Giant Congenital Sclerosing Blue Nevus of the Scalp Presenting with Rapidly Disseminated Fatal Metastases in a Pediatric Patient

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Sir:

Giant congenital blue nevi (GCBN) are rare dermal melanocytic proliferations that are distinguished by their large size, aggressive behavior, and propensity for malignant degeneration.[1] To date, only 18 cases of GCBN involving the scalp have been reported. Among these, local invasion into the calvaria/dura (n = 10), malignant transformation (n = 6), and disseminated metastases (n = 5) were observed.[2] Although the precise nature of malignant change is unclear, adults with histopathologically cellular GCBN are most commonly affected.[2] We report the first case of a sclerosing GCBN of the scalp presenting with rapidly progressive fatal metastases in a toddler.

A 17-month-old girl presented for evaluation of a 15×20-cm blue-gray plaque of the scalp that had grown commensurately with the child since birth. The patient's history was notable for posterior encephalocele repair during infancy, resulting in an occipital bone defect and chronic/stable ulceration at the vertex. Initial magnetic resonance imaging demonstrated a noninvasive parietooccipital mass with an underlying 12-cm^2 calvarial defect. Multiple incisional biopsies from ulcerated and nonulcerated regions revealed histopathologic features of sclerosing GCBN with extensive dermal/subcutaneous infiltration (ie, spindle-shaped melanocytes/dense collagenous stroma). Given these benign findings, conservative observation/local wound care was pursued in the hopes that dural-induced calvarial regeneration would improve the safety of tumor resection from the underlying scarred dura. However, the patient presented 7 months later with rapidly enlarging subcutaneous nodules, necrosis, and new-onset pain, vomiting, and ataxia (Fig. 1). Repeated magnetic resonance imaging demonstrated interval development of 2 sublesional masses with intracranial extension and calvarial/dural invasion (Fig. 2). A histopathologic diagnosis of melanoma (ie, atypical epithelioid melanocytes, large/pleomorphic nuclei, necrosis, numerous mitoses) arising within the areas reminiscent of sclerotic blue nevus was confirmed. Immunostains for BRAF V600 were negative; however, homozygous deletion of the 9p21 locus (CDKN2A/P16 tumor suppressor) was identified in 40% of cells analyzed by fluorescent in situ hybridization. Total body positron emission tomography revealed disseminated visceral metastases. The patient subsequently died 3 weeks later from widespread metastatic disease.

This case presents several unique findings that deviate from previously reported trends. Morphologically, the majority of GCBN display cellular histopathologic features, which correlate well with their proclivity for invasive growth and malignant degeneration.[1,2] In contrast, sclerosing blue nevi are predominantly benign, solitary, well-circumscribed nodules. Focal malignant melanoma arising within this subtype has been reported only once; however,

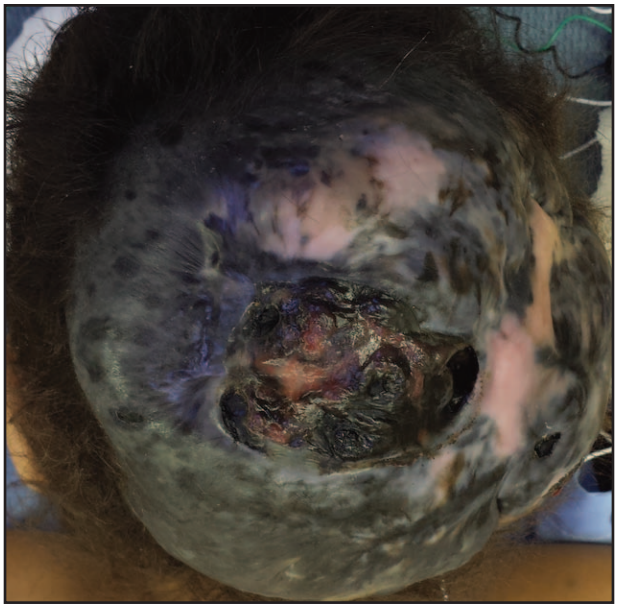

Fig. 1. Clinical appearance of the malignant sclerosing giant congenital blue nevus at the time of final presentation. Gross inspection reveals a large (15×20 cm) parietooccipital plaque with irregular contours, hyperpigmented macules, paracentral scar, and extensive necrotic ulceration at the vertex.

