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LUMPS, TUFTS AND DIMPLES
IT’S THE PITS!!
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Embryology of the Nervous System:
Gastrulation

- Primitive streak elongates from caudal to cranial end (POD 13-16)
- Prospective mesoderm ingresses through primitive groove
- Remaining epiblast cells spread out, replace ingressing mesoderm cells

- Hensen’s node at cranial end of the primitive streak
- Prospective notochordal cells ingress through node and become notochord
- Primitive streak caudally, becomes caudal cell mass
Embryology of the Nervous System: Primary Neurulation

- POD 17-27
- Neural groove forms as median hinge point
- Neural folds develop
- Neural folds fuse
- Separation of neuro- and cutaneous ectoderm (dysjunction)

Embryology of the Nervous System: Primary Neurulation

- Cranial neuropores last points to close
- Caudal neuropore at level of S2
- Cranial neuropore at lamina terminalis, just above optic chiasm
- Additional occipital neuropore
- Neuropores most common sites of dermal sinus tracts
Embryology of Nervous System: Secondary Neurulation

- Begins on POD 27
- Involves spinal cord caudal to S2 and filum terminale
- Formed from caudal cell mass
- Occurs in the presence of an intact cutaneous ectoderm
- Species specific

Embryology of the Nervous System: Ascent of Conus Medullaris

- POD 42-54: Retrogressive differentiation
- Caudal neural tube thinner
  - No mantle zone
  - Rudimentary marginal zone
  - Looks "less well developed"
- POD 54 and beyond: Differential growth of spinal cord and vertebral column

Embryology of the Nervous System: Ascent of Conus Medullaris

- Most ascent pre-natally
- Conus at or above L2-3 disc space at birth
- Conus at or above L1-2 disc space by 2 months post-natal
Level of Normal Conus Medullaris

- 100 children with brain tumors having screening whole spine imaging
- Level of conus medullaris measured
- Mode: L1-2 disc space
- Mean: Inferior third of L1
- Lower border (95% confidence limits): Middle third L2
- Below inferior third of L2 considered radiographically tethered

Kesler, Dias, Kalapos: Neurosurg Focus 2007

Innocent Coccygeal Dimples

- Coccygeal - within the gluteal cleft
- Tip of coccyx palpable within few millimeters
- Normal gluteal cleft
- Shallow, non-complex dimple
- No tufts of hair, hemangiomata, skin appendages or skin tags
- No neurological, urological, or orthopedic abnormalities
- Remnant of tail bud/Hensen’s node
Innocent Coccygeal Dimples

- Prevalence up to 4% of population
- Not related to pilonidal cysts in adults
- No convincing evidence for relationship with congenital spinal cord malformations
  - Only 7 cases with SC abnormalities in literature
    - 5/7 cases had other cutaneous markers
    - 2 not clear whether dimple was within gluteal cleft
  - Powell: 2000 cases, 1 with tract to outer dura
  - Herman – 0/53, Gibson – 0/75 had abnormalities
  - Weprin and Oakes: 1000 cases, no evidence of tethering or deterioration (imaging not done)
Spinal Dermal Sinus Tracts (DST)

- Incidence 1 in 2500 births
  - 1% cervical
  - 10% thoracic
  - 41% lumbar
  - 35% lumbosacral
- May be associated with other congenital spinal cord malformations
  - Split cord malformations, lipoma, thickened filum terminale, endodermal or dermoid cyst

Spinal Dermal Sinus Tracts (DST)

- Lumbosacral (above gluteal crease)
- Abnormal gluteal cleft
- Deep or complex
- Tufts of hair, hemangiomata, appendages or skin tags
- Neurological, urological, or orthopedic abnormalities
Embryology of DST

- Failure of neuroectoderm to separate from cutaneous ectoderm during dysjunction
  - Tongue of cutaneous ectoderm remains attached to neural tube
  - Persistence of tract variable
    - Some tracts involve subcutaneous tissues only, most remain attached to spinal cord
- Most common at posterior neuropore
  - Tract therefore ends along dorsum of spinal cord at level of S2, separate from filum terminale
Pathophysiology of Deterioration in Spinal DST

