Overview
• Large subject – focus on early detection in PCP office
• Head size: causes, interventions, followup, case study
  • Microcephaly
  • Macrocephaly
    • Benign extra axial fluid
    • Hydrocephalus
    • Lesions
    • Non-accidental trauma
• Head shape: causes, interventions, followup, case study
• Sacral dimples: causes, examinations, case study

Abnormalities in Head Size

Microcephaly
• Primary: identified at birth
• Secondary: presents later from a postnatal insult
• The brain is very small, usually 3 standard deviations below the mean.
• Affected patient often have mental retardation but otherwise have unremarkable findings on neurologic exam.
• Many have dysmorphic features: sloping forehead and prominent ears

Macrocephaly
• Defined as a head circumference greater than 2 SDs above the mean or greater than the 98th percentile.
• Bright Futures recommendations state that head circumference measurements should be obtained at each health supervision visit from birth to 24 months of age.
Abnormalities in Head Size

- Macrocephaly
  - Physical Exam: Should focus on the calvaria: signs of increased intracranial pressure such as a large tense fontanelle, splayed sutures or impaired upward gaze with “sunsetting” eyes.

- Accurate measurement of head. Compare to previous measurements
- Inspection and palpation of skull
- Presence of other dysmorphic features
- Neurologic and developmental assessments
- Signs and symptoms of increased intracranial pressure.

Benign Extraaxial Fluid

- Many children with macrocephaly have an accumulation of extracranial fluid
- Cause of the fluid is not clearly understood.
- Head circumference grows to more than the 95th% and then subsequently generally parallels the growth curve.
- Patient is otherwise stable, no developmental delays or focal findings on exam.

Case Study

- Otherwise healthy infant born at 36 weeks. Discharged from hospital with mother
- Patient making normal developmental progress
- Noted at the 6 month well child check to have climbed to the 50th percentile. Patient brought back 1 month later and found to have increased again
- Mother states that Dad has a “big head”
- Referral made to Neurosurgery.
- CT scan obtained
Benign Extraaxial Fluid Case Study

- Repeat imaging 3 months later

Benign Extraaxial Fluid

- If OFC starts follow curve, extra axial fluid improves, normal developmental patient.
- If patient showing concerns for significant developmental delays, psychomotor regression and continuing increase head circumference
  - Megalencephaly should be considered
  - Referral to neurology/neurodevelopmental
  - Full MRI will be recommended at this time.

Hydrocephalus

- Incidence:
  As an isolated congenital disorder, occurs 0.5-1.5 per 1000 live births
- Pathophysiology:
  - Production of CSF: 60% of CSF produced in the choroid plexus, with the rest being produced by in the ependyma of the cerebral ventricles, the aqueduct of Sylvius and the subarachnoid space
  - Rate of production in children is 0.33ml/kg/hour
  - Cause: Obstructed flow of CSF, impaired absorption or overproduction.

Hydrocephalus Case Study

- 9 month old infant present to NSR.
- Found to have increasing OFC; 50th% at 6mo visit; 95th % at 9 mo visit.
- Fussy infant; not developing as fast as her twin however is still meeting milestones in a delayed fashion.

Hydrocephalus Case Study

- Shunt placement: The shunt diverts the flow of CSF from the ventricles into the region of the body
  - Mostly to the abdomen
  - Also can be into right atrium of heart or pleural.

Treatment
Hardware

- Once patient has a shunt is a life long medical device
- Shunt malfunction considered to be a medical emergency
- Neurosurgery sees patient once a year; imaging on off years
- Good to have a baseline exam
- Fevers more concerning in the first 3 months of shunt placement.
  - Prolonged fever of unknown origin should consider shunt at that time

Hydrocephalus Treatment

- Endoscopic Third Ventriculostomy
  - Alternate operation to treat hydrocephalus
  - Surgery entails making a tiny hole in the floor of the third ventricles to allow free flow of CSF
  - Hole is made with an endoscope.
  - Not everyone is candidate: depends on etiology, location of the basilar artery and prior history

Hydrocephalus Treatment

- Will leave a reservoir in place.
- Innovative treatment however many surgeons are still uncertain in practice
- Combined often with choroid plexus cauterization (CPC) for full benefit.

