AMSER Rad Path Case of the Month:

Neurocutaneous Melanosis

Gibson Klapthor
Medical University of South Carolina

Dr. Jeanne Hill and Dr. Laura Spruill
Medical University of South Carolina

Patient Presentation

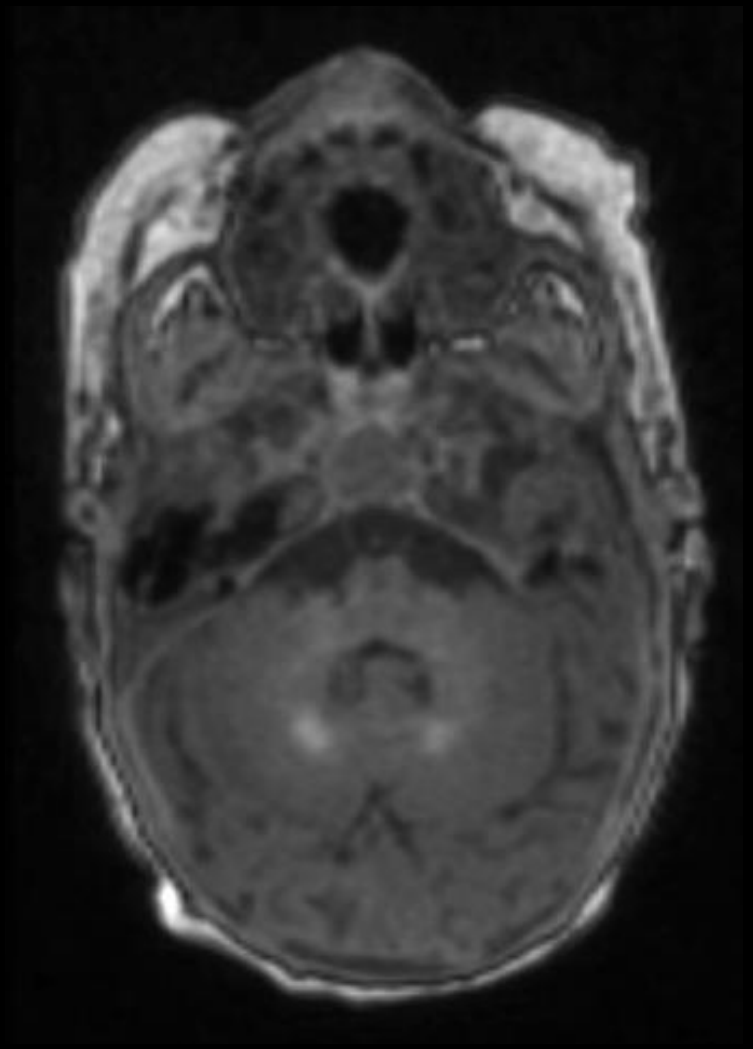
Clinical history: Pt is a 6 week old female with diffuse hyperpigmented skin lesions. A posterior mass was first identified on prenatal US at 36 weeks gestation. After birth, pt had a 3 week stay in the NICU where biopsies of the skin lesions and whole body imaging were obtained.

Pertinent social history: none

Pertinent physical exam findings: Skin: Large melanocytic nevi covering the entire body. The lesions completely cover the abdomen and genitalia and involve the head, trunk, and all extremities. Multiple satellite nevi over entire body including lips.

Pertinent Labs

- None



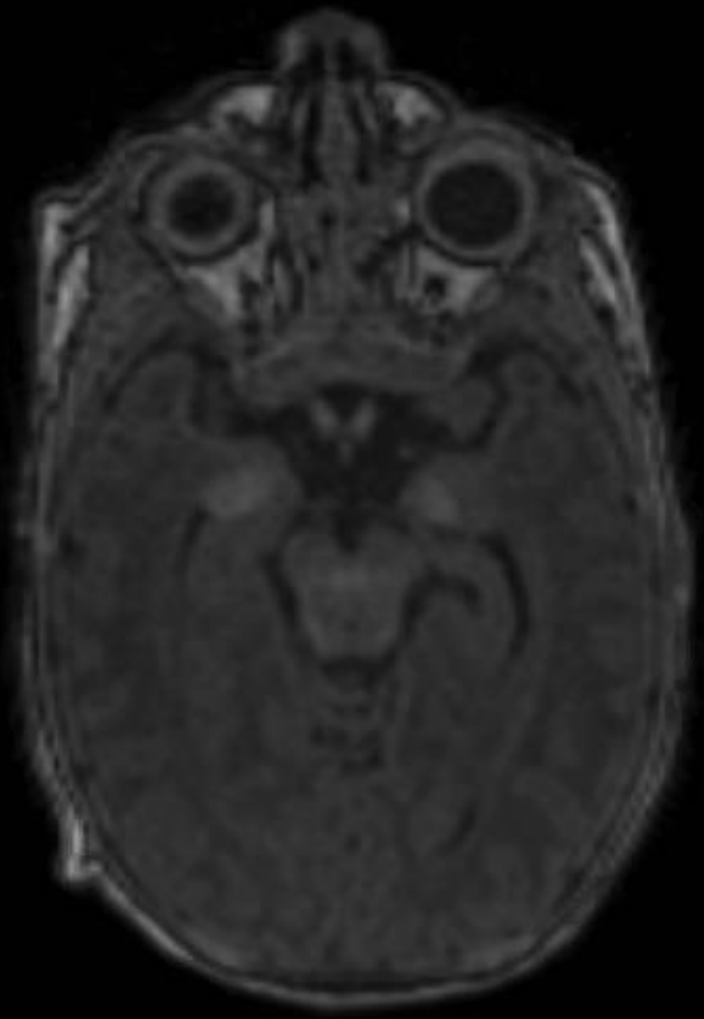


Figure 1:

