

Newborn Skin Disease Part 1: Birthmarks

Basic Dermatology Curriculum



Content for this module was developed
by The Society for Pediatric Dermatology

Goals and Objectives

- The purpose of this module is to help medical students develop a clinical approach to the evaluation and initial management of common birthmarks in newborns.
- By completing this module, the learner will be able to:
 - Identify which birthmarks require diagnostic workup or intervention.
 - Understand the clinical presentation and natural history of birthmarks, including nevus sebaceous, aplasia cutis, congenital melanocytic nevi and dermal melanocytosis.
 - Understand potential complications of congenital melanocytic nevi.

Summary of Birthmarks

Birthmarks discussed in this module:

- Congenital melanocytic nevus
- Aplasia cutis congenita
- Nevus sebaceous
- Dermal melanocytosis

- Note: Vascular birthmarks will be covered in the Vascular lesion module and Neurofibromatosis and Tuberous sclerosis will be covered in the Inherited skin conditions module

Case One

Joshua

Case One: History

ID: 3 -week-old male

HPI: Joshua's parents have brought him to clinic because they are concerned about a large brown area on his thigh. Parents want to know if this is a birthmark or something more worrisome.

PMH: full-term, vaginal birth with no complications.

FHx: Mother is a healthy 27-year-old, no history of medications during or after pregnancy. Joshua's paternal grandmother was diagnosed with melanoma at age 65.

Case One: Skin Findings



Case One: Question 1

Based on Joshua's history and this image of his lesion, what is the most likely diagnosis?

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

Case One: Question 1

Answer: B

Based on Josua's history and this image of his lesion, what is the most likely diagnosis?

- 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)

Differential Diagnosis

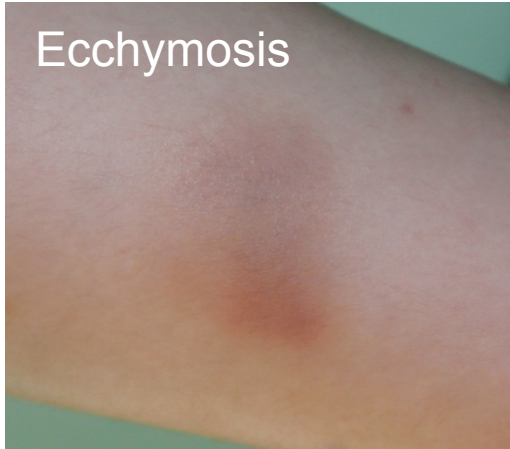
Café-au-lait macules



Dermal melanocytosis



Ecchymosis



Epidermal nevus



Congenital Melanocytic Nevi: Clinical Presentation

- Proliferations of benign melanocytes (pigment cells)
- Also known as 'Moles'
- Congenital melanocytic nevi (CMN) occur in 1 to 3 percent of newborn infants

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

Congenital Melanocytic Nevi: Classification

CMN are classified according to the projected size in adulthood into:

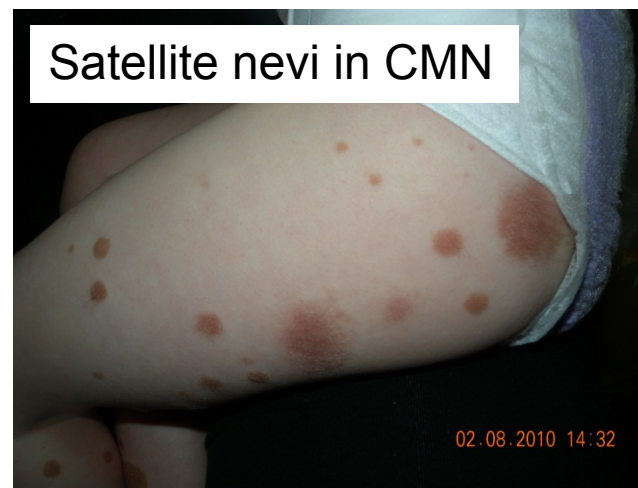
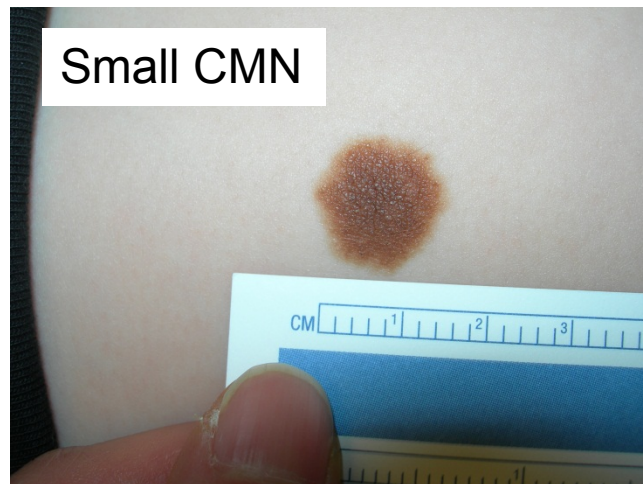
- Small (most common): diameter up to 1.5 cm
- Intermediate/Medium (more common): 1.5 to 20 cm
- Large (less common): 20 to 40 cm
- Giant (least common): 40 cm or more

