolated partial agenesis of the vermis or enlarged cisterna magna is limited and the prognosis for these conditions is uncertain.

MICROCEPHALY

Microcephaly means small head and brain.

Prevalence

Microcephaly is found in about 1 per 1,000 births.

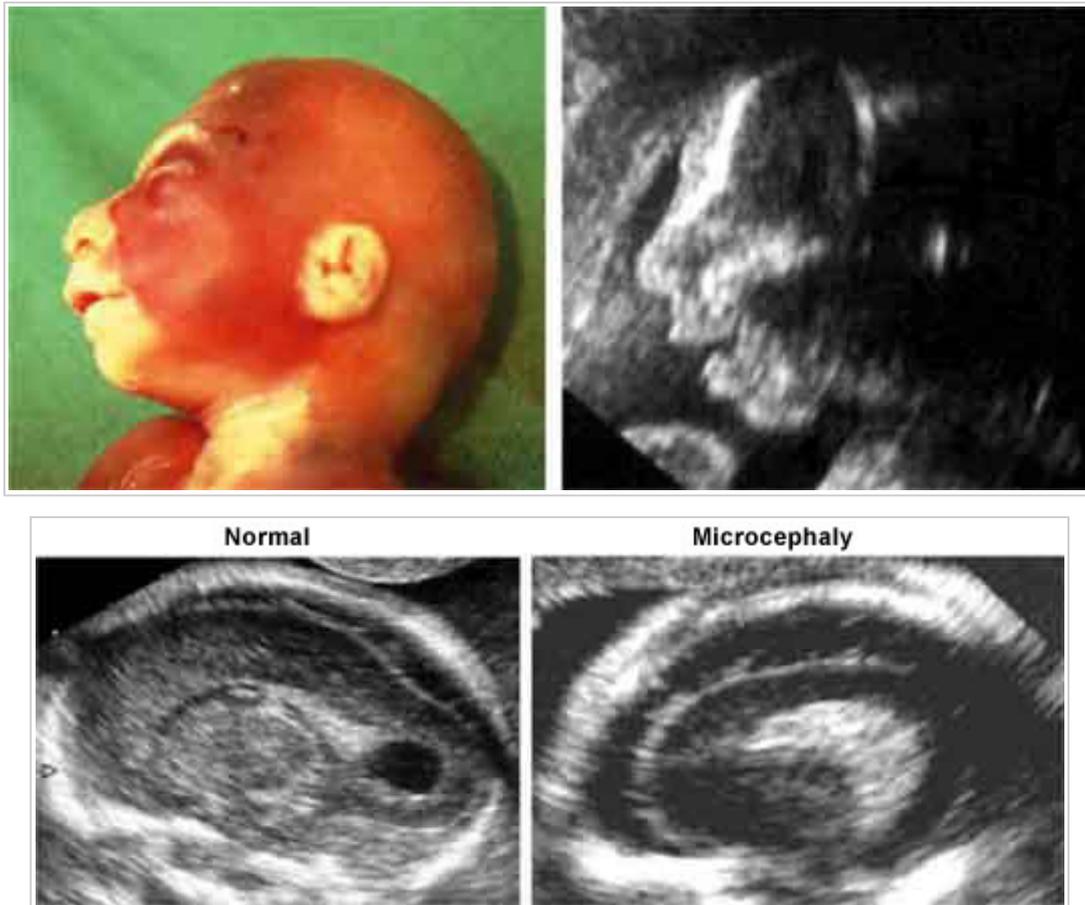
Etiology

This may result from chromosomal and genetic abnormalities, fetal hypoxia, congenital infection, and exposure to radiation or other teratogens, such as maternal anticoagulation with warfarin. It is commonly found in the presence of other brain abnormalities, such as encephalocele or holoprosencephaly.

Diagnosis

The diagnosis is made by the demonstration of brain abnormalities, such as holoprosencephaly. In cases with apparently isolated microcephaly it is necessary to demonstrate progressive decrease in the head to abdomen circumference ratio to below the 1st centile with advancing gestation. Such diagnosis may not be apparent before the

third trimester. In microcephaly there is a typical disproportion between the size of the skull and the face. The brain is small, with the cerebral hemispheres affected to a greater extent than the midbrain and posterior fossa.