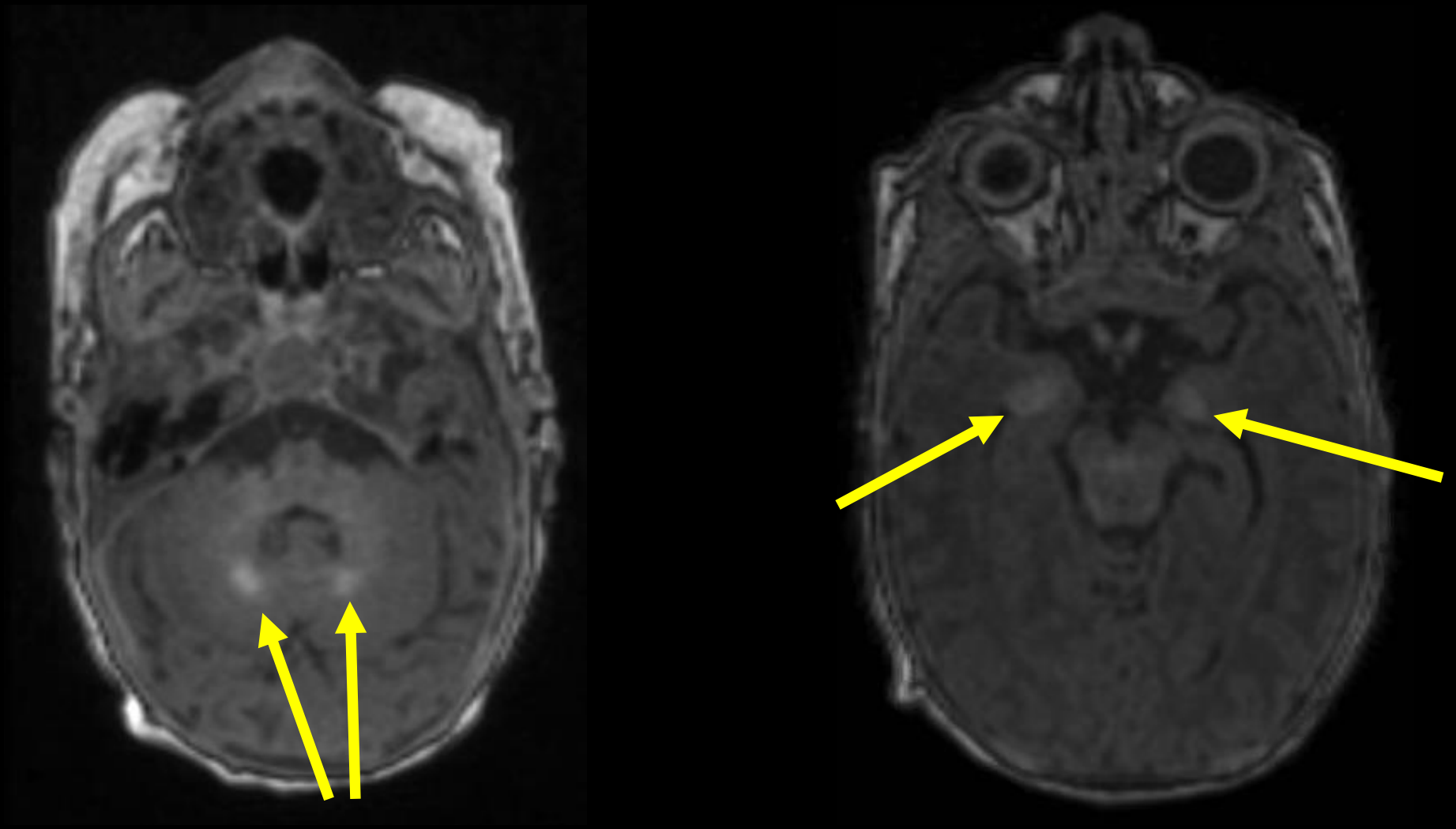
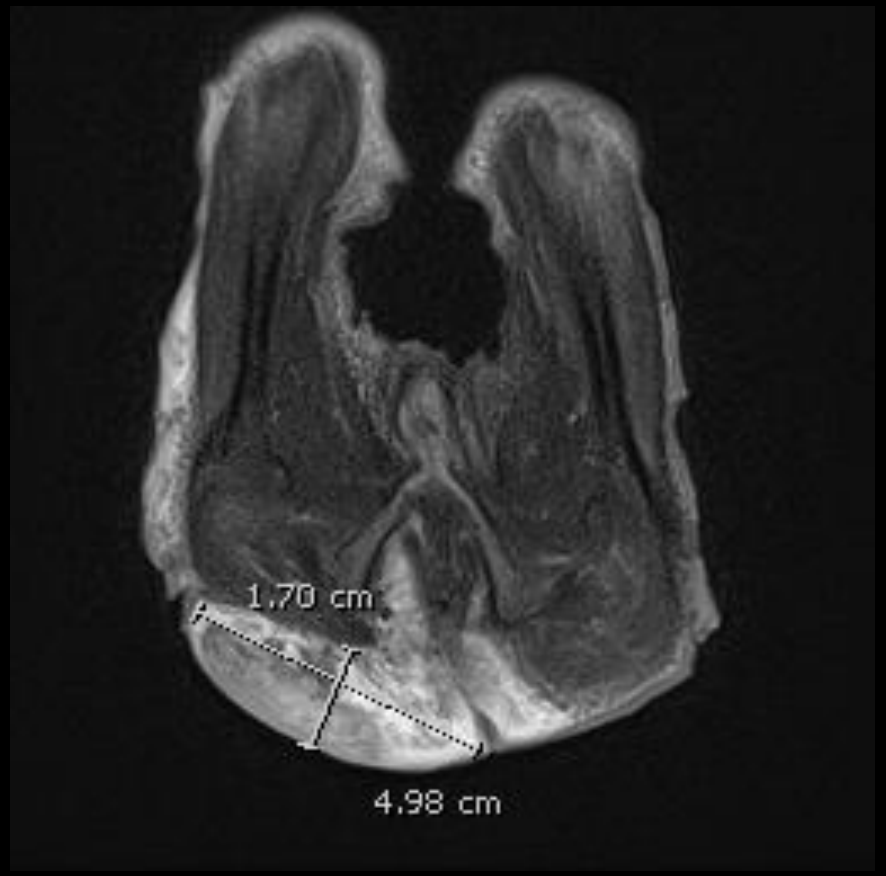


