A Case of Nevoid Basal Cell Carcinoma Syndrome in a 9 month Infant
Jeffrey Scott Harbold Jr DO, Carlos Rivera MD, Rick Lin DO FAOCD FAAD

ABSTRACT
Nevoid Basal Cell Carcinoma Syndrome (NBCCS), also known as Gorlin syndrome is an autosomal dominant genodermatosis with a prevalence of 1 in 60,000 people. [1] It can present with a multiple clinical features including musculoskeletal malformations, CNS deformities, medulloblastoma, cardiac fibroma and basal cell carcinoma. Musculoskeletal deformations include jaw keratocyst, craniofacial abnormalities, palmar/plantar pitting and bifid ribs. [2]

Most clinical features manifest in adulthood and rarely present in the pediatric population. The diagnosis can be established with a combination of two major diagnostic criteria and one minor diagnostic criterion or one major and three minor diagnostic criteria (Table 1). [3] In cases where clinical features are inconclusive, sequence analysis of the PTCH1 gene can support the diagnosis. Management for these patients includes continuous surveillance and treatment of clinical manifestations.

CASE PRESENTATION
Our patient is a 9 month old Hispanic male with an evolving asymptomatic linear atrophic plaque on the flexor surface of the left wrist extending to the left upper arm. Lesion presented at birth and has recently progressed into a linear pattern towards the proximal arm.

Additional findings on exam includes cleft lip, craniomegaly with frontal bossing, a supernumerary digit of the right foot, and plantar pits. See Figures 3-6.

A 4mm punch biopsy was obtained and histology demonstrated findings consistent with a basaloid neoplasm. The patient was referred to pediatric dermatology where genetic PTC1 testing was performed and confirmed our suspicion of NBCCS. The patient was referred to pediatric neurosurgery were an MRI head was ordered to evaluate for medulloblastoma. Patient was also referred to ophthalmology and pediatric dentistry. The extra digit was excised by plastic surgery.

Due to the size of the lesion and the age of the patient, topical imiquimod was started. Strong emphasis was placed on future treatment with plastic surgery.

DISCUSSION
Management of NBCCS involves a multidisciplinary approach. It requires continuous screening, prevention and treatment of clinical manifestations. Prevention is done by wearing protective clothing in order to minimize UV light radiation exposure, regular use of sunscreen, avoiding excessive exposure to X-rays, and close follow up with a dermatologist for screening.

Referral to dentistry or oral surgery starting at age 8 is recommended every 12-18 months for jaw keratocyst screening, although prolonged X-ray exposure with the use of panoramic dental radiograph (Orthopantogram) should be limited due to increased risk of neoplasm.

Experts recommend performing a brain MRI when patients present with abnormal neurologic findings or changes in head circumference. [5] Surveillance for medulloblastoma is recommended every 4 months until age 3 and every 6 months until age 5. [4] Dermatologic screening is recommended annually until first BCC manifests, then every 6 months or sooner if needed. [3]

Treatment options for BCC in pediatric cases include topical 5-FU, imiquimod and surgical excision. The use of Mohs micrographic surgery is effective for early lesions and can be supplemented with cryotherapy and laser treatment. Radiotherapy is a feasible option when lesions are recurrent, or surgery is contraindicated. [6]

Patients who receive prompt treatment and recurrent screenings may have a favorable prognosis. [7]