Prognosis

This depends on the underlying cause, but in more than 50% of cases there is severe mental retardation.

MEGALENCEPHALY

Megalencephaly means large head and brain.

Prevalence

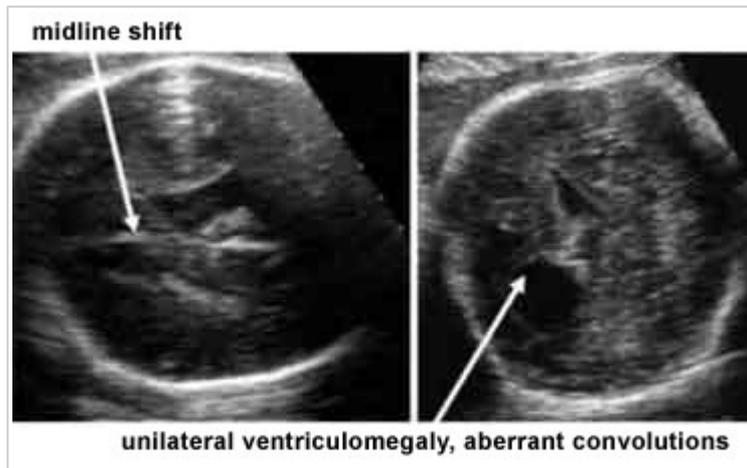
Megalencephaly is a very rare abnormality.

Etiology

This is usually familial with no adverse consequence. However, it may also be the consequence of genetic syndromes, such as Beckwith-Wiedemann syndrome, achondroplasia, neurofibromatosis, and tuberous sclerosis. Unilateral megalencephaly is a sporadic condition.

Diagnosis

The diagnosis is made by the demonstration of a head to abdomen circumference ratio above the 99th centile without evidence of hydrocephalus or intracranial masses. Unilateral megalencephaly is characterized by macrocrania, a shift in the midline echo, borderline enlargement of the lateral ventricle and atypical gyri of the affected hemisphere.



Prognosis

Isolated megalencephaly is usually an asymptomatic condition. Unilateral megalencephaly is associated with severe mental retardation and untreatable seizures.

DESTRUCTIVE CEREBRAL LESIONS

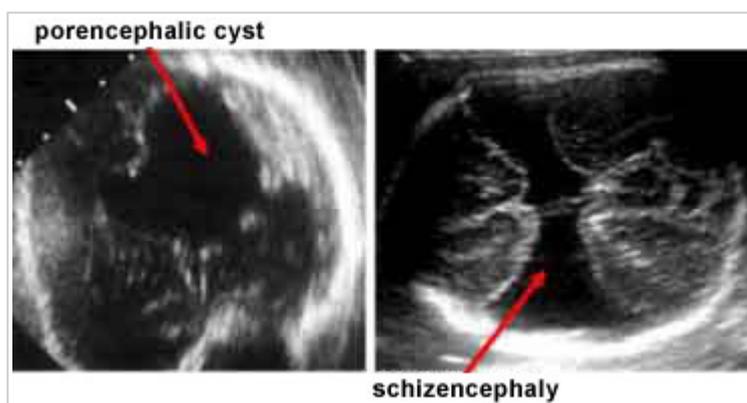
These lesions include hydranencephaly, porencephaly and schizencephaly. In hydranencephaly there is absence of the cerebral hemispheres with preservation of the mid-brain and cerebellum. In porencephaly there are cystic cavities within the brain that usually communicate with the ventricular system, the subarachnoid space or both. Schizencephaly is associated with clefts in the fetal brain connecting the lateral ventricles with the subarachnoid space.

Prevalence

Destructive cerebral lesions are found in about 1 per 10,000 births.