Figure 1: MRI Brain Axial T1-weighted. Intraparenchymal T1 hyperintensity within cerebellum (left) the bilateral amygdala (right)



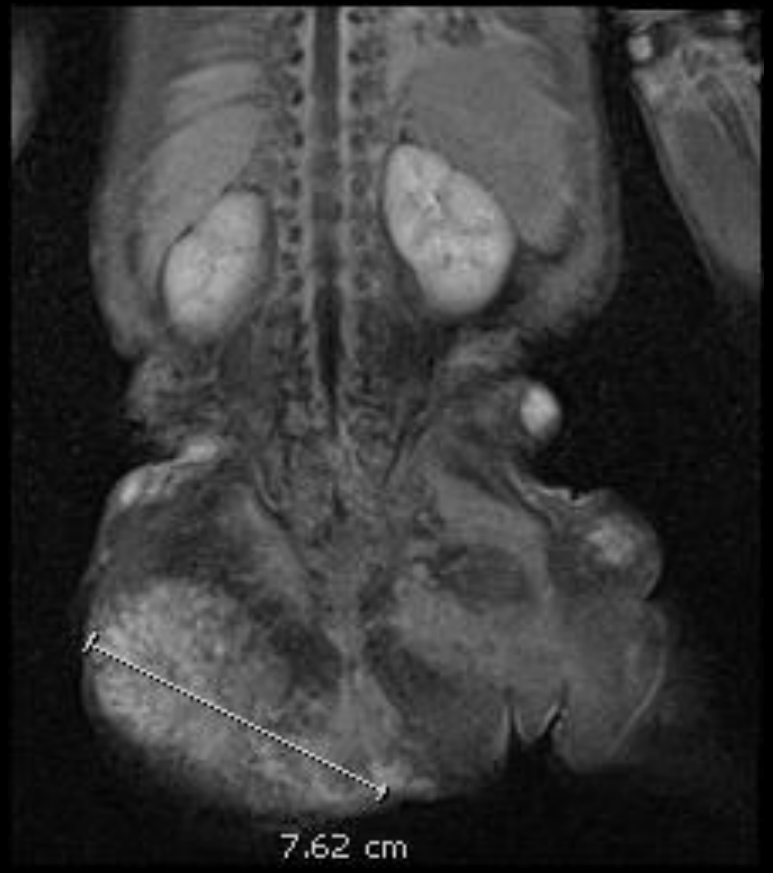
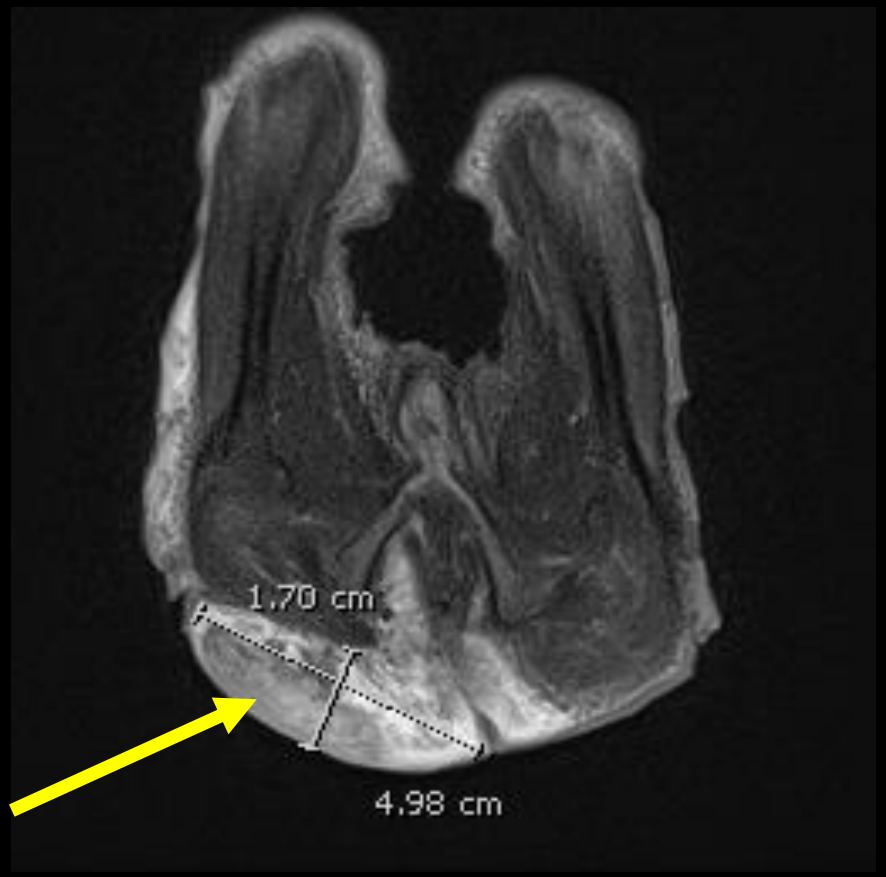


