Central nervous system

- Many congenital malformations of the CNS result from incomplete closure of the neural tube.

Anencephaly

Central nervous system

- the most common neural tube defect
- Anencephaly - means absence of the brain
  - caused by failure of closure of the neural tube at the cranial end.
- recurrence risk 2% to 3% for woman with a history of a prior pregnancy with an open neural tube defect.

Anencephaly

- absence of the cranial vault
- complete or partial absence of the forebrain
  - may partially develop and then degenerate
- the presence of the brainstem, midbrain, skull base, and facial structures.

Anencephaly

- Anencephaly may result from
  - a syndrome such as Meckel-Gruber
  - a chromosomal abnormality such as trisomy
- increased risk in patients with diabetes mellitus
- teratogens associated with NTD
  - High levels of zinc
  - Methotrexate
  - folate and vitamin deficiencies

Anencephaly

- Anencephaly is a lethal disorder
  - up to 50% of cases resulting in fetal demise
  - The remainder die at birth or shortly thereafter.
Anencephaly
Central nervous system
• Anencephaly may be detected with ultrasound as early as 10 to 14 weeks

Anencephaly
Central nervous system
• Second trimester identification of anencephaly is more obvious
  – absent cerebral hemispheres evident as well as absence of the skull.

Anencephaly
Central nervous system
• Polyhydramnios
  – 40% to 50% of cases present after 26 weeks
• Additional anomalies include
  – cleft lip and palate
  – Hydronephrosis
  – diaphragmatic hernia
  – cardiac defects
  – Omphalocele
  – gastrointestinal defects
  – talipes.

Acrania
Central nervous system
• Lethal anomaly that manifests as absence of the cranial bones with the presence of complete development of the cerebral hemispheres.

Acrania
Central nervous system
• Begins in the fourth gestational week
• progresses to anencephaly
  – the brain slowly degenerates as a result of exposure to amniotic fluid.

Acrania
Central nervous system
• Presence of brain tissue without the presence of a calvarium
• Prominent sulcal markings
**Acrania**  
Central nervous system  
- associated anomalies  
  - spinal defects  
  - cleft lip and palate  
  - talipes, cardiac defects  
  - omphalocele  
- has been associated with amniotic band syndrome

**Cephalocele**  
Central nervous system  
- cephalocele  
  - a neural tube defect in which the meninges alone or meninges and brain herniate through a defect in the calvarium

**Cerebral Meningeal Agenesis**  
Central nervous system  
- Encephalocele  
  - term used to describe herniation of the meninges and brain through the defect  
- cranial meningocele  
  - describes the herniation of only meninges

**Cephalocele**  
Central nervous system  
- Cephaloceles involve the occipital bone and are located in the midline in 75% of cases  
- they also may involve the parietal and frontal regions

**Cephalocele**  
Central nervous system  
- The presence of brain in the defect, microcephaly, and other anomalies worsens the prognosis  
- isolated cranial meningocele may have a normal outcome

**Cephalocele**  
Central nervous system  
- Sonographic  
  - A bony defect in the skull  
  - Ventriculomegaly  
  - Polyhydramnios  
- Cephaloceles located off midline are usually the result of amniotic band syndrome
Cephalocele
Central nervous system
• Coexisting anomalies
  – Microcephaly
  – agenesis of the corpus callosum
  – facial clefts
  – spina bifida
  – cardiac anomalies
  – genital anomalies.

Cephalocele
Central nervous system
• Chromosomal anomalies and syndromes
  – trisomy 13
  – Meckel-Gruber syndrome, which is an autosomal-recessive disorder characterized by encephalocele
  – Polydactyly
  – polycystic kidneys.

Spina Bifida
Central nervous system
• wide range of vertebral defects that result from failure of neural tube closure.
• meninges and neural elements may protrude through this defect.
• may occur anywhere along the vertebral column
  – most commonly occurs along the lumbar and sacral regions.

Spina Bifida
Central nervous system
• the second most common open neural tube defect.
• When covered with skin or hair, it is referred to as spina bifida occulta
• If the defect is very large and severe, it is termed rachischisis. Ψ

Spina Bifida
Central nervous system
• When the defect involves only protrusion of the meninges, it is termed a meningocele.
• More commonly the meninges and neural elements protrude through the defect and are termed a meningomyelocele.