- Lesions grow in proportion to the individual
- Projected size can be estimated

Congenital Melanocytic Nevus: Risk of Malignancy

- The importance of this classification is that it correlates with the malignancy potential.
- Small and medium sized CMN have less than 1% risk of malignant transformation.
- In large and giant lesions the risk is higher; ranging from 0-7.6%, with most studies reporting an estimated risk of about 2%.

Clinical Presentations



Congenital Melanocytic Nevus: Additional Categorization

Diameter and size has been the lone criterion for categorizing CMNs and assessing the risk of complications. The following are additional characteristics that can be used to categorize CMNs:

Localization

- Head, Trunk, and/or Extremities

Number of satellite nevi

- None, <20, 20-50, or >50

Color heterogeneity

- None, moderate, or marked

Surface rugosity (wrinkling or creasing)

- None, moderate, or marked

Dermal or subcutaneous nodules

- None, scattered, or extensive

Hypertrichosis (hairiness)

- None, notable, or extensive

Congenital Melanocytic Nevus: Diagnosis

Diagnosis is made clinically based on the natural history, morphology and in some cases dermoscopic evaluation

Histology

- Biopsy can be used to confirm the diagnosis and/or to rule out melanoma.

More information on nevi can be found in the module, [Evaluation of pigmented lesions](#)

Case One: Question 2

In addition to size, which of the following characteristics is most predictive of potential for malignancy?

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

Case Eight: Question 2

Answer: D

In addition to size, which of the following characteristics is most predictive of potential for malignancy?

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

Small and Medium-sized Congenital Melanocytic Nevi

- Monitored and managed on an individual basis
- Risk of melanoma is low
- Because potential for malignant transformation is low, usually a secondary reason might trigger removal (irritation, cosmetic concern)
- CMN require periodic checks (particularly after puberty)

Large, Giant, and Numerous Congenital Melanocytic Nevi

- Patients with large, giant, or numerous CMN are at risk of a condition called neurocutaneous melanosis, which involves the central nervous system
 - Patients at risk can be screened with MRI of the brain and spine during the first six months of life if this is suspected
- Larger CMN have a higher potential for malignant transformation into melanoma, particularly when satellite nevi are present
- Surgical removal doesn't eliminate the risk of melanoma in the setting of large/giant nevi. (50% of melanomas presented in CMN occur at another site)

Congenital Melanocytic Nevus: Prognosis and Treatment

- Treatment of CMN depends on age of the patient, size of nevus, location and depth.
- Periodic surveillance is indicated in patients with CMN
- Suspect melanoma in CMN that present new lumps or bumps, area of ulceration that doesn't heal, sudden growth, changes in border, changes in color
- Surgery is indicated in giant congenital melanocytic nevus when a melanoma develops within it
- Prophylactic surgical removal doesn't eliminate the risk of melanoma (50% of melanomas presented in CMN occur at another site)

