OB Case Study:
Anencephaly

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WARNING

GRAPHIC CONTENT INSIDE
Neural Tube Formation

- Neural plate appears during 3rd week
- Neural plate gives rise to neural folds
- Neural tube closes by 4th week
  - Cephalic end
  - Caudal end
Neural Tube Formation

- Failure of the neural folds to fuse can lead to NTDs:
  - Anencephaly
  - Spina bifida
  - Encephalocele
Neural Tube Formation

Click here if video did not play
What is Anencephaly?

- Failure of closure of one of the anterior neuropores
- Most severe neural tube defect
- Most common NTD
What is Anencephaly?

- An- = without
- Encephal/o = brain
- Meroencephaly
- Mero- = partial
Characteristics of Anencephaly

- Partial absence of the brain
  - Cerebrum
  - Cerebellum
  - Basal ganglia
- Absence of cranial vault
- Only brainstem left
Characteristics of Anencephaly

- Occurs superior to the orbits
- Facial features are preserved
- Base of skull remains
- Incompatible with life
At least 1 to 2 in 1000 births in US

Highest incidence in United Kingdom (3.5 per 1000)
Occurrence

- 4 times more common in females (males tend to have lower spinal defects)
- 6 times more prevalent in whites
- Recurrence is 3% to 5%
Theory

Failure of cranial end of neural tube to close by 4th week
Theory

Failure of cranial end of neural tube to close by 4th week
Theory

Failure of cranial end of neural tube to close by 4th week = NTD
Theory

Failure of cranial end of neural tube to close by 4th week = NTD
Theory

Failure of cranial end of neural tube to close by 4th week

= NTD

Abnormal development of calvaria
Theory

- Failure of cranial end of neural tube to close by 4th week
- Abnormal development of calvaria

NTD
Theory

Failure of cranial end of neural tube to close by 4th week = NTD

Abnormal development of calvaria = ACRANIA
Theory

Failure of cranial end of neural tube to close by 4th week = NTD

Abnormal development of calvaria = ACRANIA
Theory

Failure of cranial end of neural tube to close by 4th week

Abnormal development of calvaria

Protrusion of the brain from the cranium

NTD

ACRANIA
Theory

Failure of cranial end of neural tube to close by 4th week = NTD

Abnormal development of calvaria = ACRANIA

Protrusion of the brain from the cranium
Theory

Failure of cranial end of neural tube to close by 4th week = NTD

Abnormal development of calvaria = ACRANIA

Protrusion of the brain from the cranium = EXENCEPHALY
Theory

Failure of cranial end of neural tube to close by 4th week = NTD

Abnormal development of calvaria = ACRANIA

Protrusion of the brain from the cranium = EXENCEPHALY
Theory

Failure of cranial end of neural tube to close by 4th week = NTD

Abnormal development of calvaria = ACRANIA

Protrusion of the brain from the cranium = EXENCEPHALY

Without the protection of the skull, the brain is exposed and undergoes degeneration
Failure of cranial end of neural tube to close by 4th week

Abnormal development of calvaria

Protrusion of the brain from the cranium

Without the protection of the skull, the brain is exposed and undergoes degeneration

NTD

ACRANIA

EXENCEPHALY
Theory

Failure of cranial end of neural tube to close by 4th week

Abnormal development of calvaria

Protrusion of the brain from the cranium

Without the protection of the skull, the brain is exposed and undergoes degeneration

NTD

ACRANIA

EXENCEPHALY

ANENCEPHALY
Acrania

- Also known as exencephaly
- Occurs at the beginning of 4th GA week
- Skull mineralization begins at 9 weeks
- Skull bones fail to mineralize
- Normal bone should be echogenic

www.ultrasound-images.com
Acrania

- Can be diagnosed at 12 weeks
- Mickey Mouse head (bilobed brain appearance)
- Believed to be a predecessor of anencephaly
- True acrania is absence of entire skull (including skull base)
Anencephaly: What is Left?