Spina Bifida
Central nervous system
• Fetuses with myelomeningoceles often present with the cranial defects associated with the Arnold-Chiari (type II) – 90%
  – presents with hydrocephalus
• the cerebellar vermis, which becomes displaced into the cervical canal.
  – giving it a “banana” appearance Ψ
Spina Bifida
Central nervous system

- caudal displacement of the cranial structures causes scalloping of the frontal bones of the skull

Spina Bifida
Central nervous system

- Sonographic features of spina bifida include
  - Splaying of the posterior ossification centers with a V or U configuration
  - Protrusion of a saclike structure that may be anechoic or contain neural elements
  - cleft in the skin

Spina Bifida
Central nervous system

- associated findings include
  - Talipes
  - Cephaloceles
  - cleft lip and palate
  - Hypotelorism
  - heart defects
  - genitourinary anomalies.

Dandy-Walker Malformation
Central nervous system

- agenesis or hypoplasia of the cerebellar vermis with resulting dilation on the fourth ventricle.
  - thought to occur before the sixth or seventh gestational week

Dandy-Walker Malformation
Central nervous system

- associated with other intracranial anomalies - 50% of the time.
  - agenesis of the corpus callosum
  - aqueductal stenosis
  - Microcephaly
  - Macrocephaly
  - Encephalocele
  - gyral malformations
  - lipomas.

Dandy-Walker Malformation
Central nervous system

- associated extracranial anomalies
  - Cardiac
  - Polydactyly
  - facial clefts
  - urinary tract

- associated Chromosomal anomalies
  - trisomies 13, 18, and 21.
**Dandy-Walker Malformation**

Central nervous system

- Imaging characteristics
  - posterior fossa cyst
  - Splaying of the cerebellar hemispheres
  - enlarged cisterna magna
  - Ventriculomegaly

**Holoprosencephaly**

Central nervous system

- a range of abnormalities resulting from abnormal cleavage of the prosencephalon (forebrain)
  - three forms of holoprosencephaly
    - alobar - most severe form
    - semilobar intermediate form
    - lobar and the mildest form.

**Holoprosencephaly**

Central nervous system

- Alobar is characterized by
  - a monoventricle
  - brain tissue that is small and may have a cup, ball, or pancake configuration
  - fusion of the thalamus
  - absence of the interhemispheric fissure, cavum septum pellucidum, corpus callosum, optic tracts, and olfactory bulbs.

**Holoprosencephaly**

Central nervous system

- Semilobar presents with
  - singular ventricular cavity with partial formation of the occipital horns
  - partial or complete fusion of the thalamus
  - a rudimentary falx and interhemispheric fissure
  - absent corpus callosum, cavum septum pellucidum, and olfactory bulbs.

**Holoprosencephaly**

Central nervous system

- Lobar holoprosencephaly
  - there is almost complete division of the ventricles with a corpus callosum that may be normal, hypoplastic, or absent
  - the cavum septum pellucidum will still be absent.

**Holoprosencephaly**

Central nervous system

Sonographic findings

- common C-shaped ventricle
  - may or may not be enlarged
- Fusion of the thalamus with absence of the third ventricle
- Absence of the interhemispheric fissure
- Absence of the corpus callosum
- Absence of the cavum septum pellucidum
**Holoprosencephaly**
Central nervous system

- associated with facial abnormalities
  - Cyclopia
  - Hypotelorism
  - absent nose
  - flattened nose with a single nostril
  - a proboscis.
  - median or bilateral clefting may be present

**Agenesis of the Corpus Callosum**
Central nervous system

- a fibrous tract that connects the cerebral hemispheres
- a range of complete to partial absence of the callosal fibers that cross the midline,

**Agenesis of the Corpus Callosum**
Central nervous system

- Associated chromosomal anomalies
  - trisomies 13, 18 and 8.
- Sonographic findings
  - Absence of the corpus callosum
  - Elevation and dilation of the third ventricle
  - Widely separated lateral ventricular frontal horns
  - Dilated occipital horns (colpocephaly)

**Aqueductal Stenosis**
Central nervous system

- results from an obstruction, atresia, or stenosis of the aqueduct of Sylvius causing ventriculomegaly
- Sonographic findings
  - lateral Ventricular enlargement
  - Third ventricular dilation

**Vein of Galen Aneurysm**
Central nervous system

- rare arteriovenous malformation
  - vein is enlarged and communicates with normal-appearing arteries.
- usually isolated anomaly, has been associated with
  - congenital heart defects
  - cystic hygromas
  - hydrops.