Case Two

Ana

Case Two: History

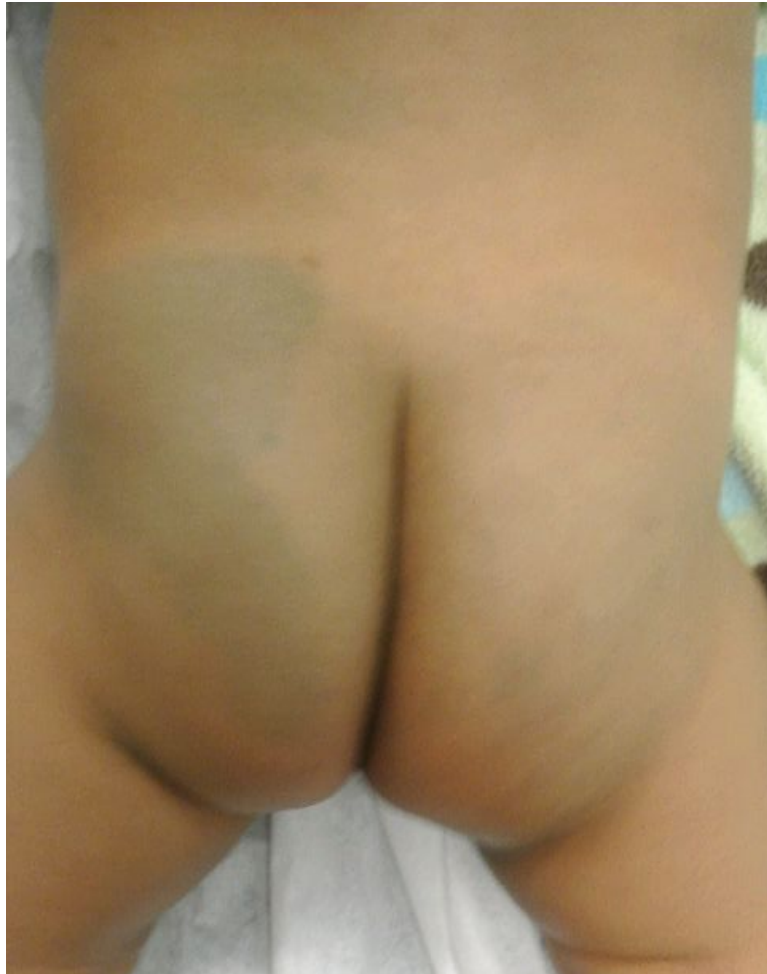
ID: 6 day-old female (Hispanic background)

HPI: at birth, Ana presents with a large gray-dark spot, located on the lower back and buttock. She asymptomatic and otherwise healthy newborn

PMH: full-term, normal pregnancy and vaginal birth

FHx: Mom is an immigrant from Mexico. She said Ana's older brother was born with a similar patch. He is currently 9 y/o and he is no longer having any evidence of that pigmentation.

Case Two: Skin Findings



Case Two: Question 1

Based on the history and previous picture, what would be the explanation for Ana's skin findings?

- A. Ana is victim of child abuse
- B. Ana has a congenital vascular malformation
- C. Ana has a congenital melanocytic nevus
- D. Ana has a severe diaper rash
- E. Ana has congenital dermal melanocytosis

Case Two: Question 1

Based on the history and previous picture, what would be the explanation for Ana's rash?

Answer. E

- A. Ana is victim of child abuse (ecchymosis usually presents with purpuric discoloration)
- B. Ana has a congenital vascular malformation (pigmentation in vascular lesions are more bluish/purplish)
- C. Ana has a congenital melanocytic nevus (pigmentation in congenital melanocytic nevus are usually light or dark brown)
- D. Ana has a severe diaper rash (usually characterized by erythema)
- E. Ana has a congenital dermal melanocytosis (also known as Mongolian spot)**

Congenital Dermal Melanocytosis (CDM): Clinical Presentation

Epidemiology

- Dermal melanocytosis is more common in individuals with darker skin types
- It is usually present at birth or becomes evident on the first weeks of life

Morphology

- CDM or Mongolian spot presents as a patch of blue-gray pigmentation with irregular shape, unclear edges and normal skin texture
- More commonly seen as a single lesion, however multiple lesions can be seen

Distribution

- Has a predilection for buttocks and lower back, but it can also affects other body parts

Congenital Dermal Melanocytosis : Diagnosis

Etiology

- Dermal melanocytosis represents the entrapment of melanocytes in the dermis during their migration from the neural crest into the epidermis.

