Checklists for Pigmented Lesions and Hamartomas

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Disclosure of Relationships with Industry

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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

- **Mosaic hamartomas** - checklist for when to evaluate for associated systemic findings and what to evaluate for
  - Congenital Melanocytic Nevi
  - Becker’s Nevi
  - Sebaceous Nevi
  - Epidermal nevi

- **Patterned pigmentation** - checklist for when to evaluate for associated systemic findings and what to evaluate for
  - Segmental Pigmentation Disorder
  - Nevoid Hyper/Hypo-melanosis
  - Blaschkonian pigmentation as a marker for McCune Albright Syndrome
  - Pigmentation of the genitals as a marker for PTEN Hamartoma Syndrome
Large Congenital Melanocytic Nevus- mosaic NRAS mutation
Initial Visit Checklist

• Does this child have neurocutaneous melanosis?
• Is there a melanoma?
• What are these weird lumps?
Neurocutaneous melanosis?

• 50% who develop symptoms do so prior to 1 year
  – most by 2 years of age
  – another small peak at time of puberty

• **Number of satellites most predictive**
  – >20 satellites 5 fold increase in risk of NCM
  – 3 or more small or medium congenital nevi with no "mother ship"

• **Size**
  – > 20cm increases risk

• **Location of LCMN**
  – Posterior midline
Large mothership with multiple satellites

Multiple small/medium congenital nevus phenotype
Imaging for NCM

• **Image if:**
  – LCMN with 10 or more satellites
  – 3 or more small or medium congenital nevi
  – LCMN with posterior midline (+ spine)

• **MRI of the brain**
  – Ideally before 6 months of age
  – Try feed and swaddle to avoid general anesthesia
  – ? Contrast
  – Spine if possible, particularly if lumbosacral involvement

• **If positive**
  – Close follow up with Pediatric Neurology
  – Increases risk for melanoma
Is there a melanoma?

• **Risk is around 5% lifetime for LCMN**
  – Half occur before 5 years old and almost all before puberty
  – Larger size $\rightarrow$ higher risk (75% assoc w > 40 cm)
  – Truncal location and multiple satellites $\rightarrow$ increase risk
  – Risk much higher for melanomas including cutaneous and extra-cutaneous if NCM

• **Cutaneous MM present as deep, fast growing or ulcerated nodules in the mothership**
  – Palpate
  – Pictures

• **CNS melanoma is actually more common**
  – Especially if there is NCM with a LCMN
What are these weird lumps?

- **Proliferative nodules**
  - Don’t increase risk of melanoma
  - Ulcerate less often and less extensively than melanoma
  - Atypical histologic feature common on biopsy
    - Cytologic atypia, architectural disorder, pagetoid spread, high mitotic index
    - IH, FISH seem to have limited value
  - Get expert and second opinions
First Follow up Checklist

• Will it fade?
• Should we go straight to the surgeon?
• Support groups
Will it fade?
Should we go to the surgeon?

- Get to know your family
- Join the support group before we have this discussion
- Complex discussion, best with experienced surgeon
  - Does not eliminate risk
  - Scars vs nevi - function and form
  - Issues of general anesthesia
Support Groups -
*parents immediately, child before school age*

Nevus Outreach - [www.nevus.org](http://www.nevus.org)

Nevus Network - [www.nevusnetwork.org](http://www.nevusnetwork.org)

The Congenital Nevus Support Group
2018 - Our 35th Year!
Follow up Checklist LCMN

• Serial exam with palpation every 3 months first year
  – Q 6 months until age 5 then annually
• Total body photography
• Counsel regarding xerotic skin and hypohidrosis
• Counsel not to let this limit activity due to fear
Individual Small or Medium CMN
MM in small/intermediate CMN

- Most were superficial
- Age range 18 to 79 years.

Illig L, et al. Congenital nevi less than or equal to 10 cm as precursors to melanoma. 52 cases a review, and a new conception. Arch Dermatol. 1985;121:1274-81.
Checklist Small and Medium CMN

• Risk of Melanoma low
  – <1% over a lifetime
  – Occur after puberty

• Periodic evaluation after puberty with photos

• Discussion of removal
  – Functional concerns
  – Psychosocial concerns
  – Usually wait till after 3 yo or until child can participate in discussion
The exception to the rule

- 8 year-old Report of MM arising in CMN
- Change over months
- No regular medical monitoring done prior to visit for nodule
- PET scan and sentinel node neg
- NED 12 months later

Favorite References


Becker’s Nevus
Becker’s Nevus Checklist

• Is it in a female over the breast?
  – Watch breast development
  – Consider spironolactone 50-100 qd

• If extensive rare skeletal or muscular abnormalities
  – Scoliosis most common

• Increased sebum production
  – Tinea versicolor, acne, pityrosporum folliculitis
    • Selenium sulfide wash
Nevus Sebaceous is mosaic HRAS or KRAS Mutation
Epidermal Nevus is mosaic of HRAS, KRAS, FGFR3 or PIK3CA mutations
Checklist for EN and NS