- Spinal cord tethering
- Bacterial meningitis (portal of entry)
- Aseptic meningitis (desquamation of epithelial debris within CSF)
- Mass effect from expanding dermoid inclusion cyst
Imaging of Spinal DST

- Spinal Ultrasound
  - Good screening tool, but not sensitive
  - Probably not worthwhile after 6 months of age
- Magnetic resonance Imaging
  - Definitive neuroimaging study
  - More sensitive than sonography
  - Missed 2/3 of tracts in one study
  - Conus may not be abnormally low
- Need for operation depends upon clinical appearance and location of dimple!!!!

Clinical Presentation of Spinal DST

- Focal neurological deficits in 2/3 at initial neurosurgical evaluation
  - Motor weakness 39%
  - Sensory changes 25%
  - Gait changes 18%
  - Sphincter disturbances 21%
  - Bowel and bladder changes 14%
- Age < 1 yr: 50% with neurological deficits
- Age > 1 yr: 92% with neurological deficits
- Infection in 3/28 (1 with meningitis)
Management of Spinal DST

- Linear vertical incision, ellipse around tract
- Follow tract to defect in lumbodorsal fascia
- Laminectomy adjacent to tract
- Identify penetration of dura
- Open dura cranial and caudal to track, ellips dural opening around tract
- Follow tract to conus medullaris
- Look for separate filum terminale
Meningocele Manque

- Likely related embryologically to DST
- Scarified, ‘cigarette paper’, ‘cigarette burn’ skin lesion
- May be tender to touch
- No CSF leakage
- Fibrous tract or atretic peripheral nerves, dorsal root ganglion cells
- 50% associated with split cord malformations
Cranial Dermal Sinus Tracts

- Less common than spinal DST
- Occipital, retro-auricular, nasofrontal location
- Modes of deterioration
  - Intracranial suppurative infection
    - Epidural abscess, subdural empyema, recurrent bouts of meningitis, brain abscess
  - Growing intracranial dermoid or epidermoid masses

Occipital DST

- Cutaneous pit or tract usually located near occipital external protuberance
- Skull X-rays and CT/or scans demonstrate bone defect at site of cutaneous lesion
- MRI better demonstrates soft tissue tract
- Invariable intracranial extension
- Associated hairy nevus (2/8 cases) or subcutaneous mass (4 of 8 cases)
Frontonasal DST

- Dimple or dermoid extending anywhere from nasal tip to glabella
- Innocuous looking
- Travel between nasal bone and nasal cartilage toward anterior skull base
- 90% end extra-cranially
- 10% extend through foramen cecum, anterior to crista galli, and end intracranially
  - Extradural, intra-falx, subarachnoid space, lamina terminalis
Embryology of Cranial DST

- Failure of dysjunction at cranial neuropores
  - Occipital midline: occipital lobes and/or cerebellum
  - Lamina terminalis

A. Dura
   Frontal Bone
   Fonticulus
   Nasofrontalis
   Nasal Bone
   Prenasal Space
   Nasal Cartilage

B. Foramen
   Cecum
   Dura
   in Contact
   with Skin
Surgical Management of Frontonasal DST

- If no intracranial extension on neuroimaging, excise superficial tract, follow toward skull base
- If tract ends extracranially, no need for further exploration
- If intracranial extension found, plan intracranial exposure at same or later operation
- If obvious intracranial extension, operation planned jointly with plastic and neurosurgery
Flammeus Nevus and Spinal Lipomas

- Flat, not raised, port wine in color
- Similar in appearance to ‘stork bite’
- Irregular outline, blanches with pressure
- May be associated with other cutaneous markers of spinal dysraphism
  - Dermal sinus tract, subcutaneous masses
  - Meningocele manque
  - Fawn's tail
  - Hairy nevus

