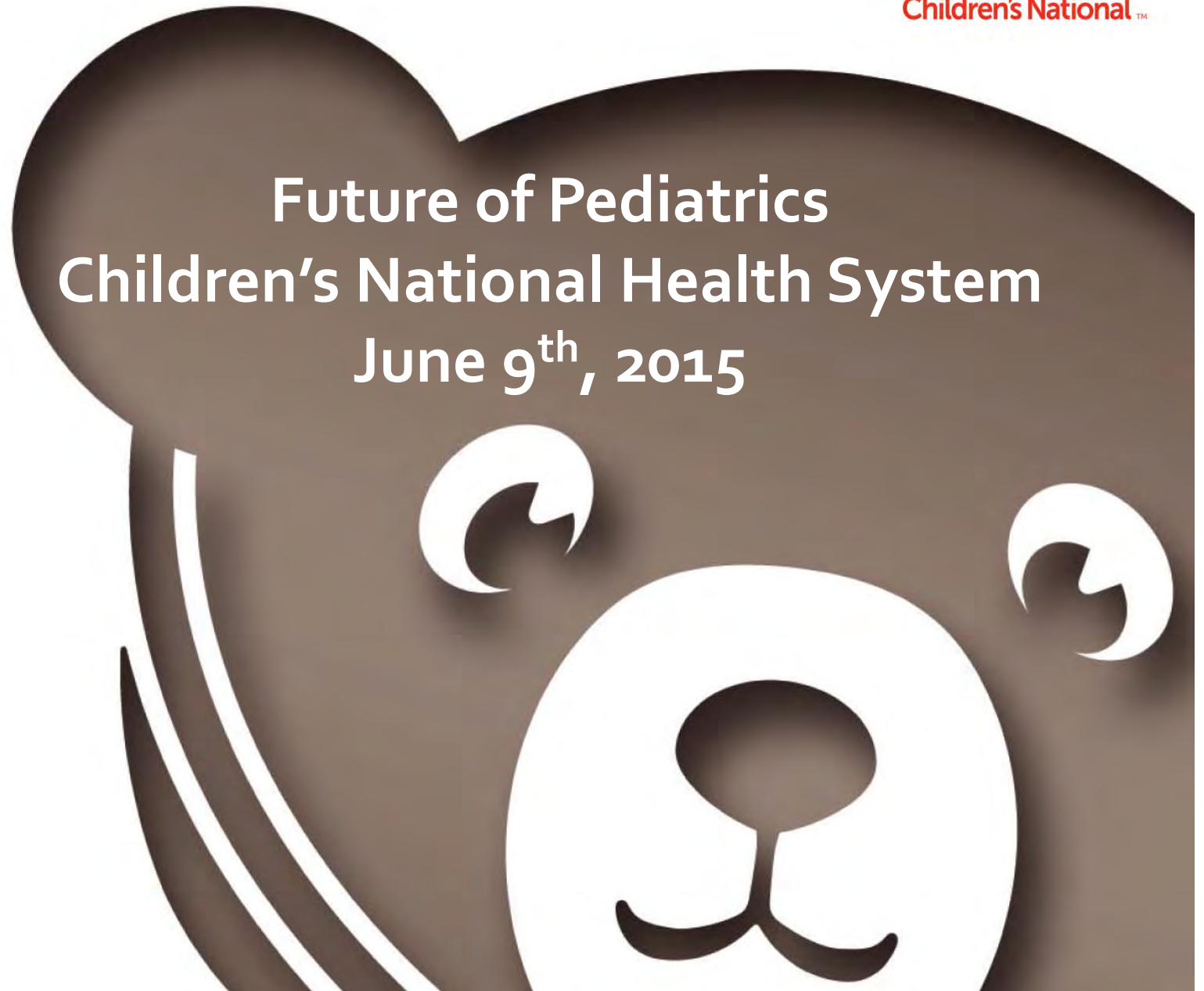




**Future of Pediatrics**  
**Children's National Health System**  
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# Congenital Pigmented Lesions

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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

Küsters-Vandavelde HV, Willemsen AE, Groenen PJ, Küsters B, Lammens M, Wesseling P, Djafarihamedani M, Rijntjes J, Delye H, Willemsen MA, van Herpen CM, Blokk WA. Experimental treatment of NRAS-mutated neurocutaneous melanocytosis with MEK162, a MEK-inhibitor. *Acta Neuropathol Commun.* 2014 Apr 8;2:41

# Dermal Melanocytosis (formerly Mongolian Spots)



# Extensive Dermal Melanocytosis: GM1-Gangliosidosis, Type 1

Bloch LD, Matsumoto FY, Belda W Jr, Giugliani R, Menezes LF, Kim CA, Machado MC. Dermal melanocytosis associated with GM1-gangliosidosis type 1. *Acta Derm Venereol.* 2006;86(2):156-8.

# Café-au-lait macule (CALM)







# Multiple CALM

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

# Multiple CALM

- NF1
  - $\geq 6$  CALM
  - $\geq 0.5$ cm in size in children
  - $\geq 1.5$ cm in adults
- MEN Type 1
- Tuberous sclerosis
- Many others
  - Search OMIM.org

cafe-au-lait macule

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Search: 'cafe-au-lait macule'

Results: 1 - 10 of 281 | [Show 100](#) | [Download As](#) ▾ | 1 2 3 4 5 6 7 8 9 10 Next Last

- 1 : [\\* 613113. NEUROFIBROMIN 1; NF1](#)  
Cytogenetic location: 17q11.2 , Genomic coordinates (GRCh37): 17:29,421,944-29,704,694  
Matching terms: cafeaulait, macule
- 2 : [# 162200. NEUROFIBROMATOSIS, TYPE I; NF1](#)  
Cytogenetic location: 17q11.2  
Matching terms: cafeaulait, macule
- 3 : [# 162210. NEUROFIBROMATOSIS, FAMILIAL SPINAL](#)  
Cytogenetic location: 17q11.2  
Matching terms: cafeaulait, macule
- 4 : [114030. CAFE-AU-LAIT SPOTS, MULTIPLE](#)  
Matching terms: cafeaulait, macule
- 5 : [# 131100. MULTIPLE ENDOCRINE NEOPLASIA, TYPE I; MEN1](#)  
MEN1 SOMATIC MUTATIONS, INCLUDED  
Cytogenetic location: 11q13.1  
Matching terms: cafeaulait, macule
- 6 : [\\* 607108. PAIRED BOX GENE 6; PAX6](#)  
Cytogenetic location: 11p13 , Genomic coordinates (GRCh37): 11:31,806,339-31,839,508  
Matching terms: cafeaulait, macule
- 7 : [# 276300. MISMATCH REPAIR CANCER SYNDROME; MMRCS](#)  
Cytogenetic locations: 2p21 , 2p16.3 , 3p22.2 , 7p22.1  
Matching terms: cafeaulait, macule

[mim.org/entry/162210?search=cafe-au-lait+macule&highlight=cafeaulait,macula,macule](http://mim.org/entry/162210?search=cafe-au-lait+macule&highlight=cafeaulait,macula,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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