Diagnosis

- CDM is diagnosed clinically based on the history and clinical presentation
- In most cases, no work up is necessary.
- Extensive Dermal melanocytosis may be the initial presentation of some rare metabolic disorders (GM1 gangliosidosis or Hurler syndrome) or spinal meningeal anomalies
- Consider further work up in cases where there is extensive involvement with failure to thrive or failure to meet developmental milestones.

Case Two: Question 2

What would you tell Ana's mother regarding her condition?

- A. Ana needs to be referred to plastics for removal of the lesions
- B. Ana has higher risk of developing skin cancer (melanoma)
- C. Ana doesn't need any treatment
- D. Hydroquinone cream is indicated to treat Ana's condition

Case Two: Question 2

What would tell Ana's mother regarding her condition?

Answer. C

- A. Ana needs to be referred to plastics for removal of the lesions (surgical removal is no indicated in this condition)
- B. Ana has higher risk of developing skin cancer (Mongolian spots have no risk to develop skin cancer)
- C. Ana doesn't need any treatment. (CDM usually improves in appearance before adulthood)**
- D. Hydroquinone cream is indicated to treat Ana's condition (this medication is used to treat melasma but has no indications for Mongolian spots)

Case Three

Kenny

Case Three: History

ID: 3-day-old male

HPI: Kenny presents at birth with hairless lesion located on the left side of his forehead and scalp, extending close to his eyebrow

PMH: full-term, vaginal birth with no complications

FHx: Mother is a healthy 33-year-old, no history of medications during or after the pregnancy

Case Three: Skin Findings



Case Three: Question 1

Based on Kenny's history and this image of Kenny's lesion, what is the most likely diagnosis?

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

Case Three: Question 1

Answer: D

Based on Kenny's history and this image of her lesion, what is the most likely diagnosis?

- 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 Sebaceous**
- E. Seborrheic dermatitis (erythematous and scaly plaques often seen on the scalp and face)

Differential Diagnosis



Nevus Sebaceous: Clinical Presentation

- Nevus sebaceous or 'organoid hamartoma' is seen in 0.3% of newborns
- It consists of overgrown epidermis (upper layers of the skin), sebaceous glands, hair follicles, apocrine glands and connective tissue.
- Occurs primarily on the scalp or 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.

Case Three: Question 2

Kenny's 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 sebaceous often heralds serious, systemic disease
- B. Nevus sebaceous has a high risk of malignant transformation, and surgical excision is strongly indicated
- C. Nevus sebaceous has a low risk of malignant transformation, but the lesion should be monitored for changes
- D. Nevus sebaceous has no malignant potential and does not require treatment, surgical excision may be used for cosmetic reasons
- E. Nevus sebaceous will grow rapidly for several months and begin to resolve after one year, without treatment

Case Three: Question 2

Answer: C

Kenny's 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 sebaceous often heralds serious, systemic disease (Nevus sebaceous can be associated to other conditions, but this is rare)
- B. Nevus sebaceous has a high risk of malignant transformation, and surgical excision is strongly indicated (Risk of malignant transformation in childhood is low, and surgical excision is no longer automatically indicated)
- C. **Nevus sebaceous has a very low risk of malignant transformation, but the lesion should be monitored**
- D. Nevus sebaceous has no malignant potential and does not require treatment, surgical excision may be used for cosmetic reasons (There is a low risk of malignant transformation)
- E. Nevus sebaceous will grow rapidly for several months and begin to resolve after one year, without treatment (nevus sebaceous grows in proportion to the individual)

Nevus Sebaceous: Diagnosis

Diagnosis

- Nevus sebaceous is usually diagnosed clinically, based on history and clinical features
- A large nevus sebaceous rarely presents associated with disorders of the eye, brain and skeleton (Sebaceous nevus syndrome)
- Most nevus sebaceous remain benign throughout life
- Most growths that can arise from nevus sebaceous are benign, and very rarely skin cancer may arise from it