Figure 2:

Axial T2



Coronal T1 post contrast

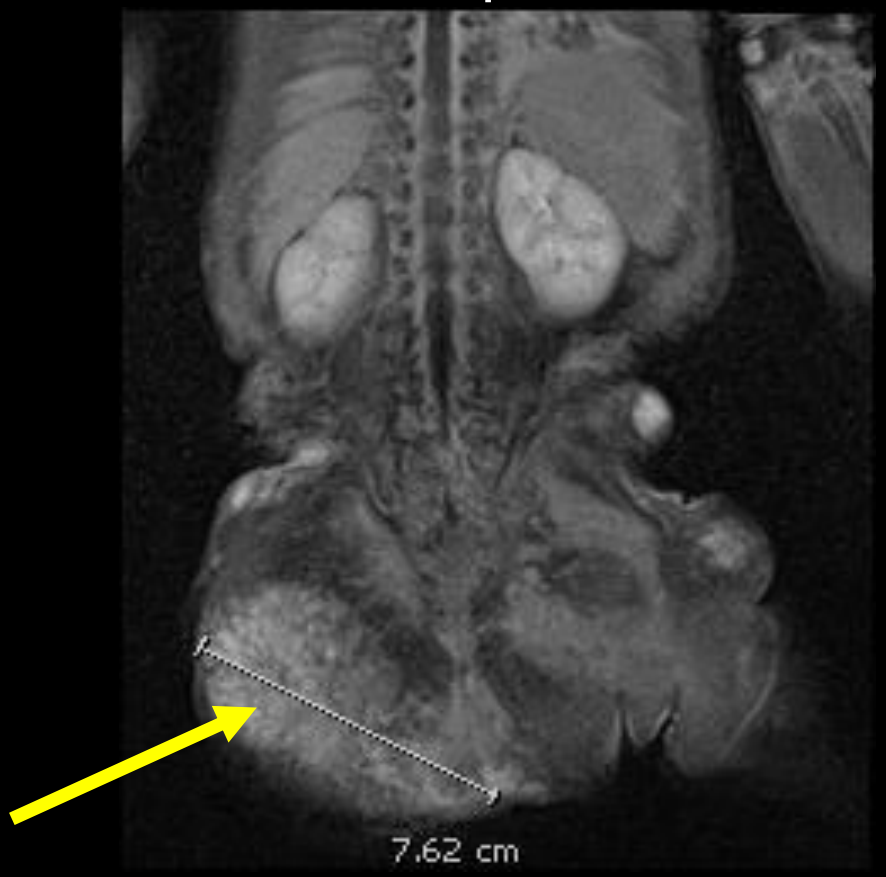
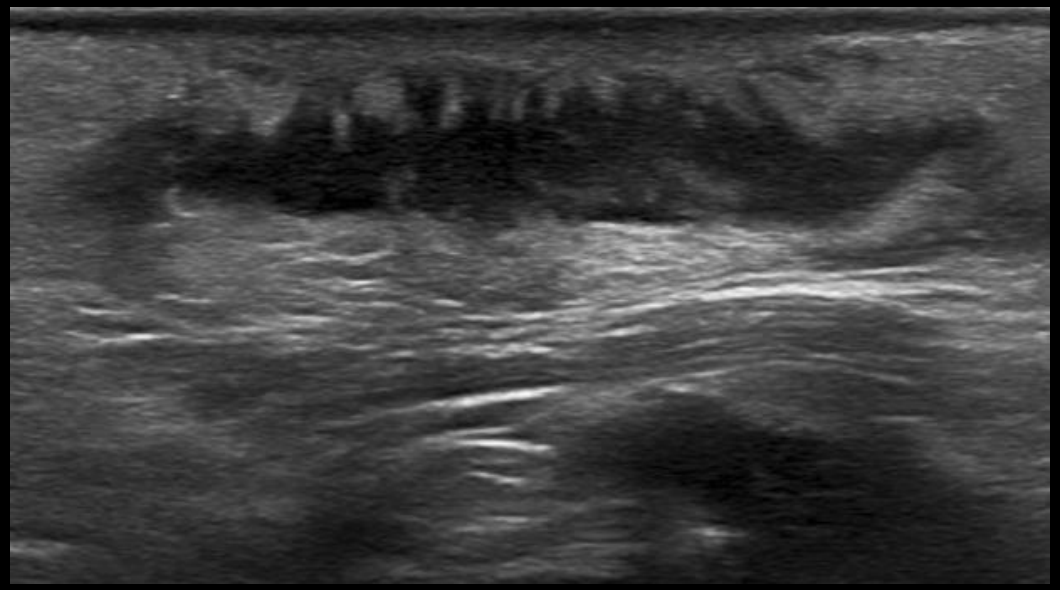