Tubbs, JNS:Pediatrics, 2004

Management of Flammeus Nevus

- Incidence of underlying spinal cord malformations is unknown
  - 21 (17%) of 120 patients with occult spinal dysraphism in one series had isolated flammeus nevus as the only manifestation (N=21)
    - Lipoma 3
    - Fatty filum terminale 5
    - Dermal sinus tract with lipoma 2
    - Syringomyelia 8
    - Meningocele manque 1
    - Split cord malformation 2
  - No specific malformation associated with flammeus nevus

Tubbs, JNS:Pediatrics, 2004
Hypertrichosis (Fawn’s Tail)

- Most commonly associated with, and specific for, split cord malformations (SCM)
  - Diastematomyelia, diplomyelia
- Present in 11/14 (79%) of patients with SCM
- Associated with other cutaneous markers
  - Flammeus nevus in 3/14 (21%)
  - Dysplastic skin in 2/14 (14%)
  - Subcutaneous mass in 2 (14%)
Human Tails and Pseudotails

- **True tail**: contains both cutaneous and bony structures (stiff), remnant of human tail
- **Pseudotail**: lacks bony structures (soft)
- Differentiation arbitrary and confusing, not helpful since BOTH are associated with congenital dysraphic malformations
  - Congenital spinal anomalies in 50%
  - Spinal cord tethering in 25%
- MRI for further evaluation
- Surgical exploration, excision and spinal cord untethering

Lu, Pediatr Neurol 1998
Atretic Parietal Encephaloceles

- Small, usually elevated lesions with dysplastic skin over dome
- Whorling pattern of surrounding hair
- No CSF leakage
- Bisects sagittal sinus
- Underlying MRI abnormalities common, MRV demonstrates persistent embryonic prosencephalic vein
- Neurological/cognitive abnormalities in 40%
Cutis Aplasia Congenita

- Rare, most often involves midline scalp
- May involve scalp, scalp and bone or scalp, bone and dura
- Overlies superior sagittal sinus
  - Significant hemorrhage possible
- Sometimes associated with other malformations
  - Cerebral malformations, TEF, frontonasal dysplasia, congenital heart defects, facial palsy, mental retardation, cleft-lip/palate
  - Adams Oliver syndrome (CAC + limb defects)

Cutis Aplasia Congenita: Management

- Early identification of lesion
- Do NOT allow to dry out. Keep moist!!!!
  - Cover with wet gauze and plastic wrap
  - Wet to wet dressings
  - Cover with petroleum based antibiotic ointment
- Two surgical management strategies
  - Non-operative: wet to wet dressing changes
  - Operative: primary closure or scalp rotation flaps
Conclusions

• Innocent coccygeal dimples are located within the gluteal cleft
  • No associated cutaneous lesions
  • Depth of pit not important
  • No need for further workup, imaging, or surgery

• Pathological lumbosacral DST are *more cranially located*, outside of gluteal cleft
  • Imaging important for surgical planning but of no importance for determining need for surgery
  • Might as well go directly to MRI rather than US
  • Prophylactic surgical exploration necessary

Conclusions

• Cranial DST
  • Recognize importance of seemingly innocent midline dimple or bump anywhere on nose
  • Suspicion in cases of intracranial suppurative infections in expected locations, or recurrent bouts of unexplained meningitis
  • Neuroimaging may include both CT and MRI
  • Do not inject dye into any tract
  • Combined surgical approach with experienced pediatric neurosurgical and plastic/craniofacial surgical expertise

Conclusions

• Flammeus nevus is associated with various forms of spinal dysraphism (incidence unknown)
• Hypertrichosis (fawn’s tail) most often associated with, and specific for, SCM
• Human tails associated with spinal cord malformations in 50%, tethering in 25%
Conclusions

- Atretic encephaloceles look benign, associated with underlying brain malformations
  - Whorls of hair, skin covered
- Cutis aplasia congenita similar but without whorls of hair, not skin covered, sometimes has underlying skull and dural defect
  - Hemorrhage from SSS, keep it moist!
  - Surgical and non-surgical management proposed

Midline cutaneous anomalies other than coccygeal dimples should be considered to have associated underlying CNS malformations and should be referred to a specialist for evaluation and treatment