• Detailed Physical Exam to determine
  – Extent
  – Multifocality (mouth, genitals, scalp)
  – musculoskeletal abnormalities

• Detailed History to ask about
  – Developmental milestones
  – Seizures
  – Issues with vision
Localized NS
Localized NS Checklist

• Tumor growth
  – Benign:
    • trichoblastomas, syringocystadenoma papilliferum
  – Malignant:
    • Basal cell cancer most common
    • < 1% in 651 excised NS in children (Rosen et al 2009)

• Excise? When?
  – Psychosocial, risk of general anesthesia, family ethic/culture
  – There is no rush ➔ after 3 yo
  – Check in around puberty when thickening
Localized EN Checklist

- Less risk of tumors
- No medical reason to excise, based on disfigurement
- Rule out associated overgrowth syndromes like CLOVES, SOLAMEN, Proteus
Extensive/Multifocal NS Checklist

• **Neurology Consult**
  – 7% in recent cohort of 196 patients w neurologic issues
  – More common with *centrofacial involvement*
  – Intellectual disability and seizures most common
  – Screening imaging not helpful unless symptoms
    • 75% will have normal imaging

• **Ophthalmology Consult**
  – 2% in recent cohort of 196 patients
  – More common with neurologic abnormalities
  – Choristomas, colobomas, strabismus most common

• **Skeletal exam and look for scoliosis**
  – *Scoliosis, gait, limb length, shoe wear patterns*
  – Think about hypophosphatemic vitamin D-resistant rickets
    • *Bone pain, impaired mobility, bony deformities* (birth to puberty)
**Extensive/Multifocal EN Checklist**

- Neurology Consult
- Ophthalmology Consult
- Skeletal Exam
- Consider hypophosphatemic vitamin D-resistant rickets
  - Bone scans, calcium, phosphorous

- ***Is it epidermolytic keratinocytic nevus***
  - Biopsy at some point to look for epidermolytic hyperkeratosis
  - K1, K10 mutations → extensive or over the gonads then offspring can have epidermolytic ichthyosis
Favorite References


A funny café au lait spot:
Could this be NF1?
Checklist for patterned pigmentation

• **What is the pattern here?**

• **Detailed physical exam**
  – Are there other birth marks, CALM, skin findings, or stigmata of NF1?
  – Could this be McCune Albright?
    • Is this café au lait or just café?
    • Is this jagged coast or smooth?

• **Detailed History**
  – Developmental milestones
  – Issues with vision
  – Endocrine or precocious puberty
Segmental Pigmentation Disorder

• Blocky, segmental, hyper/hypo-pigmented, patches with midline cutoffs
• Smooth borders
• Café au lait, not just café

• Generally good prognosis
  (Hogeling M, Frieden IJ. Br J Dermatolog 2010)
  – Ask about developmental milestones
  – Talk about CNS and eyes but no routine referrals
  – Sun protection, self tanners
If the pigmentary mosaicism is more blaschkonian
Nevoid Hyper/Hypo-pigmentation

• Useful term for pigmentary change in more blaschkonian pattern
• Perhaps more associated systemic findings then segmental pigmentation disorder
    • 54 patients with nevoid hyper and hypo pigmentation
    • 15/54 had neurologic abnormalities, usually developmental delay and seizures
    • 3/54 hemihypertrophy
    • 2/54 cardiac: PDA, VSD
    • 1 with conical teeth
    • 1 with scoliosis
Is this McCune Albright?

• CALM is most common presenting sign
  – Usually present at birth
  – Unilateral with sharp midline cutoff, segmental
  – blaschkoid
  – Usually darker- just café, with no milk
If worry for McCune Albright:

• Bone survey for **polyostotic fibrous dysplasia**
  – Craniofacial 90% by 3.5 yrs old
    • Painless lump
  – Extremities 90% by 14 yrs old
    • Limp, pain, pathologic fracture

• Endocrine abnormalities
  – Hyperparathyroid, pituitary adenomas (GH), adrenal adenomas (Cushing, aldosteronism)

• Precocious puberty
  – Menstrual spotting
  – Scrotal thickening and enlargement
CALM looking macule of the genitals
If lentigines of penis or vulva:

• Look for
  – Macrocephaly
  – Lipomas
  – Vascular malformations
  – Oral papillomas, acral keratoses, acanthosis nigricans
  – Joint hyperextensibility, scoliosis

• Ask about
  – Hypotonia, developmental delay, autism
  – Hamartomatous intestinal polyps (PHx, FHx)
PTEN Hamartoma Syndrome
(Bannayan-Riley-Ruvalcaba)

- In a patient with “autism”
Favorite References


Figure 3

Large verrucous epidermal nevus on head of a Thai man who as a child said he remembered the life of his paternal uncle, who was killed with a blow on the head from a heavy knife.