Figure 2: MRI Abd / Pelvis W Wo. Multiple subcutaneous lesions (T2 hyperintense (left), T1 intermediate (right), nondiffusion restricting (not shown)) with the largest area of abnormality involving the right buttock (arrows) and perineal region. Abnormal T2 signal is also seen along the lateral right leg (left).



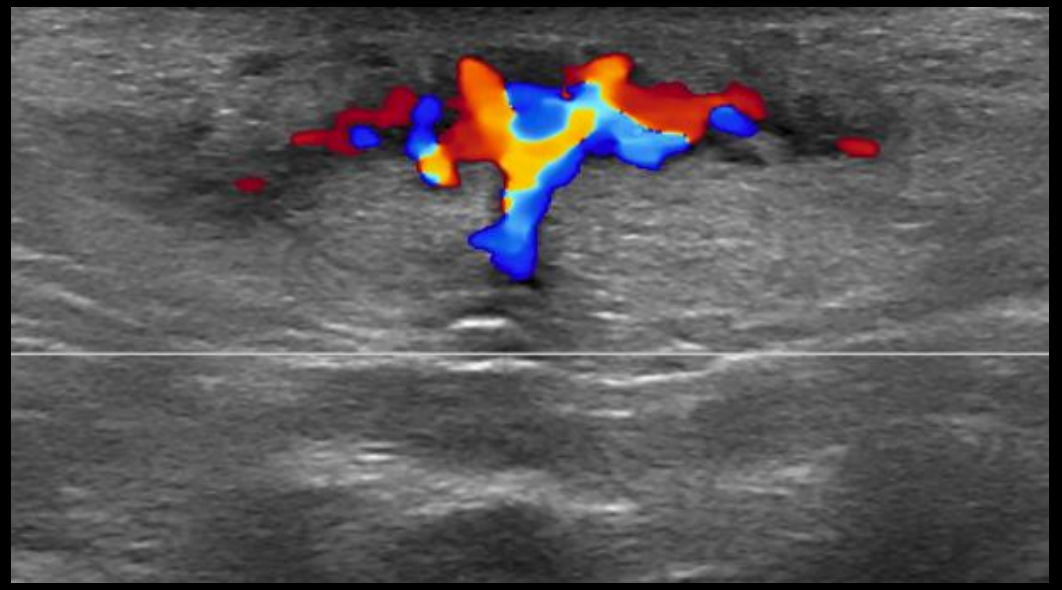


Figure 3:

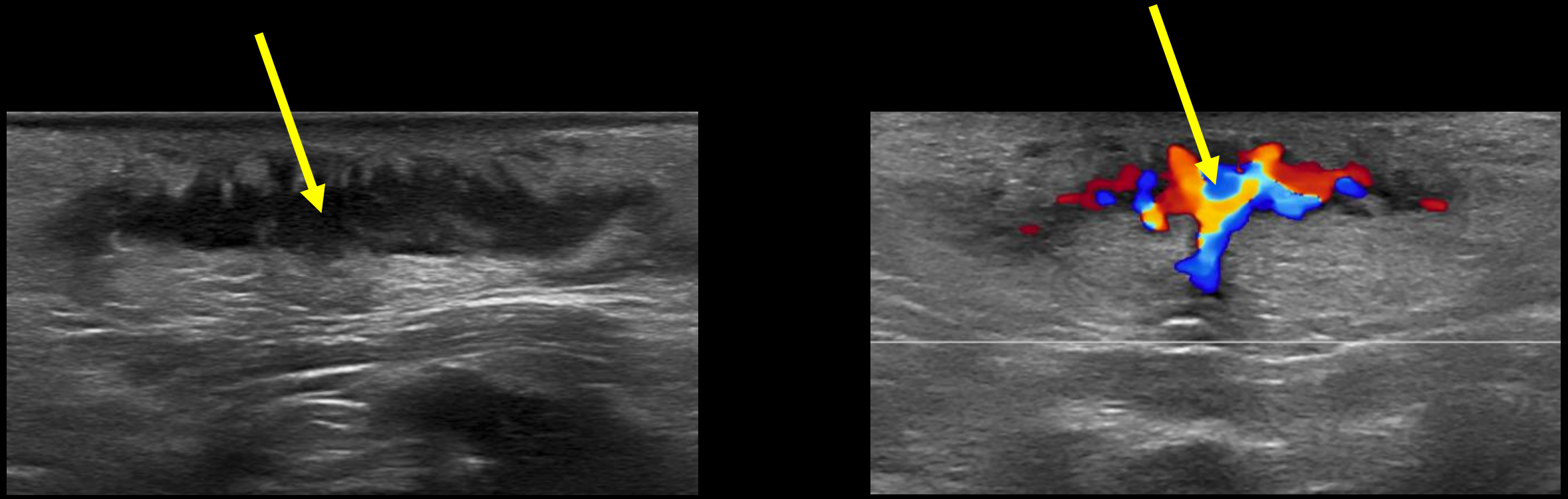


Figure 3: US Rt Gluteal Area. Soft tissue mass with hypoechoic component (left) containing color Doppler signal (right) and multiple arterial waveforms on pulse wave Doppler (not shown)

DDX (based on imaging)

- Congenital melanocytic nevus
- Atypical mole
- Becker's nevus syndrome
- Nevus of Ota / Nevus of Ito
- Malignant melanoma

