Cephalic Disorders

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Cephalic Disorders

• Abnormal development of the nervous system
• Neurological disorders that are present from birth (i.e. congenital)
• Genetic and environmental
  – Exposure to medication, infection, and radiation before birth
• Most caused early in development
Prevention, Treatment, and Prognosis

• Adding folic acid to a mother’s diet before and during pregnancy helps decrease the probability of a fetus developing neural tube defects
  – Women of child-bearing age should take 0.4 mg of folic acid daily

• Treatment is often times limited to symptomatic and supportive help

• Prognosis is dependent upon the disease; some people with Cephalic Disorders function almost normally, but most are either disabled or die soon after birth

• http://my.clevelandclinic.org/disorders/cephalic_disorders/hic_cephalic_disorders.aspx
Nervous System Development: 4 major processes

- Cell proliferation: creation of new cells
- Cell migration: movement of cells
- Cell differentiation: acquisition of characteristics specific to cell type
- Cell death

http://www.ehd.org/images/gasserbook/Gasser_Fig6-1gs.jpg
Anencephaly

• Defect of neural tube where the head of the neural tube does not close
• Happens between day 23 and 26 of pregnancy
• Results: missing major parts of the brain, skull, and scalp
• Infants born with no forebrain, and what is left of the brain tissue is exposed
Anencephaly

- These infants are usually blind, deaf, unconscious, and have no feeling of pain.
- 1000-2000 American children are born with Anencephaly per year.
- More females than males are affected, but there is no known cause.
Colpocephaly

- Enlargement of occipital horns (posterior part of lateral ventricles)
- This happens when white matter fails to thicken in the cerebrum
- Infants have microcephaly (small head) and mental retardation
- Other possible conditions include motor problems, muscle spasms, and seizures
Colpocephaly

• May be diagnosed late in pregnancy, but often misdiagnosed as hydrocephalus
• May be more accurately diagnosed after birth when other physical signs are present (see previous)
• Prognosis dependent upon the severity of the condition
Holoprosencephaly

- No development of prosencephalon (forebrain)
- Forebrain fails to divide (therefore, no bilateral hemispheres)
- Causes abnormal face and brain structure
- A spectrum of defects, from serious malformations that cause death, to facial defects (including cyclopia and median cleft lip)
- Seizures and mental retardation can occur
Holoprosencephaly

• 1/2 of the causes are related to chromosomal abnormalities
  – Can be associated with Patau’s syndrome (trisomy 13) and Edwards’ syndrome (trisomy 18)
• Increased risk in diabetic mothers
• Poor prognosis, many do not show mental progress

Hydranencephaly

- Absence of cerebral hemispheres, area replaced by sacs of CSF
- Usually, normal brainstem and cerebellum formation
- Infants can appear normal initially, but after a few weeks, the infant becomes irritable and has hypertonia (increased muscle tone)
- After a few months, seizures and hydrocephalus can develop

http://library.med.utah.edu/WebPath/PEDHTML/PED009.html
Hydranencephaly

- Infants may develop hydrocephalus
- An extreme form of porencephaly
- Can be caused by stroke, injury, infection, or traumatic disorders after the 12th week of pregnancy
- Most children die before age 1

Iniencephaly

• Neural tube defect; includes extreme retroflexion of the head (head bends backwards) and defects of the spine
• Infant is usually short with a very large head
• The skin of the face is connected to the chest, and the skin of the scalp is connected to the back; generally there is no neck
Iniencephaly

- Associated abnormalities include:
  - Anencephaly
  - Cephalocele (cranial contents protrude from skull)
  - Hydrocephalus
  - Cyclopia
  - No mandible (lower jaw)
  - Cleft lip and palate
- More common in females
- Poor prognosis (usually only a few hours of life)

27 weeks, dorsally protruding brain tissue

Lissencephaly

- Means “smooth brain”
- Microcephaly (small head) and lack of sulci in brain
- Caused by a problem in neuronal migration
- Symptoms include: unusual appearance of face, difficulty swallowing, psychomotor retardation
- Also, seizures and hydrocephalus
Lissencephaly

- Can be caused by viral infections of the mother of fetus during the first trimester, insufficient blood supply to brain in early pregnancy, or a genetic disorder (X-linked, and chromosome 17-linked)
- Associated with Miller-Dieker syndrome and Walker-Warburg syndrome
- Prognosis varies; some develop almost normally, some fail to develop beyond the 5-month level, and many die before 2 years
- Most common causes of death are respiratory problems

http://webspace.webring.com/people/dl/lfurlotte99/brain.gif
Megalencephaly

- Also called macrencephaly
- Abnormally large and usually malfunctioning brain
- Head can be large at birth or can become abnormally large in early years of life
- May be related to a problem in the regulation of cell proliferation and reproduction
- Symptoms include development delay, corticospinal dysfunction, seizures, and convulsive disorders
Megalencephaly

