MID-LINE ANOMALIES: WHEN TO WORRY?

F019 Lumps & Bumps in Children

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DISCLOSURE OF RELATIONSHIPS WITH INDUSTRY

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DISCLOSURES

I have no relevant disclosures.
Primary Investigator for epidermolysis bullosa studies
Learning Objectives

• Identify the most common mid-line lesions seen in pediatric patients
• Recognize potential underlying anomalies and properly order additional testing
Lumps & Bumps

- Developmental
  - Aplasia cutis congenita
  - Encephalocoele/atretic
  - Meningocoele/atretic
  - Dermoid cyst/sinus
  - Umbilical anomalies
  - Other developmental cysts/sinuses/retained structures

- Neoplasms
  - Benign (congenital nevi, epidermal nevi)
  - Non-benign

- Vascular
  - Malformations (VM, AVM, LM)
  - Neoplasms (Hemangioma etc.)
Lumps and Bumps: Acquired Neoplasms

- Juvenile xanthogranuloma
- Mastocytoma
- Spitz nevus
- Keloid
- Acquired nevi
- Pyogenic granuloma
- Angiofibroma
- Granuloma annulare
- Pilomatrixoma
- Non-benign

Courtesy of Ingrid Polcari, MD
Aplasia cutis
Encephaloceles
Meningoceles
Dermoid
Spinal dysraphism

Branchial/
Thyroglossal/
Bronchial cysts

Omphalomesenteric duct remnant
Vitelline duct remnant
Aplasia Cutis

- Congenital absence of epidermis, dermis and sometimes subcutaneous tissues
- Extension to dura or meninges is possible
- Scalp > extremities, trunk
  - usually sporadic
  - solitary (70%) or multiple
  - Usually in close proximity to hair whorl
- Often confused with forceps or scalp electrode injury
- Ulceration, erosion or scar
- Membranous vs non-membranous ACC

- Clinical diagnosis
- Treatment usually unnecessary

Browning JC. Dermatol Therapy. Volume 26, Issue 6, Article first published online: 9 DEC 2013
ACC: Cause for worry?

- More than 2 findings
  - ACC plus another lump or bump
- Hair collar sign
  - With membranous ACC: form fruste of neural tube defect?
  - Encephaloceles, meningoceles, heterotopic brain tissue
  - MRI to rule out underlying scalp defect or intracranial connection
- Other defects present: consider a syndrome
  - *Adams-Oliver syndrome*: ACC, transverse limb defects, cardiac, CNS abnormalities
  - *Trisomy 13, 4p-syndrome, oculocerebrocutaneous syndrome*

Aplasia cutis

- Good history and physical exam
- Associated findings?
- Consider imaging or clinical follow-up
- Refer if concerning features
Aplasia cutis
Encephalocoeles
Meningocele
Dermoid
Spinal dysraphism
Omphalomesenteric duct remnant
Vitelline duct remnant
Branchial/
Thyroglossal/
Bronchial cysts
Dermoid sinus (or cyst)

- Epithelium lined tract (or cyst), may contain adnexal structures (hair follicles, sebaceous glands)
- Protruding tuft of hair pathognomonic
- Can be difficult to visualize if covered by hair
- Thickening of the scalp, hypertrichosis, dimpling
- Sinuses are portals for infection that can lead to abscesses, osteomyelitis, meningitis (*staph aureus*)
Dermoid cyst

- Cysts most commonly on orbital ridge
- 3% midline (45% with CNS connection)
- Progressive enlargement with resulting bony defects
Imaging of midline lesions

- **MRI: image of choice** for sinuses/midline lesions.
  - Review of 103 nasal dermoids: correct result in 87% of the cases, no false positives and 13 (12%) false negatives.