- A mass that is:
  - Spongy
  - Vascular
  - Mostly composed of hindbrain
  - Rudimentary brain stem
  - Functioning neural tissue

www.neuropathology.neucom.edu
Angiomatous Stroma / Cerebrovasculosa

- Thick membrane
- Covers remnants of brain

Anencephaly. All that is left is a small, vascular mass of disorganized neural tissue (cerebrovasculosa) mixed with choroid plexus.

www.neuropathology.neucom.edu
Associated Findings

- Polyhydramnios (no control of swallowing)
- Spina bifida
- Craniorachischisis (congenital fissure of the skull and vertebral column)
- Cleft lip and palate

http://www.jle.com/en/revues/medecine/mtg/e-docs/00/04/40/1D/article.md?fichier=images.htm
Associated Findings

- Hydronephrosis
- Diaphragmatic hernia
- Cardiac defects
- Omphalocele

Intestine protruding through hole in diaphragm

www.nlm.nih.gov
Associated Findings

- GI defects
- Talipes
- Dysraphism (widening of upper cervical spine)
- Iniencephaly (retroflexed fetal head due to lack of cervical vertebrae)
Lab Values: AFP

- Elevated maternal serum alpha-fetoprotein (MSAFP)
  - Synthesized in yolk sac and later in fetal liver
  - Detected in fetal spine, GI tract, liver, and kidneys
  - Peaks between 15 and 18th GA week
Lab Values: AFP

- Elevated maternal serum alpha-fetoprotein (MSAFP)
  - Cutoff is 2.0 or 2.5 MoM (multiples of the median)
  - Detection rate for anencephaly is 95% or higher
  - Levels with anencephaly are >5.0 MoM
Lab Values: Acetylcholinesterase

- Exposed neural tissue in amniotic fluid
- Found also in maternal blood
Lab Values: Other Tests

- Amniocentesis
- Triple screen
- Karyotype testing
Sonographic Findings

- Embryo’s head identified by US at 7 weeks
- Normal amounts of brain can be seen in 1st trimester (even with anencephaly)
- Cranial vault does not ossify until after 1st trimester
Sonographic Findings

- Normal cranial anatomy can be visualized after 12 to 14 weeks GA
- CRL may be normal early in GA
- Absence of brain and cranial vault
- Absence of parietal and frontal bones
Sonographic Findings

- Folding of the ears
- Cerebrovasculosa
- Froglike appearance (bulging orbits) due to absent frontal bone
Patient History

- 25-year old
- Female
- G1
- 24w 3d Gestational Age
- No known history of genetic disease
  - Patient refused sequential screening
- No known complications with this pregnancy
- No prenatal or ultrasound history during early gestation due to socioeconomic reasons
Case Images: Ultrasound
Case Images: Ultrasound
Case Images: Ultrasound

Depth = 8.41 cm
Case Images: Ultrasound
Case Images: Ultrasound

![Ultrasound Image]

- D1 = 1.67 cm
- D2 = 2.36 cm
- Circum = 6.36 cm
- Area = 3.08 cm²
Case Images: Ultrasound
Case Images: Ultrasound
Case Images: Ultrasound

![Ultrasound Image]

Depth = 17.41 cm

10:18:42 am
6C2-S
5.5 MHz
150 mm
OB New
Compound Soft
75 dB SC1/+1/2/4
Gain = 2 dB Δ=2
Store in progress
Case Images: Ultrasound
Causes

- Multifactorial (genetic + environmental)
- Amniotic band syndrome?
  - Amputations
  - Entanglement
- Meckel-Gruber syndrome?
- Trisomy 13?
Risks

- Diabetes mellitus
- Hyperthermia
- Valproic acid (an antiepileptic drug)
- Folate and vitamin deficiencies

www.diabetesinsider.com
Risks

- Teratogens:
  - Zinc
  - Valproic acid
  - Methotrexate
  - Amnionopterin
Prognosis

- Fetal demise occurs in up to 50% of cases
- Death is imminent after birth
Prevention / Planning

- Genetic counseling
- Vitamin therapy
- Folic acid
  - 1 to 2 months prior to conception
  - During 1st trimester

http://www2.massgeneral.org/livingwithtsc/care/counseling.htm
1. Familial exencephaly - anencephaly sequence and translocation.
2. Neonate with meroacrania: radiological findings and review of the literature.
3. Anencephaly: pitfalls in pregnancy outcome and relevance of the prenatal exam.
Main Points:

- “The incidence of anencephaly and all neural tube defects is decreasing, mainly because of folic acid supplementation.”
- “Typically we talk about exencephaly or acrania as being, that there is some dysmorphic brain tissue, whereas anencephaly there’s no brain tissue above the brainstem.”
- “30 to 50-fold increase of the likelihood of having another baby with a neural tube defect.”

(D. Shaver, personal communication, November 19, 2010)
References

Which gender and race has a higher risk for anencephaly?

A. Males, Hispanics
B. Females, Whites
C. Females, Blacks
D. Males, Asians