• Affect males more often than females

• Prognosis is dependent upon the underlying cause of the disorders

Fig 2. Hemimegalencephaly. Axial T2 FSE (fast spin echo) [A] shows an enlarged right cerebral hemisphere. 3D SPGR (spoiled gradient recalled echo) coronal reformat [B] also exhibits ipsilateral cortical thickening, with shallow sulci resembling pachygyria. Ventricular asymmetry is very conspicuous.

http://www.scielo.br/img/revistas/anp/v69n1/24f02.jpg
Microcephaly

- Characterized by a small circumference of the head
- Can be congenital or develop over the first years of life
- Face continues to grow, leading to a receding forehead
Microcephaly

- Body is underweight and dwarfed
- Delayed speech and motor development
- Hyperactivity and mental retardation are common

[Image link: http://childrenshospital.org/cfapps/research/data_admin/Site3078/Images/Microcephaly_bigger.jpg]
Porencephaly

- Cyst or cavity in a cerebral hemisphere
  - Remnants of destructive lesions
  - Or result of abnormal development
- Can occur before or after birth
- Diagnosis usually made before age 1
- Delayed growth and development, spastic paresis (slight/incomplete paralysis), hypotonia (decreased muscle tone), seizures, and macrocephaly or microcephaly
Porencephaly

• Poor or absent speech development
• Epilepsy
• Mental retardation
• Prognosis varies
Schizencephaly

- Abnormal slits in the cerebral hemispheres
- A form of porencephaly
- Those with bilateral clefts (on both sides) are usually developmentally delayed and have delayed speech and language skills
- Those with unilateral clefts can be weak in one side of the body with normal or near normal intelligence

http://neuropathology-web.org/chapter3/images3/3-4l.jpg
Schizencephaly

- Can have microcephaly, hemi- or quadripareisis (weakness or paralysis on one side/all four extremities), and hypotonia
- Probably a very early neurological disruption
- A genetic origin for one type of Schizencephaly, also, exposures to medications and toxins can cause Schizencephaly
- Sometimes associated with heterotopias (isolated islands of neurons) - failure of migration of neurons
Less Common Disorders

Acephaly

- Absence of head
- A parasitic twin attached to sibling by the neck
- Lacks a head and heart

Figure 3. Post-mortem anteroposterior direct X-ray. Fetal cranial bone structures are not visible.
Exencephaly

- Brain located outside of skull
- Usually an early stage of anencephaly
- During fetal growth, the neural tissue degenerates
- Usually fetus is not carried to term
- Image: 12 weeks

http://radiographics.rsna.org/content/24/1/157/F5.expansion.html
Macrocephaly

- A descriptive, not diagnostic term
- Larger head circumference than average
- Characteristic of a variety of disorders
- In half of the cases, mental development is normal
- Can be caused by an enlarged brain or hydrocephalus
- May be associated with dwarfism, neurofibromatosis, and tuberous sclerosis

http://images.radiopaedia.org/images/23711/9bfdef7fd712c732ce52af608c854d.jpg
Micrencephaly

- Characterized by a small brain
- May be caused by disturbance in nerve cell proliferation
- Associated with maternal alcoholism, diabetes, or rubella (German measles)
- Genetic factor in some cases

- Severe intellectual impairment common
- Motor function problems may not appear until later in life

http://upload.wikimedia.org/wikipedia/commons/c/c3/Microcefalia.jpg
Otocephaly

- Primary feature: agnathia - total or virtual absence of lower jaw
- Lethal due to airway function
- Can occur with Holoprosencephaly
Less Common Disorders: Craniostenoses

• Deformities in the skull due to premature fusion of cranial sutures
Brachycephaly

- Premature fusion of coronal suture
- Causes shortened front-back diameter of skull
Oxycephaly

- Premature closure of either:
  - Coronal suture and any other suture
  - All sutures
- Most severe of craniostenoses

Plagiocephaly

- Premature unilateral fusion of coronal or lambdoid (occipital and parietal bones) sutures
- Characterized by asymmetrical distortion of skull

http://www.hanger.com/orthotics/services/plagiocephaly/PublishingImages/Plagiocephaly_head%20deformity.jpg
Scaphocephaly

- Premature fusion of sagittal suture (joins together parietal bones)
- Most common craniostenosis
- Presentation: long, narrow head

Trigonocephaly

- Premature fusion of metopic suture (joins two halves of frontal bone)
- Presentation: triangular prominence of forehead and eyes that are close-set