Etiology

Hydranencephaly is a sporadic abnormality that may result from widespread vascular occlusion in the distribution of the internal carotid arteries, prolonged severe hydrocephalus, or an overwhelming infection such as toxoplasmosis or cytomegalovirus. Porencephaly may be caused by infarction of the cerebral arteries or hemorrhage into the brain parenchyma. Schizencephaly may be a primary disorder of brain development or it may be due to bilateral occlusion of the middle cerebral arteries.



Diagnosis

Complete absence of echoes from the anterior and middle fossae distinguishes hydranencephaly from severe hydrocephalus in which a thin rim of remaining cortex and the midline echo can always be identified. In porencephaly there is one or more cystic areas in the cerebral cortex, which usually communicates with the ventricle; the differential diagnosis is from intracranial cysts (arachnoid, gliependymal), that are usually found either within the scissurae or in the mid-line and compress the brain. In schizencephaly there are bilateral clefts extending from the

lateral ventricles to the subarachnoid space, and is usually associated with absence of the cavum septum pellucidum.

Prognosis

Hydranencephaly is usually incompatible with survival beyond early infancy. The prognosis in porencephaly is related to the size and location of the lesion and although there is increased risk of impaired neurodevelopment in some cases development is normal. Schizencephaly is associated with severe neurodevelopmental delay and seizures.

ARACHNOID CYST

Arachnoid cysts are fluid-filled cysts contained within the arachnoid space

Prevalence

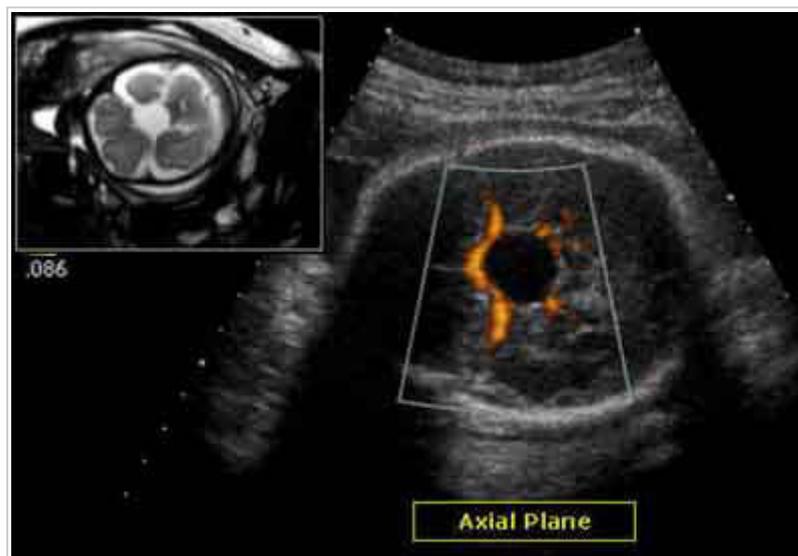
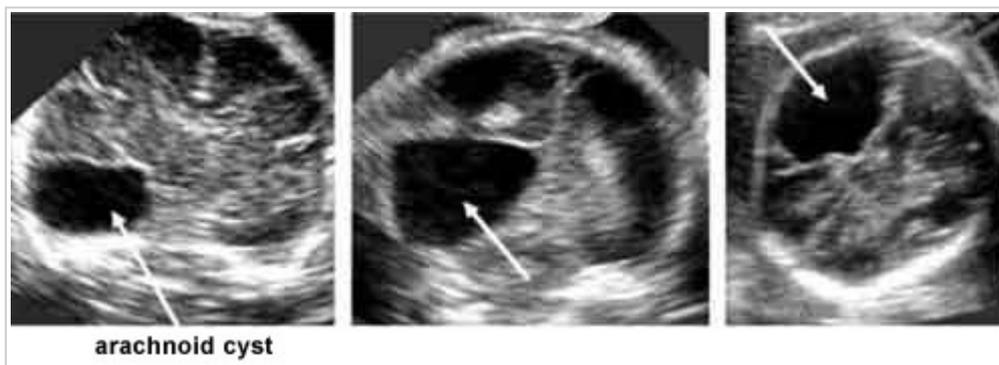
Arachnoid cysts are extremely rare