- **CT helpful** (can aid in bony anatomy)

- Surgical and radiological findings were concordant in 90 (87%) cases. (4 cases intracranial extension was not noted but was evident intraoperatively)

BEJ Hartley et al. Nasal dermoids in children: A proposal for a new classification based on 103 cases at Great Ormond Street Hospital, 2015-01-01Z, Volume 79, Issue 1, Pages 18-22,
Cephaloceles

- Cranial meningoceles: meninges, CSF
- Encephaloceles: brain tissue
  - Occipital most common
  - Anterior: broad nasal root
  - May have associated cutaneous signs

- Flesh, colored or bluish hue
- Enlarge with increased intracranial pressure (crying, Valsalva)
Embryology

- Midline fusion of ectodermal and neuroectodermal tissue occurs at cephalic and caudal ends of the neural tube: look in occipital and lumbosacral regions

Atretic Meningocele

- Congenital ectopic rests of meningeal tissue
- Occipital/parietal scalp
- Flesh colored, glistening or bullous appearance
- Associated PWS or hair collar sign
- May have skull defect or fibrous tract, but no CNS connection (so no enlargement with increased ICP)
- MRI to differentiate from meningocoele

- Differential: membranous aplasia cutis
- Treatment: excision by neurosurgeon
Completely Intraosseous Atretic Meningocele
Davidson L., Gonzalez-Gomez I. · McComb J.G.

Fig. 1
Coronal $T_2$-weighted (a) and sagittal $T_2$-weighted (b) unenhanced MR images of the patient. The intraosseous lesion is hyperintense on $T_2$ (arrow) and hypointense on $T_1$ (arrow). The overlying scalp and subjacent brain are normal in appearance.

Pediatr Neurosurg 2009;45:308–310
Atretic encephaloceles

- Ectopic rests of neuroglial tissue
- No intracranial connection, so no enlargement with ICP
- May have skull defect or fibrous stalk
- Previously known as nasal gliomas
  - Scalp, oronasopharynx, palate, orbit
- Firm, red-blue noncompressible nodules
  - May have overlying telangiectasia

- MRI to differentiate from encephalocele
- Treatment: excision by neurosurgeon
Associated signs? Symptoms?
Size change with increased ICP?
Imaging?
Referral
Aplasia cutis

Encephaloceles/
meningocoeles

Dermoid

Spinal
dysraphism

Omphalomesenteric
duct remnant
Vitelline duct
remnant

Branchial/
Thyroglossal/
Bronchial cysts
Developmental tags/cysts

- Accessory Tragus:
  - Preauricular most common
    - Hearing exam
  - Less common at lateral commissure of the mouth
  - Line of fusion of first branchial arch
- Cartilaginous rest of the neck
  - Remnants of branchial arches

- Management: excision
  - May contain cartilage
Midline anterior neck inclusion cyst: A novel superficial congenital developmental anomaly of the neck

Rabina Walsh MD | Jeffrey North MD | Kelly M. Cordoro MD | Ana Isabel Rodriguez Bandera MD | Leonard Kristal MD | Ilona J. Frieden MD

FIGURE 2 Representative pathology demonstrates a dermal cyst lined by stratified squamous epithelium with maturational through a granular layer and containing orthokeratotic keratin. Hematoxylin and eosin, (A) 20x and (B) 40x.

FIGURE 1 Each patient presented with a cystlike yellow-white papule on the midline lower anterior neck.
Median raphe cysts

- Aka: Congenital sinus and cysts of the genitoperineal raphe, mucous cysts of the penile skin, parameatal cysts
- Consequence of incomplete fusion of ventral aspect of the urethral or genital folds, likely resulting in ‘tissue trapping’
- Asymptomatic unless super-infection occurs
- Lined by pseudostratified epithelium, except at distal penis, where is stratified
- Recent review suggests 4 types: urethral, epidermoid, glandular, and mixed
- Treatment: observation, removal if symptomatic

Branchial cleft cyst

• Cyst contains laminated keratin
• Sinus tract lined by squamous epithelium with or without cilia or goblet cells
• Cyst often surrounded by lymphoid follicles
Branchial cleft cyst/sinus