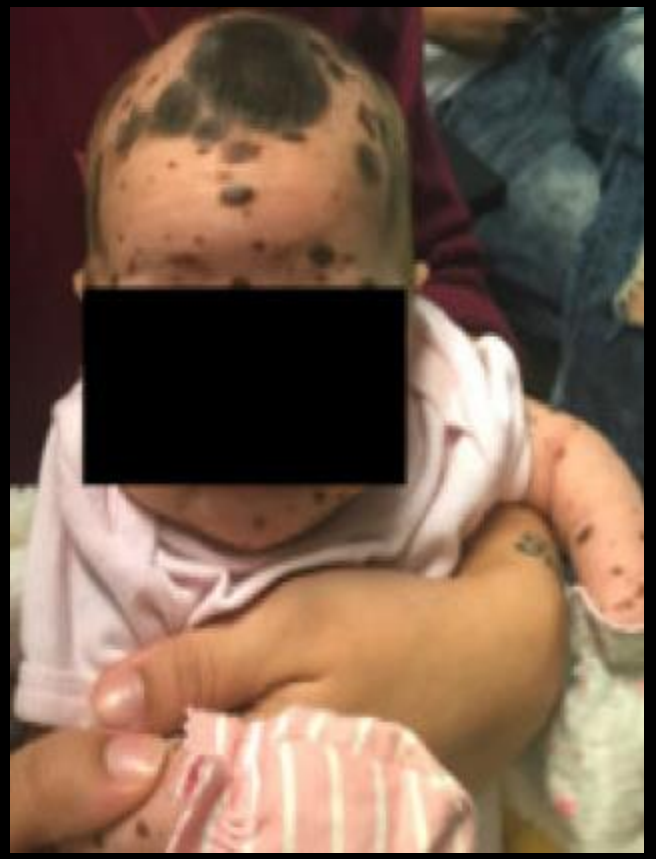


Figure 4: Hyperpigmented skin lesions

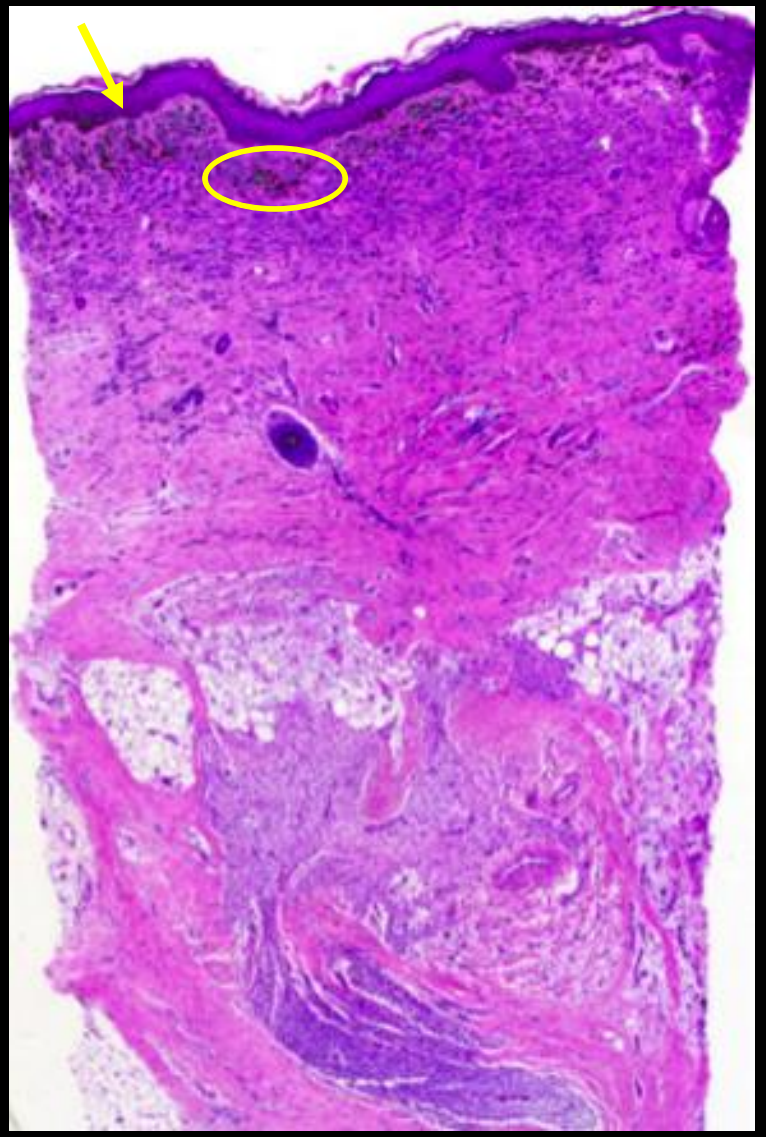


Figure 5: Microscopic pathology of gluteal biopsy. Low power view demonstrating melanocytic proliferation with an epidermal (arrow) and dermal (circle) component

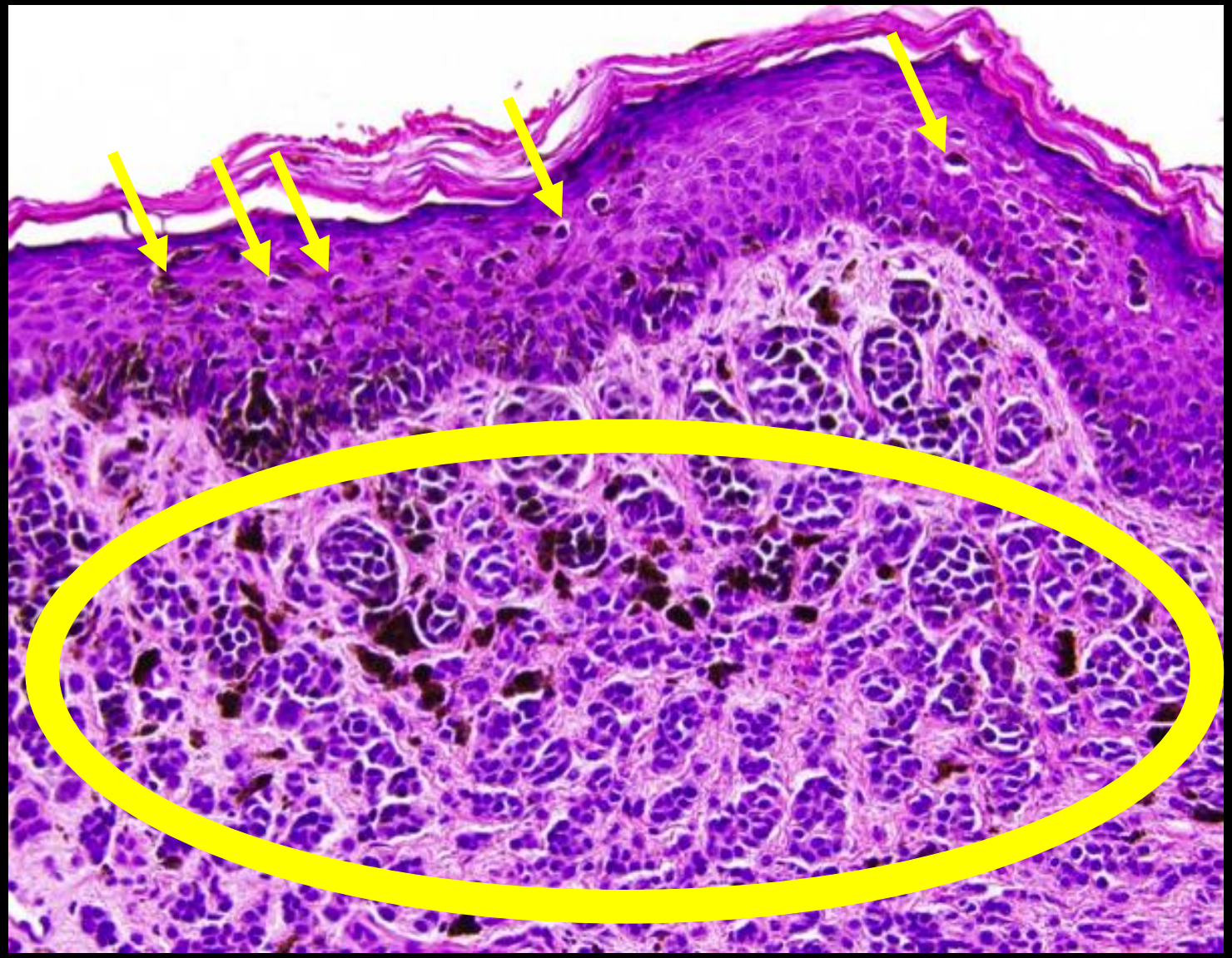


Figure 6: Microscopic pathology of gluteal biopsy. High power view demonstrating melanocytic proliferation with an epidermal (arrow) and dermal (circle) component