Nevus Sebaceus: Diagnosis

Biopsy

- Atypical cases may warrant histological evaluation
- Characteristic histological features are more developed in adolescence and adulthood
- Histologic changes in infants tend to be subtle

Treatment

- Intermittent interval follow up is recommended
- Referral a dermatologist or surgical removal is indicated if a lump, warty growth, non-healing sore or any other concerning change is observed

Case Four

Sophia

Case Four: History

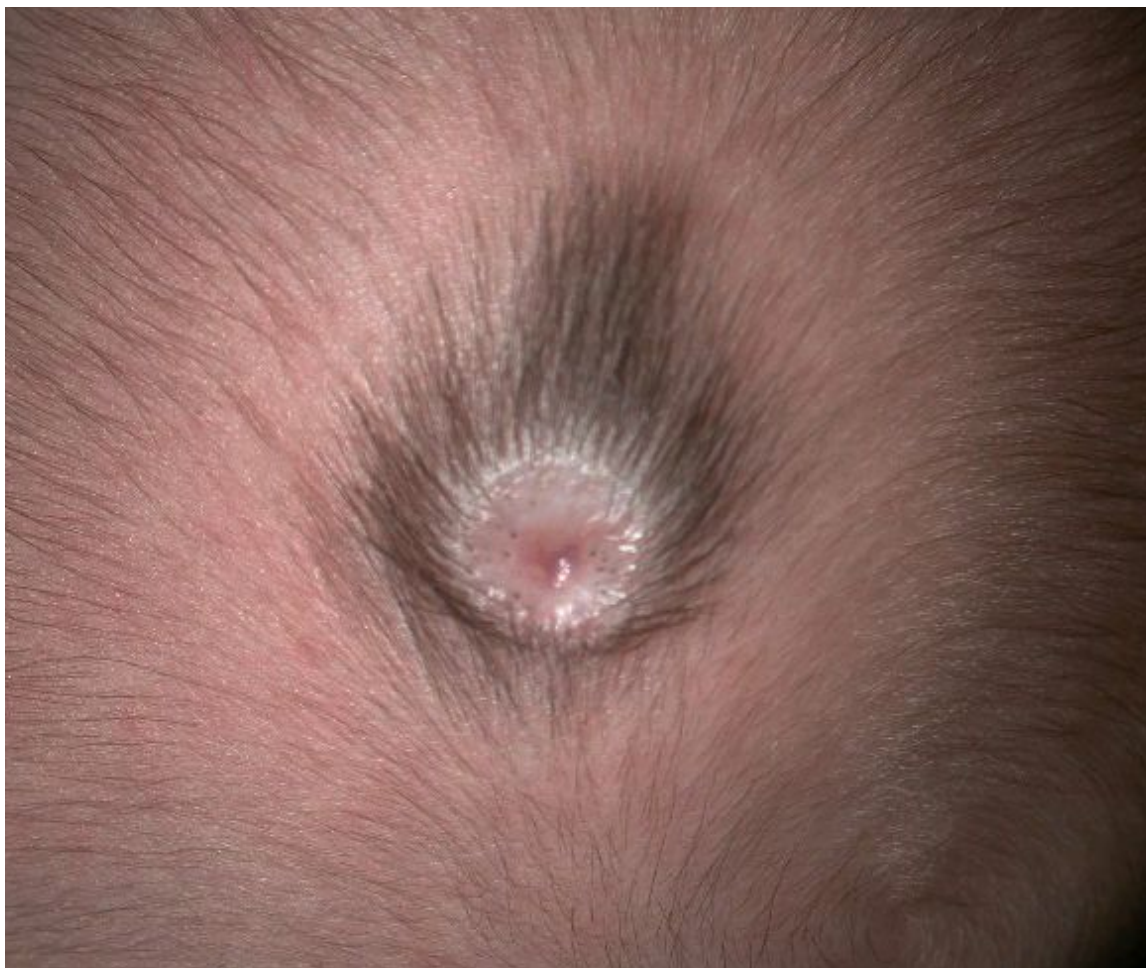
ID: 20-day-old female

HPI: Sophia presented at birth one deep ulcer on the scalp. Parents state now the ulcer has healed, leaving skin changes behind. She is afebrile with no systemic symptoms.

