More Than Meets the Eye?

A checklist approach to cutaneous markers of congenital malformations

Renee Howard, MD
Professor of Dermatology, UCSF
Director of Dermatology
UCSF Benioff Children’s Hospital Oakland
DISCLOSURE OF RELATIONSHIPS WITH INDUSTRY

Renee Howard MD

F043 - Is it Only Skin Deep? A "Checklist" Approach to Diagnosing and Managing Birthmarks and Neonatal Skin Diseases

DISCLOSURES

I have no relevant relationships with industry
Objectives

- Outline differential diagnosis of skin signs of developmental anomalies at four “hotspots”: nose/brow, scalp, lower back, neck
- Checklists for diagnosis and treatment
Checklist

- What?
  - High risk or low risk?
    - Vascular stain, pigmentary change, pit, aplasia cutis, nodule, SC mass?
How? Management

- Active nonintervention
- Imaging
- Referral
  - Neurosurgery
  - ENT
  - Plastic Surgery
  - Pediatric Surgery
Where?

Developmental hotspots = Embryological fusion lines

From Kos and Drolet. Neonatal Dermatology 2nd Edition
10 month-old with subtle lump nasal bridge, slowing increasing in size
Nose and Brow
Nasal Midline

- Dermoid cyst/sinus, glioma, encephalocele
- High risk CNS extension
- Common embryologic origin
Dermoid Cyst

- Rare 1:20,000
- Most common midline congenital anomaly
- Mean age at presentation 29 mos
Dermoid Cyst

- Entrapment of surface ectoderm
- Midline or over suture lines higher risk
- Can rupture, become inflamed or infected, erode bone
- Persist

Nasal Dermal Sinus Tract (DST)

- Congenital midline pit
- Tuft of hair, oily or clear discharge (CSF!)
- 10-30% extend deep +/- cyst anywhere along the tract
- Complications infectious or aseptic meningitis
58 superficial
38 intraosseous
10/102 cases
intracranial
Nasal Dermoid Cyst, DST

- **Who:** healthy newborn or toddler
- **What:** pit, nodule
- **When:** sinus congenital, dermoid
  - first few years of life
- **Where:** glabella to nasal tip midline
- **How**
  - Imaging-MRI with contrast for soft tissue, CT for bony defects prn
  - Refer to: ENT + Neurosurgeon
Lateral Brow

Most common location of Dermoid Cysts
Lateral Brow Dermoid Cyst

- **Who:** healthy infant or toddler
- **What:** slowly growing nodule
- **When:** first few years of life
- **Where:** lateral 1/3 eyebrow
- **How**
  - No imaging needed as no intracranial extension
  - Active nonintervention
  - Refer to: ENT or plastic surgeon
Imagine you are attending...

- There are two patients in two rooms
- One requires quick reassurance--low risk
- The other needs imaging and/or surgical referral--high risk
- You’re THE DECIDER!
Imagine you are attending...

- **Low risk: active nonintervention**
  - Off midline
  - Soft slow growing
  - Mobile

- **High risk: imaging/refer**
  - Midline membranous aplasia cutis congenita
  - Hair collar
Which scalp lesion is high risk?
Which scalp lesion is high risk?

- High risk: imaging/refer
  - Midline membranous aplasia cutis congenita
  - Hair collar

- Low risk: active nonintervention
  - Scar like
  - No other skin or hair changes
Scalp Aplasia Cutis Congenita (ACC)

- 86% of ACC located on scalp
- Most sporadic though can be syndromic
- Presents at birth as erosion or scar like area with alopecia
- Hair collar
Scalp aplasia cutis congenita (ACC)

- New classification based on clinical appearance
  - Membranous-developmental
  - Nonmembranous-destruction
- Most scalp ACC membranous
- 15-20% have underlying bony defect
Scalp ACC: When to Image

- Midline
- Membranous
- Hair collar
- Vascular stain
Which scalp nodule is higher risk?
Hemangioma vs. Dermoid Cyst

Natural history, overlying skin changes
Ultrasound if unsure
Active nonintervention
Young man with hx of lump and alopecia since early childhood
“Tip of the Iceberg” phenomenon
Scalp Dermal Sinus Tracts (DST)