Hydrocephalus Case Study

- Patient underwent placement of a ventricular-peritoneal shunt with a programmable valve
- Patient is developing slower than twin which has been hard for parents.
- Involvement of early intervention services
- Patient has undergone 3 shunt workups in the last year (usually more frequent in the first year with a non-verbal baby)

Brain Tumor: Epidemiology

- Estimated 4,000 children under age 20 are diagnosed annually with a primary benign or malignant brain tumors.
- Of there 2875 will be less than 15 years of age, and 1125 between the ages of 15-19.
- Slightly higher incidence in boys (4.61 per 100,000) than girls (4.55 per 100,000)

Statistics published by the American Brain Tumor Association (ABTA) in 2009

Average Annual Age-Adjusted Incidence Rates† of Primary Brain and CNS Tumors by Age and Behavior, CBTRUS Statistical Report: NPCR and SEER data from 2006-2010.

Average Annual Age-Adjusted Incidence Rates† of Primary Brain and CNS Tumors by Year and Behavior, CBTRUS Statistical Report: NPCR and SEER data from 2006-2010.

Brain Tumor Case Study
- 8 mo female presents to PMD office.
- Patient more fussy; tugging at ears last 2 weeks more.
- Parents have noted head to be increased in size; concerned that patient appears to be more top heavy.
- Otherwise normal self; interactive, saying "mama" "dada", normal eye movements, smiling.

Past Medical & Surgical History:
- No previous hospitalizations, chronic medical problems, or surgeries. Normal pregnancy and delivery.

Objective:
Pulse 200 (crying) I Temp (Src) 99.1 °F (37.3 °C) I Wt 21 lb (9.526 kg) I HC 48.3cm (19.02")

General: Alert, cooperative, in no apparent distress, appears stated age

Skin: Normal color, no rashes.

Head: Anterior fontanelle full, but not bulging. Head large for age.

Eyes: Sclerae white, No discharge.

Neuro: Alert, occasionally she had a faint smile. She seemed to have mild difficulty supporting her head when upright manifested by a slight wobbliness of her head.

Brain Tumor Case Study
Patient taken to operating room where lesion was excised
Patient continue to need ventricular drainage postoperatively (hydrocephalus) resulting in a ventricular peritoneal shunt placement
Patient found to have a CHOROID PLEXUS PAPILLOMA. Benign lesion treated with surgery.
Will be followed with frequent MRI’s for now.
Brain Tumor

- Brain tumors are followed closely by Oncology/Neurosurgery/Neurology/Rehab in the brain tumor clinic
- Patient will be followed closely by the Primary Care Provider.
- Neurodeficits depend on where the tumor is located.
- Establish a new neuro baseline. Does the patient now have a shunt? When is next followup? Any concerns related to possible shunt/tumor/treatment side effects call Brain tumor team
- Lots of FEAR

Signs and symptoms of a brain tumor/subsequent deficits

- Depends on location of tumor and associated hydrocephalus
- Mass effect of tumor itself and surrounding edema

Non-accidental Trauma

- Macrocephaly often is a symptom of abuse
- Need to do a head CT to rule out blood

Non-accidental Trauma

Important to note increased rate in first year
- Parent education on taking break/crying babies

Distribution of fatal AHT/fatal TBI cases for children under 1 year old, by month of age, USA, 2003–2007.


AHT: Abusive Head Trauma

Cause and morphologic features of SDB in 50 infants’ autopsies.


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**Non-accidental Trauma Case Study**

- 5mo old male presents to the ER after suffering a seizure
- Previously healthy full term infant was noted at 4mo WCC to have a jump in head circumference. Referral made for macrocephaly. Upcoming appointment in 2 weeks
- Parents report 1 prior episode a week ago when he appeared to have some seizure-like activity. Patient taken to Emergency Room where by the time he arrived, he appeared normal except for macrocephaly. Sent home.
- Mother is unable to securely date when she noted his head was large, but thinks it has been from about 3 months ago. He had missed his 2-month immunizations.

**Non-accidental Trauma Case Study**

- Patient required emergent drainage of subdural hematomas and seizure management
- Represented 2 months later with full bulging fontanel and increased head circumference. Rebled into area and required a second drainage of subdurations
- Unable to wean drain – required placement of subdural to peritoneal shunt.

**Other Causes of Macrocephaly**

- Megalencephaly
  - The mucopolysaccharidoses, Canavan disease and Alexander’s disease are examples of metabolic conditions
  - Disproportionate large number of children who have autism have enlarged head circumferences. More research being done on “brain overgrowth” in early years.

**Abnormal Head Shape**

- PCP are often the first providers to evaluate the patient’s head shape.
- Often an abnormal head shape is noted in the first hours of life.
- Most parents will note, “my babies head just doesn’t look right”
- Heads look bad at birth however should normalize by 3 weeks of age.
- Important to catch craniosynostosis before 6months of age so it can be corrected.

