Congenital Pigmented Lesions
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Disclosures

• I have no conflicts of interest or relevant financial relationships to disclose.
• I will not be discussing off-label uses of any medications.
Objectives

(1) Evaluate the classification of congenital melanocytic nevi.

(2) Evaluate dermal melanocytosis.

(3) Identify the diagnostic workup and differential diagnosis for children with multiple café-au-lait macules.
Congenital Melanocytic Nevi (CMN)

• Classified by projected size in adulthood
  – Small: <1.5cm
  – Medium: 1.5cm-19.9cm
  – Large/Giant: >20cm

• Risk of malignant melanoma (MM) is increased in large/giant nevi

• Evidence regarding prophylactic excision of congenital nevi to prevent MM is controversial
Dermoscopy/Epiluminescence Microscopy
Small CMN
Medium CMN
Giant CMN
Satellite Nevi
Neurocutaneous Melanosis (NCM)

• Melanocytic proliferation within the leptomeninges or brain parenchyma in the context of a CMN

• Risk factors for NCM
  • Presence of a GCMN
  • Male sex
  • Satellite nevi or multiple CMN
  • Head, neck, or posterior midline location

NCM

• Symptomatic patients generally present before age 2 → poor prognosis
• MRI brain/spine required for diagnosis → no effective treatment, no clear guidelines on who needs imaging
Novel Treatments for NCM

• Targeted molecular therapies used in the treatment of MM are now being used experimentally for treatment of NCM
• Pediatric experience is limited but this may be a future therapeutic option

Dermal Melanocytosis
(formerly Mongolian Spots)
Extensive Dermal Melanocytosis:
GM1-Gangliosidosis, Type 1

Café-au-lait macule (CALM)
Multiple CALM

- NF1 (≥6 CALM, ≥0.5cm in size in children, ≥1.5cm in adults)
- MEN Type 1
- Tuberous sclerosis
- Many others (search OMIM)
Multiple CALM

- NF1
  - ≥6 CALM
  - ≥0.5cm in size in children
  - ≥1.5cm in adults
- MEN Type 1
- Tuberous sclerosis
- Many others
  - Search OMIM.org
Search: 'cafe-au-lait macule'
Results: 1 - 10 of 281

1:  *613113. NEUROFIBROMIN 1; NF1*
   Cytogenetic location: 17q11.2, Genomic coordinates (GRCh37): 17:29,421,944-29,704,694
   Matching terms: cafeau-lait, macule

2:  #162200. NEUROFIBROMATOSIS, TYPE 1; NF1
   Cytogenetic location: 17q11.2
   Matching terms: cafeau-lait, macule

3:  #162210. NEUROFIBROMATOSIS, FAMILIAL SPINAL
   Cytogenetic location: 17q11.2
   Matching terms: cafeau-lait, macule

4:  114030. CAFE-AU-LAIT SPOTS, MULTIPLE
   Matching terms: cafeau-lait, macule

5:  #131100. MULTIPLE ENDOCRINE NEOPLASIA, TYPE 1; MEN1
   MEN1 SOMATIC MUTATIONS, INCLUDED
   Cytogenetic location: 11q13.1
   Matching terms: cafeau-lait, macule

6:  *607108. PAIRED BOX GENE 6; PAX6*
   Cytogenetic location: 11p13, Genomic coordinates (GRCh37): 11:31,806,339-31,839,508
   Matching terms: cafeau-lait, macule

7:  #276300. MISMATCH REPAIR CANCER SYNDROME; MMRCS
   Cytogenetic locations: 2p21, 2p16.3, 3p22.2, 7p22.1
   Matching terms: cafeau-lait, macule
Summary

• Congenital melanocytic nevi (CMN) are categorized by projected size in adulthood.
• Large/giant CMN have an increased risk for MM. Children with GCMN should have lifelong surveillance with a dermatologist.
• Dermal melanocytosis is most common on the back but can occur on other body sites.
• Multiple café-au-lait macules can be associated with multiple genetic syndromes.
Questions?

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