Thyroglossal duct cyst

- Midline, non-tender, moves with swallowing
- Thyroidhyoid (60%) >> submental, suprasternal, intralingual
- Differential: ectopic thyroid, dermoid, sebaceous cyst, lymphadenitis, goiter, lipoma
- Reasons for excision: infection, sinus formation, malignancy, cosmesis
- Diagnosis: ultrasound and +/- radioisotope scan
- Refer to otolaryngology for removal

Bronchogenic cyst

- Most common on the neck, esp suprasternal notch
- Cyst contains keratin or mucin
- Lining: varies from pseudostratified columnar (w or wo goblet cells or cilia) to squamous epithelium
- Lining may be surrounded by mucous glands, smooth muscle, lymphoid follicles or cartilage

**FIGURE 4** Location of midline anterior neck inclusion cyst (MANIC) compared with that of other congenital anomalies of the midline anterior neck. red = cutaneous bronchogenic cysts, green = thyroglossal duct cysts, orange = midline cervical clefts, blue = MANIC
Aplasia cutis
Encephaloceles/
meningocoeles
Dermoid

Branchial/
Thyroglossal/
Bronchial cysts

Spinal
dysraphism

Omphalomesenteric
duct remnant
Vitelline duct
remnant
A Neonate with an Unusual Midline Defect and Cardiovascular Anomaly

Martin Poryo, Paul Hoffmann, Hans-Joachim Schäfers, Clemens-Magnus Meier, Katrin Altmeyer, Hashim Abdul-Khalil, Michael Zemlin, Sascha Meyer

Fig. 1 Presentation of the strand with extension from the xiphoid process to the umbilical cord.

Omphalomesenteric duct remnant

• Ectopic gastric, small intestinal or colonic intestinal mucosa present within eroded periumbilical skin
• Diffuse lymphocytes in the stroma
• *consider connecting fistula or concurrent intestinal malformation
Umbilical lesions

• Differential includes:
  • Patent urachus
  • Omphalomesenteric duct / remnant (aka umbilical polyp)
  • Persistent Vitelline duct /remnant
  • Umbilical granuloma
When to worry?

- > 2 different congenital midline lumbosacral lesions
  - lipomas
  - dermal sinuses
  - tails
  - aplasia cutis
  - dermoid sinus/cyst
  - hemangioma >/2.5 cm

- A lesser risk of OSD:
  - hemangioma <2.5 cm
  - atypical dimple
  - hypertrichosis

- Lower risk of OSD
  - hyperpigmentation/hypopigmentation
  - melanocytic nevi
  - simple dimple
  - port wine stain
  - teratomas

Imaging for lumbosacral lesions

- Of the 180 patients evaluated with radiography, U/S and MRI 50 patients (28%) had spinal dysraphism (with 64 cutaneous stigmata)
- < 6 mos old: ultrasonographic examination in cases of flat cutaneous stigmata it missed only 5% of cases
- With bulky overlying masses (lipoma, hemangioma) u/s missed 15% of cases
- MRI recommended as imaging modality of choice

Segmental infantile hemangiomas
When to Worry?
Beard distribution-
Check the Airway!
Perineal Hemangiomas-
SACRAL, PELVIS, LUMBAR Syndromes

SACRAL: Spinal dysraphism, Anogenital anomalies, Cutaneous abnormalities, Renal and urologic anomalies, Angioma of Lumbosacral localization


LUMBAR: Lower body IH, Urogenital anomalies (and ulceration), Myelopathy, Bony, Anorectal and arterial, Renal
Midline infantile hemangiomas

- Infants with midline lumbosacral infantile hemangiomas are at increased risk of spinal anomalies
- Ulceration and additional cutaneous anomalies associated with an increased risk
- MRI recommended (ultrasound sensitivity 50%)
  

- Tethered cord, spinal lipoma, intraspinal hemangiomas in >50% of cases. Sinus tract was found in 40%.

Take-Home Points

• If a lump or bump is midline, you need to consider underlying anomalies or connections
• Midline pits/sinuses need further work-up
• >2 cutaneous signs if higher risk of underlying anomaly
• Consider mode of imaging (MRI/ultrasound)
• Refer if in doubt!