**Craniosynostosis: Metopic**

- 10% incidence
- Triangular shape head
- Bitemporal narrowing
- Parietal bossing
- Metopic ridge

Craniosynostosis: Unicoronal
- 20-30% incidence
- Anterior plagiocephaly
- Nasional deviation
- Flattening of the frontal bone on affected side

Craniosynostosis: Sagittal
- 40-60% incidence
- Bitemporal narrowing
- Frontal bossing
- Occipital cupping
- Palpable sagittal ridge

Craniosynostosis: Lambdoid
- 1-2% incidence
- Trapezoid shape
- Tilted skull base
- Occipitomastoid Bulge

Positional Skull Deformities
- Examine the head from the top of the head down.
- View the position of the ears
- Note the position of the cheekbones (maxilla)

Treatment:
- Prevention
- Physical therapy to rule out torticollis and encourage other positioning
- If there is progression or lack of improvement referral to Craniofacial clinic should be considered at around 4-6 months of age.
- Skull-molding helmets (work best 4-12 months)
- More than half infants improve by 6 months of age just with positioning.
- Reposition and neck exercises can take 2-3 months to improve.

Sacral Dimple
Sacral Dimple

- Neural tube defects are among the most common birth defects. Can be open or close.
- Open: myelomeningocele
- Closed: skin covered lesions under which the nervous tissue is not formed normally:
  - split cord malformation
  - dermal sinus tract
  - tethered spinal cord
  - Intraspinal lipoma

Sacral Dimple

- More than 50% of occult spinal dysraphisms (OSD) are diagnosed with a dimple
- Otoscopic examination of the dimple to determine bottom of pit
- Careful physical examination: attention to neurology and orthopedic concerns
  - Most are found midline overlying the spine
    - Any finding of abnormal hair growth, hemangioma, subcutaneous mass (Lipoma) or caudal appendage
    - A deviated or duplicated gluteal cleft

Sacral Dimple

- Concerning
  - Multiple dimples
  - Dimple greater than 5mm
  - Location greater than 2.5cm above the anal verge
- Benign
  - Solitary sacrococcygeal pits within the gluteal cleft

A prototypical benign sacral dimple that is located within the gluteal cleft (less than 2.5cm above the anus) and solitary

Sacral Dimple

- Next steps:
  - 1) monitor 2) refer to a specialist 3) order a scan
- Imaging: US vs. MRI is facility dependent.
- Prefer MRI as the gold standard
- Chern et al (2012) found majority of infants referred for lumbar US because of cutaneous finding did not have OSD.
  - 5% will have an abnormal US results; 1% will have surgical intervention
  - Abnormal US finding have poor sensitivity and good specificity. Abnormal results need to be followed by a MRI
Sacral Dimple: Case Study

- Patient is a 1-month old infant who is referred for evaluation of a deep sacral dimple, and a discoloration of the skin over the lumbar area, and a small skin appendage in the perineum
- Patient had all normal in utero scans. Born full term via vaginal deliver

- PHYSICAL EXAMINATION:
  - The lesion on the perineum is approximately 1.5 mm in diameter and raised about 1 mm above the surface of the skin, and at this time is not discolored. A small area at the posterior aspect at the left labia
  - The second abnormality is a deep intergluteal dimple approximately 2.5 cm from the anus. When wiped with a Q:
    - swab, one can see the bottom.
  - Hemangioma over the lumbar area, well above the gluteal cleft, that is approximately now 3 x 2 cm with an irregular border.

- Surgery is prophylactic.
- Therefore can be performed at a safe age (6-9 months of age)
- Will wait to perform MRI when infant is older for anesthesia (greater than 3 months of age)

- Patient had MRI performed at 3.5 months of age

- At 9 months of age patient taken to the operating room

- OPERATION:
  - 1. Resection of lumbar spinal intradural tract.
  - 2. Detethering of filum.
  - 3. Operative microdissection.

- Patient recovered well from surgery
- Patient with normal movement/sensation following surgery
- Patient slow to potty train at 3.5 years of age
- Followed annual in NSR/NDV clinics
  - At risk for cord retethering
    - Worsening bowel bladder symptoms
    - Weakness, sensation changes
    - Gait imbalances.

Conclusion

- Available to discuss specific patients/symptoms via the provider contact line 206 987 7777
- Our website does contain information on the team and information on conditions treated
Questions?
Thank you.