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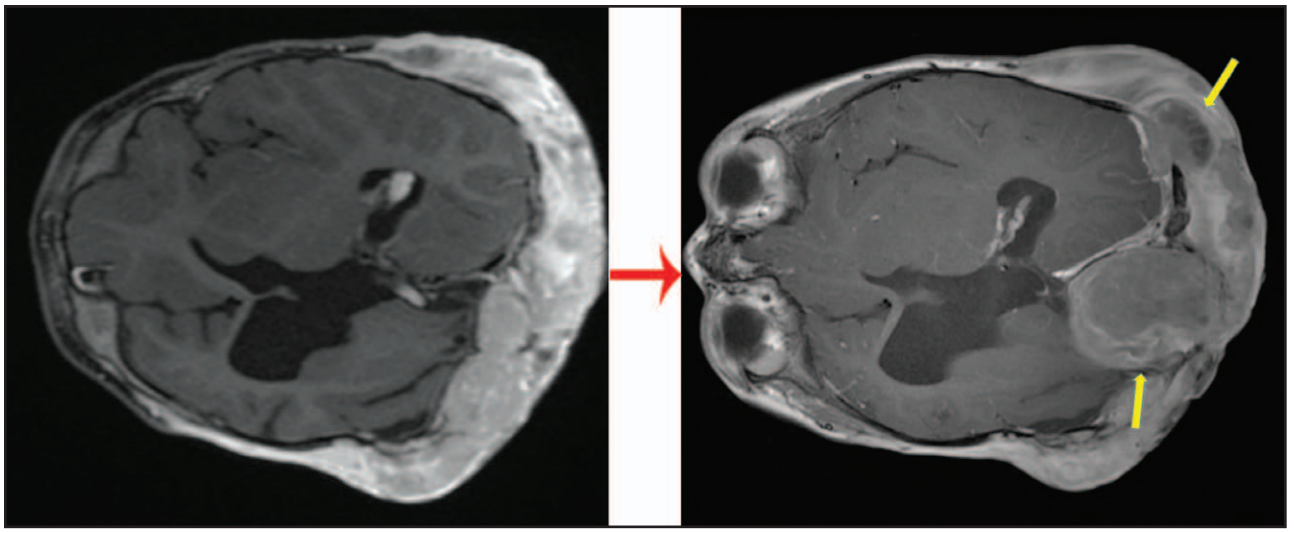

Fig. 2. Axial T1-weighted magnetic resonance image of the brain at the time of (left) initial presentation and (right) 7 months later demonstrating interval development of 2 invasive masses (arrows) along the deep aspect of the parietooccipital plaque. The larger extraaxial mass on the right (4 × 5 cm) is seen extending intracranially through the preexisting posterior calvarial defect, with involvement of the underlying dura and focal mass effect. A second, smaller mass (2 × 2 cm) is visualized on the left with parenchymal-sparing invasion through the calvarium. Stable encephalomalacia and dystrophic appearance of the right cerebral hemisphere are consistent with the previous resection of a parietooccipital encephalocele.

no association with GCBN or disseminated metastases was documented.[3] Furthermore, the onset of malignant change during childhood is exceedingly rare (n = 2) and may signify a more aggressive clinicopathologic phenotype.[2,4] Diagnosis in our case was made on the basis of rapidly progressive metastatic disease and malignant histopathologic findings. Although cytogenetic analyses have been advocated for histologically equivocal tumors, their utility remains limited in the absence of established criteria for malignancy. Interestingly, however, homozygous deletion of the 9p21 locus—as observed in our patient—is detected in a high percentage of locally invasive/metastasizing melanomas.[5] Although the significance of this finding in GCBN has not been validated, it may indicate a more clinically aggressive form of malignant transformation.

Currently, guidelines for the management of GCBN do not exist. When feasible, early prophylactic removal should be considered before extensive local invasion and/or metastasis precludes curable resection. In all cases, vigilant follow-up and biopsy of any new lesions are mandatory, and parents should be counseled regarding the significant, yet unpredictable, risk of malignant transformation. Further data are needed to determine the optimal management strategy, including adequate resection margins and indications for sentinel node biopsy and systemic adjuvant therapy in these rare but highly morbid tumors.

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DISCLOSURE

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