Birthmarks: When to worry, when to reassure
Aimee Smidt, MD, FAAD, FAAP
Associate Professor, Depts of Dermatology and Pediatrics
University of New Mexico School of Medicine
November 2016

Goals and Objectives
1. Identify and describe newborn congenital skin lesions aka “birthmarks”
2. Identify common vascular birthmarks; distinguish worrisome from benign
3. Identify common pigmented birthmarks; distinguish worrisome from benign
4. Identify other birthmarks; distinguish worrisome from benign

Description: Primary Lesions

- MACULE: Flat < 1 cm
- PAPULE: Raised < 1 cm
- VESICLE: Clear fluid-filled < 1 cm
- PUSTULE: White fluid-filled
- PATCH: Flat > 1 cm
- PLAQUE: Raised > 1 cm
- NODULE: Raised + Deep > 1 cm
- TUMOR: Raised + Deep > 1 cm
- BULLA: Clear fluid-filled > 1 cm

Descriptions, the most important part...

- Location/Distribution
- Solitary vs Multiple
- Size (measure!)
- Color
- Primary lesion
- (Secondary changes)
- Configuration

This newborn has a subtle, mottled vascular patch. The most likely diagnosis is:

A. Aplasia cutis
B. Capillary malformation/port wine stain
C. Forceps injury
D. Infantile hemangioma
(Infantile) Hemangiomas

- Up to 10% healthy infants
- Benign vascular tumor
- Known associations: Female, Preterm
- Superficial, Deep, or Combined
- Localized vs Segmental
- Complications:
  - Disfigurement/QoL issue
  - Ulceration (pain, scarring)
  - Function-Threatening

Hemangioma Growth Pattern

Hemangiomas: When to Worry

- Function-Threatening or Symptomatic (based on location):
  - Periorbital
  - Lip
  - Nasal tip
  - Perineum
  - Airway
  - Hepatic (5+ skin)

Hemangiomas: When to Worry/Refer

- Cosmetic/Psychosocial
- "Beard" distribution (airway)
- Ulcerating (pain, scarring)
- Multiple (>5) – Abd US
- Lumbosacral
- Large Facial (PHACES)
- Atypical History or Appearance

PHACE Syndrome

P – Posterior Fossa Abnormality
H – Hemangioma (Large, Facial)
A – Arterial Anomalies (Head/Neck)
C – Cardiac/Coarctation
E – Eye Associations

Workup:
- Head/Neck MRI/MRA
- EKG/Echo (needs to visualize aorta)
- Ophtho evaluation

LUMBAR Syndrome

- Analogous to PHACE but affecting pelvis
- Often ulcerating but thin hemangioma, segmental in perineum to lumbosacral area

Workup:
- Spine/pelvis MRI
- Appropriate referral to Urology, Neurosurgery as necessary
Hemangiomas: Management

- Appropriate treatment in many: “Active nonintervention”
- Periodic monitoring during expected proliferative phase
- We emphasize regression, not necessarily complete resolution

Therapies:
- Oral Propranolol
- Topical Timolol
- Less commonly used: oral, IL, or topical steroids
- (Surgery)

Congenital Hemangiomas

- Fully formed at birth – no growth phase
- Subtypes
  - Rapidly involuting (weeks to months)
  - Partially involuting (years)
  - Non-involuting
  - Involuting (fetal growth)
- Management
  - Active nonintervention*
  - Imaging/Close monitoring if diagnosis in question

This newborn has a dull pink-red homogenous patch over the hemiface. The most likely diagnosis is:

A. “Angels Kiss” birthmark
B. Birth trauma
C. Capillary malformation/port wine stain
D. Infantile hemangioma
E. I don’t know – we need to wait to find out

Capillary Malformations/ “Port wine stains”

- Vascular misdevelopment, not tumor
- Present at birth, no proliferative phase in infancy
- Can thicken, darken over time
- Treatment – Laser if desired
  - Can start any time
  - Q 6 weeks
  - 8-10 sessions + “touch-ups”
  - Bond with baby first!

Capillary Malformations: When to Worry

Sturge-Weber Syndrome
- Sporadic disorder characterized by:
  - Facial port wine stain
  - Ipsilateral leptomeningeal vascular malformation
  - Vascular malformation of choroid (eye)
- Seizures, DD, glaucoma
- Almost exclusively with V1 distribution
- +/- other dermatomes
- Risk 8% with classic V1 distribution
- Risk increases if multiple dermatomes: risk if bilateral 24%
- Any periorcular involvement warrants Ophtho exam!
- Brain MRI for diagnosis

Capillary Malformations: When NOT to worry

- Synonyms:
  - Fading macular stains
  - Angels Kiss
  - Stork Bite
  - Nevus simplex
  - Nevus simplex complex
- No treatment or evaluation typically needed
- Fade with time (months to years)
- If centrofacial may be more persistent
- May develop dermatitis
Newborn with a large brown plaque on his leg. Parents want to know if this is a birthmark or something more worrisome. He is full term, vaginal birth with no complications. There is a FH of melanoma. What is the most likely diagnosis?