Etiology

Unknown; infectious process has been hypothesized but it is unlikely that this may explain the congenital cysts

Diagnosis

Arachnoid cysts appear on antenatal ultrasound as sonolucent lesions with a thin regular outline, that do not contain blood flow, do not communicate with the lateral ventricles and anyhow are not associated with loss of brain tissue. They occur most frequently in the area of the cerebral fissure and in the midline. Large cysts may cause significant mass effect and the distinction from porencephaly may be difficult. Interhemispheric cysts associated with agenesis of the corpus callosum most likely are not arachnoid cysts, but rather gliependymal cysts.



*Arachnoid cyst (axial Plane) and Circle of Willis
MRI showing the relation with the cerebral peduncles*

Prognosis

Large cysts may cause intracranial hypertension and require neurosurgical treatment. However, a normal intellectual development in the range of 80-90% is reported by most series. Spontaneous remission has been described both in the postnatal as well as in the antenatal period. Gliependymal cysts, that should be suspected in those cases with associated agenesis of the corpus callosum probably reflect a greater degree of derangement in the development of the brain and this may be reflected in a worse outcome.

CHOROID PLEXUS CYSTS

These cysts, which are usually bilateral, are in the choroid plexuses of the lateral cerebral ventricles.

Prevalence

Choroid plexus cysts are found in about 2% of fetuses at 20 weeks of gestation but in more than 90% of cases they resolve by 26 weeks.

Etiology

The choroid plexus is easily visualized from 10 weeks of gestation when it occupies almost the entire hemisphere. Thereafter and until 26 weeks, there is a rapid decrease in both the size of the choroid plexus and of the lateral cerebral ventricle in relation to the hemisphere. Choroid plexus cysts contain cerebrospinal fluid and cellular debris.

Diagnosis

The diagnosis is made by the presence of single or multiple cystic areas (greater than 2 mm in diameter) in one or both choroid plexuses.



Prognosis

They are usually of no pathological significance, but they are associated with an increased risk for trisomy 18 and possibly trisomy 21. In the absence of other markers of trisomy 18 the maternal age-related risk is increased by a factor of 1.5.

VEIN OF GALEN ANEURYSM

This is a mid-line aneurismal dilation of the vein of Galen due to an arteriovenous malformation with major hemodynamic disturbances.

Prevalence

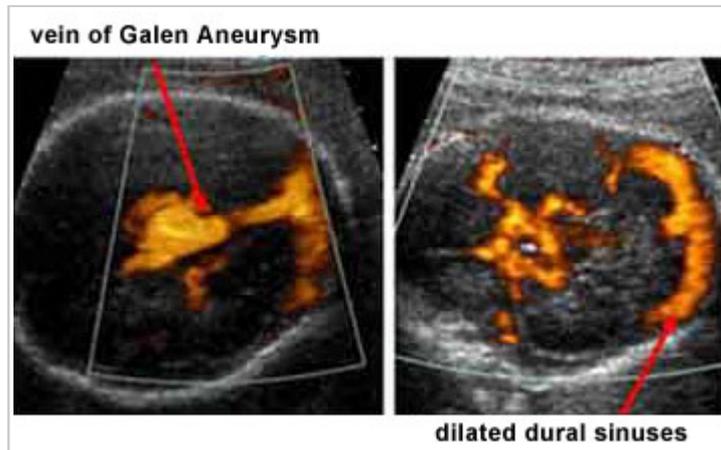
Vein of Galen aneurysm is a very rare abnormality.

Etiology

Vein of Galen aneurysm is a sporadic abnormality.

Diagnosis

The diagnosis is made by the demonstration of a supratentorial mid-line translucent elongated cyst. Color Doppler demonstrates active arteriovenous flow within the cyst. There may be associated evidence of high-output heart failure.



Prognosis

In the neonatal period about 50% of the infants present with heart failure and the rest are asymptomatic. In later life hydrocephalus and intracranial hemorrhage may develop. Good results can be achieved by catheterization and embolization of the malformation.