Final Dx:

Neurocutaneous melanosis
(subtype of congenital melanocytic nevus)

Case Discussion

Background

- Neurocutaneous melanosis is a rare nonfamilial syndrome characterized by giant and/or multiple congenital melanocytic nevi of skin and melanotic lesions in the central nervous system.
- Thought to be due to neural crest aberration during early embryonic development.
- CNS lesions primarily occur in pons, amygdala, cerebrum, cerebellum, and spinal cord. Typically asymptomatic but can cause symptoms of mass effect such as seizures and raised ICP. This is more likely if the lesions undergo transformation to leptomeningeal melanoma which occurs in almost half of patients.
- Generally has poor outcome with few treatment options. Some individuals with NCM have survived into adulthood, but most die in childhood before 10 years of age.

Case Discussion

Epidemiology:

- Incidence is unknown, but it is uncommon, with only 100 to 200 cases reported in the literature.

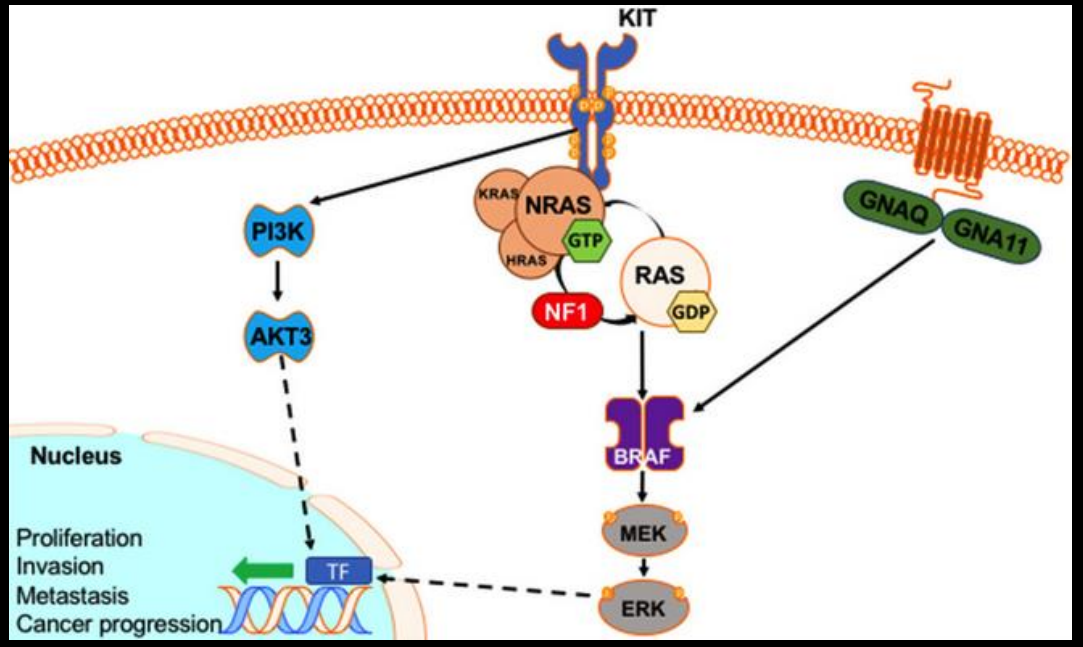
Risk factors for development of melanoma:

- A large CMN, especially if >40 cm predicted final size and in a posterior axial location
- Multiple satellite nevi
- More than two medium-sized CMN (especially if numerous)

Case Discussion

Pathogenesis of neurocutaneous melanosis

- The precise pathogenesis is not well understood, but disordered neural crest cell differentiation and melanocyte embryogenesis are suspected.
- The prominent involvement of the leptomeninges and skin over the spine supports the suggestion that the primary defect is abnormal migration of nevus cell precursors.
- In one study of 32 truly congenital nevi, no BRAF mutations were detected (as is commonly seen in melanoma), but 81% (26/32) had mutations in NRAS which drives excess production of melanin-producing cells



Shaughnessy M, Klebanov N, Tsao H. Clinical and therapeutic implications of melanoma genomics. *Journal of Translational Genetics and Genomics*. Sep 28, 2018. Licensed under CC BY 4.0.

References:

1. Bell D, Gerstenmaler F, et al. Neurocutaneous melanosis. *Radiopaedia*. Accessed 1/1/2020. https://radiopaedia.org/cases/neurocutaneous-melanosis

2. Hunt R, Schaffer J, Bolognia J. Congenital melanocytic nevi. In: *UpToDate*, Post, TW (Ed), UpToDate, Waltham, MA, 2020.

3. Islam, M. P. (2015). Neurocutaneous melanosis. In Neurocutaneous Syndromes (pp. 111–117). https://doi.org/10.1016/b978-0-444-62702-5.00007-x

4. Shaughnessy M, Klebanov N, Tsao H (2018). Clinical and therapeutic implications of melanoma genomics. *Journal of Translational Genetics and Genomics*.

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