TITLE: Pediatric Oculo(dermal) Melanocytosis: Risk factor for melanoma

ABSTRACT: A 15-year-old boy with ocular melanocytosis presents with episcleral pigmentation of the right eye. This report details the clinical characteristics of oculo(dermal) melanocytosis, the classifications, and the complications associated with this condition.

CASE HISTORY:
- 15-year-old Hispanic male
- CC: black pigmented spots on white part of right eye, first noted at age 7 years, over time has been getting worse/spots seem larger
- Ocular and Systemic History: unremarkable, 1st eye exam
- Medications: none, NKDA

PERTINENT FINDINGS:
- Unaided DVA: OD 20/30+, OS 20/20-
- Refractive Error: OD +0.75 -1.25 x 180 (20/20), OS +0.25 -0.50 x 005 (20/20)
- EOMs: full and unrestricted OD, OS; Pupils: PERR(-)APD; Cover Test: orthophoria D/N
- IOP GAT OD: 19, OS: 20 @ 3:20pm
- Slit Lamp Exam:
  - Adnexa: clear OU, no skin pigmentation around either eye/cheek area
  - Conjunctiva: ocular melanosis superior and inferior OD, clear OS
  - Iris: heterochromia, OD hyperpigmentation
- Dilated Fundus Exam:
  - C/D Ratio: 0.5H x 0.55V OD, 0.25r OS
  - Peripheral Retina: clear OU, subtle overall diffusely darker fundus OD
- Other Testing:
  - OCT ONH: size of ONH and C/D ratio OD>OS
  - OCT Anterior Chamber Angle: angles unremarkable OU, increase of iris pigmentation OD blocking OCT signal compared to OS
  - Fundus Photos: subtle, diffusely darker fundus OD
  - Anterior Segment Photos: OD dark grey, flat lesions with indistinct borders at layer of episclera, superior and inferior; iris heterochromia
  - B Scan: unremarkable (performed because the literature shows that patients with oculo(dermal) melanocytosis are at higher risk for choroidal melanomas)
  - Gonioscopy: OD increased pigment in anterior chamber angle compared to OS

DIFFERENTIAL DIAGNOSIS:
- Dermal Pigmentation: blue nevi, Mongolian spot, lentigo, melasma, Sturge Weber Syndrome
- Scleral Pigmentation: nevus, complexion associated melanosis, PAM, melanoma

DIAGNOSIS AND DISCUSSION: ocular melanocytosis OD
- Ocular melanocytosis is a congenital pigmented lesion that typically presents as a unilateral patchy-looking flat, blue-gray discoloration of the sclera; there is also an increase in intraocular pigmentation of the same eye
- When there is associated cutaneous (usually eyelid and adjacent skin) hyper pigmentation in addition to ocular involvement, it is called oculodermal melanocytosis or Nevus of Ota
- Structures most commonly involved in oculo(dermal) melanocytosis are: skin, episclera, iris, choroid, and anterior chamber angle²

- In one-third of patients with oculodermal melanocytosis, the eye is not involved and a smaller group may present only with ocular involvement (ocular melanocytosis)³

- Etiology: excess melanocytes that have not migrated completely from the neural tube to surface structures during embryogenesis

- Most (50%) develop the nevus/pigmentation at or soon after birth, but it is not uncommon for pigmentation to appear around puberty or young adulthood¹

- Important sequelae of oculo(dermal) melanocytosis:
  - Glaucoma: elevated IOP occurs in approximately 10% of eyes⁴
    - May be caused by angle abnormalities due to neural crest anomalies or mechanical occlusion by melanocytes in an open angle
  - Malignant melanoma: Patients are at greater risk for development of intraocular and central nervous system malignant melanomas; malignant change, involving choroid, skin, iris, orbit or brain, occurs in 4.6% of all reported cases⁵
  - Patients with uveal melanomas associated with oculo(dermal) melanocytosis are at a two times risk for metastasis compared to those with no melanocytosis⁶

**TREATMENT/MANAGEMENT:** Patient and parent educated on the condition and increased risk of glaucoma in the future, change of conversion to melanoma, possible involvement of ears, mouth, or brain. Send report to PCP to recommend an ENT consult, MRI of orbits and surrounding tissues, and referral to UCSF Ocular Oncology Department to ensure earliest detection if any conversation to malignant melanoma occurs.

- Closely monitor for development of glaucoma and/or melanomas - early detection is key

**CONCLUSION:** Patients with oculo(dermal) melanocytosis are at an increased risk of developing glaucoma and intraocular malignant melanomas. For these reasons, early diagnosis is key and twice-yearly examinations with proper imaging should be the norm. Examinations should include a thorough inspection of the anterior segment with slit lamp photography and anterior segment OCT. Also, a thorough dilated fundus exam, including fundus photography, OCT and ultrasonography, should be performed to confirm the absence of a tumor. It is important to note, as seen in this case, ocular pigmentation may present without dermal involvement and therefore close attention must be paid to ensure a proper, early diagnosis.

**REFERENCES**