- Located parietooccipital
- With or without associated cyst
- Intracranial extension through midline occipital skull defect into dura

Scalp Dermoid Cyst

- 2nd most common location
- Presents < 4 years, 40% congenital
- Usually asymptomatic, firm, fixed to underlying bone
- Slowly enlarge then stabilize

Scalp Dermoid Cyst

- Ten year retrospective by neurosurgeons
- 46% eroded bone
- Higher rate in patients older at time of surgery
- They conclude: refer for excision right away

Scalp: ACC, Pit, Nodule

- **High risk**
  - Present at birth
  - Midline, especially parieto-occipital
  - Nodule, membranous ACC, pit
  - Multiple signs
    - Hair collar
    - Vascular stain

- **Low risk**
  - Later onset
  - Off midline
  - Soft mobile nodule, scar-like ACC
  - Other signs
    - Hyperpigmentation

8 mo with 2 skin lesions
Hotspot: Lumbosacral Spine

- Pits
- Aplasia cutis
- Nodules and subcutaneous masses
- Vascular stains
- Segmental hemangiomas
Why is a dermatologist talking about PRIMARY NEURULATION?
Consequences of Undiagnosed LS anomalies

- Infection
- Aseptic meningitis
- Spinal cord compression
- Tethered cord

Atretic meningocele.

Mark Dias et al. Pediatrics 2015;136:e1105-e1119

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Intraspinal Lipoma
Fawn tail in child with split cord malformation.

Mark Dias et al. Pediatrics 2015;136:e1105-e1119
Dermal sinus tracts presenting as a pit
Benign coccygeal dimple within the gluteal cleft and overlying the tip of the coccyx.

Innocent coccygeal pit in intergluteal cleft
The Problem with Pits LS Spine

- Myth that a dimple or pit is innocent if its base can be visualized – superficial component of some DST’s are blind-ended

- Location of the dimple along the craniocaudal axis most important
  - Flat part of sacrum at S2 greatest risk

- Neurosurgeon’s decision to treat is made based on presence of a pathologic dimple, regardless of imaging findings -- REFER
Cutaneous Signs LS Spine

- **High risk**
  - Midline mass, membranous ACC
  - Thick tuft of terminal hair
  - Pit above intergluteal crease
  - Vascular stains
  - Segmental hemangioma

- **Low risk**
  - Off midline
  - Diffuse fine hair
  - Pit within intergluteal crease
  - Pigmentary changes
  - Isolated deviation of intergluteal crease
Checklist for LS Spine

- Who: other midline defects, GU or GI anomalies
- What: pit, ACC, nodule, vascular stain, segmental hemangioma, tail, terminal hair tuft
  - More than one anomaly
- When: present at birth
- Where: midline, above the gluteal crease
- How: ultrasound if first six months, MRI and...
How

- Refer to neurosurgery if high risk REGARDLESS of imaging
11 year old with slowly enlarging nodule
Thyroglossal, Branchial Cleft

http://www.ghorayeb.com/branchialcleft.html

Neck: Developmental Anomalies

- Thyroglossal duct cyst
- Midline cervical cleft
- Midline anterior neck inclusion cyst
- Branchial cleft cyst
- Thymic cyst
- Cartilaginous rests/wattles
- Bronchogenic cysts
Neck: Pits, Tags, Nodules

- **High risk**
  - Present at birth
  - Midline or just anterior to SCM
  - Nodule, pits, especially with discharge

- **Low risk**
  - Later onset
  - Off midline, not anterior to SCM
  - Firm blue plate like nodule or milia like superficial cyst even if midline

Figure 1

More Than Meets the Eye
Remember Importance of “Where”:

Illustration  Textbook of Neonatal and Infant Dermatology, 3rd Edition
Checklist

- Who: neurodevelopmental issues, other midline defects?
- What: vascular stain, pit, aplasia cutis, nodule, mass?
  - High risk, low risk
- When: congenital, later?
- Where: midline, at “hotspot”?
- How: imaging, referral?
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Photo courtesy of Ann Petru