A. Café-au-lait macule  
B. Congenital melanocytic nevus  
C. Dermal melanocytosis  
D. Ecchymosis  
E. Epidermal nevus

Answer: B

A. Café au lait macule (CALMs are well demarcated, light brown, flat lesions with no hair)  
B. Congenital melanocytic nevus  
C. Dermal melanocytosis (ill-defined, blue-gray patches, resembling a bruise)  
D. Ecchymosis (medical term for a bruise, non-raised skin discoloration due to the escape of blood into the tissues)  
E. Epidermal nevus (raised tan or brown linear birthmarks due to overgrowth of the epidermis, usually become verrucous with time)

Congenital Melanocytic Nevi: Clinical

Proliferations of benign melanocytes (pigment cells)  
Also known as ‘Moles’  
Occur in 1-3% of newborns

Morphology  
• Macules, papules, or plaques at birth  
• Tan, brown, dark brown, or black in color  
• Texture is smooth, verrucous, or cobblestone-like  
• Hair may or may not be present  
• Pigmentary and surface changes can develop with time

CMN: Classification

CMN are classified according to the projected size in adulthood into:  
- Small (most common): diameter up to 1.5 cm  
- Intermediate/Medium (more common): 1.5-20 cm  
- Large (less common): 20-40 cm  
- Giant (least common): 40 cm +

Lesions grow in proportion to the individual  
Projected size can be estimated (about double in adulthood)  
Correlates with malignancy potential  
- Small and medium CMN have <1% risk of malignant transformation  
- Larger CMN have a higher potential for malignant transformation, particularly when satellite nevi are present  
- Surgical removal doesn’t completely eliminate the risk of melanoma, but may improve QoL

This newborn has a giant very dark brown plaque over most of his back. In addition to size, which of the following characteristics is most predictive of potential for other issues?

A. Development of nodules  
B. Degree of hypertrichosis  
C. Lesion distribution  
D. Number of satellite nevi  
E. Surface rugosity

CMN: When to worry

- Large, giant, or numerous CMN/“satellite nevi” are at risk of a condition called neurocutaneous melanosis  
- Patients at risk should be screened with MRI of the brain and spine  
- Larger CMN have a higher potential for malignant transformation into melanoma, particularly when satellite nevi are present  
- Surgical removal doesn’t completely eliminate the risk of melanoma, but may improve QoL.
CMN: Prognosis and Treatment

• Treatment depends on age of the patient, size of nevus, location, family preference.
• Periodic surveillance is indicated in patients with CMN but does not need to be urgent
• Suspect melanoma if new lumps or bumps, area of ulceration that doesn’t heal, sudden growth, changes in border, changes in color (ABCDEs)
• Surgery is indicated in giant congenital melanocytic nevus when a melanoma develops within it
• Reassurance and monitoring most common approach

Other benign pigmented “birthmarks”

Dermal melanocytosis/“Mongolian spots”

Pigmentary mosaicism

This child was born with a smooth, hairless, linear plaque on the temple. What is the most likely diagnosis?

A. Aplasia cutis congenita
B. Congenital melanocytic nevus
C. Juvenile xanthogranuloma
D. Nevus sebaceus
E. Seborrheic dermatitis

Answer: D

A. Aplasia Cutis (absence of a portion of skin most commonly located on the scalp)
B. Congenital melanocytic nevus (well delimited dark macule-plaque with or without hair)
C. Juvenile xanthogranuloma (solitary yellowish papule-nodule usually not present at birth)
D. Nevus Sebaceus
E. Seborrheic dermatitis (erythematous and scaly plaques often seen on the scalp and face)

Nevus Sebaceus or ‘organoid hamartoma’

• 0.3% of newborns
• Overgrown epidermis (upper layers of the skin), sebaceous (oil) glands, hair follicles, apocrine glands and connective tissue.
• Occurs primarily on the scalp/face
• Presents as a solitary, smooth, yellow-orange hairless patch, often oval or linear in shape.
• Usually become more pronounced around adolescence, and often appearing bumpy, warty or scaly.
• (May look more pronounced at birth due to effect of Mom’s hormones)

The parents are relieved that you are able to diagnose the growth. What will you tell them about prognosis and treatment?

A. Early detection was critical because nevus sebaceus often heralds serious, systemic disease
B. This has a high risk of malignant transformation, and surgical excision is strongly indicated
C. This has a low risk of malignant transformation, but the lesion should be monitored for changes
D. This has no malignant potential and does not require treatment, surgical excision may be used for cosmetic reasons
E. This will grow rapidly for several months and begin to resolve after one year, without treatment
Nevus Sebaceous: Management

- Usually diagnosed clinically, based on history and clinical features
- A very large nevus sebaceous rarely presents associated with disorders of the eye, brain and skeleton (Sebaceous Nevus Syndrome)
- Most nevus sebaceus remain benign throughout life
- Most growths that arise within nevus sebaceus are benign
- Very rarely skin cancer may arise, usually in adulthood
- Reassurance and non-urgent monitoring is most appropriate
- Surgical removal often performed in childhood or adolescence, depending on family preference

Related: Epidermal Nevi

- Warty/stuck on papules or plaques
- Pink-tan to brown or gray
- Often linear, curvilinear
- Present at birth
- Any location
- Overgrowth of epidermis only
- No malignant potential
- Sometimes surgical removal later in life for aesthetic reasons
- If very extensive, concern for epidermal nevus syndrome (CNS, eye, bony associations)

Thank you!
asmidt@salud.unm.edu