PMH: Full-term, vaginal birth assisted with forceps.

FHx: Mother is a healthy 25-year-old, with no history of medications during or after pregnancy. Her last pap test before her pregnancy was unremarkable

Case Four: Skin Findings



Case Four: Question 1

During physical exam you found a circular healed ulcer surrounded by a dark tuft of hair. Based on the history and the clinical findings which would be your diagnosis?

- A. Aplasia cutis congenita
- B. Epidermolysis bullosa
- C. Herpes simplex
- D. Birth trauma
- E. Staphylococcal impetigo

Case Four: Question 1

During physical exam you found a circular healed ulcer surrounded by a dark tuft of hair. Based on the history and the clinical findings which would be your diagnosis?

Answer. B

- A. **Aplasia cutis congenita**
- B. Epidermolysis Bullosa (EB is characterized by skin fragility, lesions seen are blisters or bullae that can be present at birth or develop secondary to friction or trauma after birth)
- C. Herpes simplex (this viral infection presents with clusters of small vesicles)
- D. Birth trauma (usually located bilaterally on the face/scalp)
- E. Staphylococcal impetigo (lesions are characterized by fragile bullas/pustules followed by superficial erosions)

Aplasia Cutis Congenita (ACC): Clinical Presentation

ACC refers to the absence of skin present at birth that can be localized or widespread

- It has no sexual or racial predilection
- Its incidence has been reported to be 2.8 cases per 10,000 newborns
- It can be an isolated finding or be associated with other developmental anomalies

Morphology

- Lesions are well demarcated and may be scarred, superficially eroded or deeply ulcerated
- Their size varies (very small to large) and can be circular, oval, linear or stellate
- Lesions may be covered by a membranous epithelium giving them a bulla-like appearance (bullous aplasia cutis)

Aplasia Cutis Congenita (ACC): Clinical Presentation

Distribution

- Scalp is most frequently affected, 86% occurs on the vertex of the scalp, but ACC can be found in any part of the body
- Single lesions are more common up to 70-75%, 20% of the lesions present in pairs and the remainder are multiple
- Some times a darker tuft of hair surrounds the periphery of the scalp defect (hair collar sign) and may indicate an underlying neural tube defect

Examples of aplasia cutis



Case Four: Question 2

True or False

All patients with aplasia cutis, regardless of where the area of ACC is located, require an MRI to rule out internal involvement.

Case Four: Question 2

Answer. False

Not all patients with ACC require imaging. If the ACC presents a hair collar sign, if it has a midline location or if there are other bumps/lumps or palpable defects underlying the area of ACC, imaging should be requested since this might indicate deeper embryologic defects.

ACC: Diagnosis

Diagnosis

- ACC is diagnosed clinically based on the morphology and history of the lesions
- No specific laboratory tests are required for this condition
- Imaging studies are rarely required for small lesions of the scalp with no other anomalies associated
- Genetic counseling may be considered if associated anomalies are found
- Atypical or very large scalp defects, and those with hair collar sign, should be imaged to rule out underlying bone, vascular, or soft tissue defects

ACC: Treatment and Prognosis

Treatment

- Therapy of ACC depends primarily on the size, depth, and location of the cutaneous defect
- Gentle cleansing and the application of bland ointment or topical antibiotic ointment can be indicated to prevent infection
- Referral to Neurosurgery for surgical repair may be indicated for large or multiple scalp defects

Prognosis

- In general prognosis for ACC is excellent
- However larger lesions with periosteal, dural or bone defects carry additional risk and management considerations as mentioned above

Summary table

Congenital melanocytic nevus	Dermal melanocytosis	Nevus sebaceous	Aplasia cutis congenita
Proliferation of benign melanocytes	Entrapment of melanocytes in dermis	Organoid hamartoma	Absence of skin present at birth
Pigmented macules, papules or plaques; +/- hair	Blue-gray pigmentation, ill defined	Smooth, yellow-orange hairless patch	Presents as superficial erosion, ulceration or scar
Risk of melanoma in large/giant CMN	Benign, predilection buttocks/lower back	More pronounced around adolescence	Atypical/large or hair collar sign indicate need for imaging



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End of Module