**Vein of Galen Aneurysm**
Central nervous system

- Sonographic findings
  - space that may be irregular in shape and is located midline and posterosuperior to the third ventricle
  - Turbulent flow with Doppler evaluation
Choroid Plexus Cyst
Central nervous system

- round or ovoid anechoic structures found within the choroid plexus
- common - identified in approximately 1% of antenatal ultrasound examinations
- not associated with other anomalies
  - often resolve by 22 to 26 weeks of gestation

Choroid Plexus Cyst
Central nervous system

- ranging in size from 0.3 to 2 cm
- Unilateral or bilateral cysts
- Solitary or multiple
- Unilocular or multilocular
- Enlargement of the ventricle with large cyst

Choroid Plexus Cyst
Central nervous system

- Choroid plexus cysts have been identified in association with aneuploidy, most commonly trisomies 18 and 21.
- sonographic survey for anomalies that might suggest aneuploidy should follow identification of a choroid plexus cyst

Choroid Plexus Cyst
Central nervous system

- survey of the heart, and a survey of the feet and hands to look for abnormal posturing and polydactyly.
- Amniocentesis for karyotyping may be offered

Porencephalic Cysts
Central nervous system

- Porencephalic cysts, also known as porencephaly
  - cysts filled with cerebrospinal fluid that communicate with the ventricular system or subarachnoid space.
- may result from
  - hemorrhage, infarction, delivery trauma, or inflammatory changes in the nervous system.

Porencephalic Cysts
Central nervous system

- brain parenchyma undergoes necrosis, brain tissue is resorbed, and a cystic lesion remains.

Sonographic findings
- cyst within the brain parenchyma without mass effect
- Communication of the cyst with the ventricle or subarachnoid space
- Reduction in size of the affected hemisphere
Hydranencephaly
Central nervous system
• destruction of the cerebral hemispheres by occlusion of the internal carotid arteries.
  – Brain parenchyma is destroyed and is replaced by cerebrospinal fluid

Hydranencephaly
Central nervous system
• may be associated with polyhydramnios
• etiology usually involves congenital infection or ischemia.
  – cytomegalovirus and toxoplasmosis

Hydranencephaly
Central nervous system
• Sonographic findings
  – Absence of normal brain tissue with almost complete replacement by cerebrospinal fluid
  – An absent or partially absent falx
  – Presence of the midbrain, basal ganglia, and cerebellum

Ventriculomegaly (Hydrocephalus)
Central nervous system
• Ventriculomegaly - dilation of the ventricles within the brain.
• Hydrocephalus occurs when ventriculomegaly is coupled with enlargement of the fetal head.
• Enlargement of the ventricles → obstruction of cerebrospinal fluid flow.

Ventriculomegaly (Hydrocephalus)
Central nervous system
• If the result of aqueductal stenosis it is referred to as noncommunicating hydrocephalus.

Ventriculomegaly (Hydrocephalus)
Central nervous system
• obstruction may be outside of the ventricular system, such as with an arachnoid cyst, and is referred to as communicating hydrocephalus.
Ventriculomegaly (Hydrocephalus)
Central nervous system
• when an obstruction occurs the ventricles dilate as the flow of cerebrospinal fluid is blocked.
• Enlarged ventricles exert pressure on the brain tissue
  – sometimes producing irreversible brain damage.

Ventriculomegaly (Hydrocephalus)
Central nervous system
• manifestation of a syndrome or chromosomal abnormality.
  – associated with trisomy 21, and identified in trisomies 13 and 18.

Ventriculomegaly (Hydrocephalus)
Central nervous system
• may be quantitated by measuring the ventricular atrium across the glomus of the choroid plexus.
  – considered dilated when its diameter exceeds 10 mm

Ventriculomegaly (Hydrocephalus)
Central nervous system
• Sonographic findings:
  – Lateral ventricular enlargement exceeding 10 mm
  – A “dangling choroid sign” gravity-dependent choroid plexus
  – Possible dilation of the third and fourth ventricles
  – Fetal head enlargement when the BPD and HC exceed those for the established gestational age

Ventriculomegaly (Hydrocephalus)
Central nervous system
• 80% of fetus with ventriculomegaly have associated anomalies
• surveyed for defects involving
  – the face, heart, kidneys, abdominal wall, thorax, and limbs.
• amniocentesis to rule out chromosomal anomalies
• laboratory tests to rule out congenital infections.

Microcephaly
Central nervous system
• abnormally small head that falls 2 standard deviations below the mean.
• occurs because the brain is reduced in size
**Microcephaly**
Central nervous system

- Teratogens linked with microcephaly
  - congenital infections (rubella, toxoplasmosis, cytomegalovirus)
  - maternal alcohol abuse
  - heroin addition
  - mercury poisoning
  - maternal phenylketonuria
  - Radiation
  - hypoxia.

**Microcephaly**
Central nervous system

Sonographic findings

- Measurements include
  - BPD
  - OFD
  - HC

- Ratios comparing the head to other parameters – helpful

- Serial measurements should be performed monthly