Rapidly enlarging nontender lesion on a child's face

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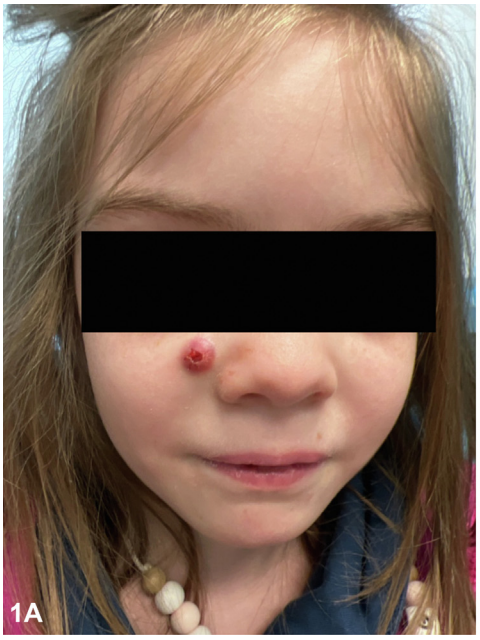

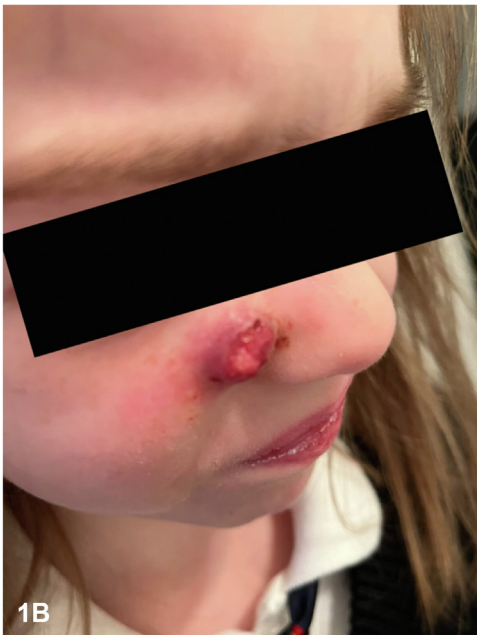

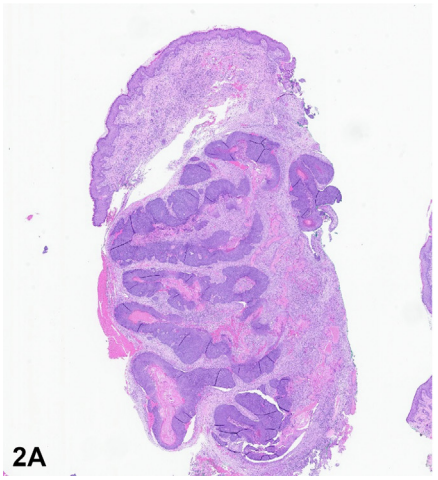

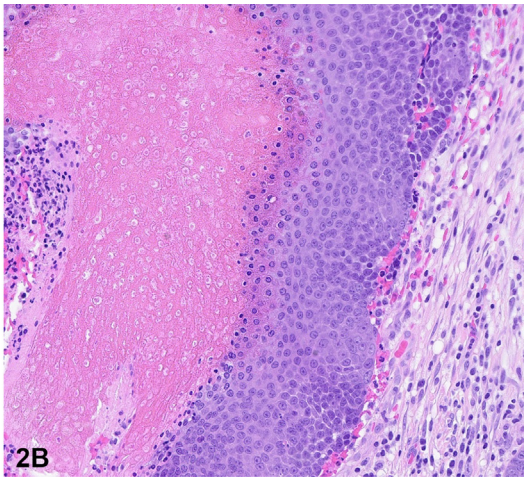

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CASE DESCRIPTION

An otherwise well 7-year-old Caucasian female presented to a dermatology outpatient clinic with a 2-month history of a rapidly enlarging non-tender nodule on the right cheek which measured approximately 1 cm. This patient had no medical history and was not on any medications. Two weeks later the lesion had grown to measure approximately 3 cm and significant erosion was noted (Fig 1). She was referred to plastic surgery for urgent excision. Representative image of histopathology is shown (Fig 2).

Question 1: What is the most likely diagnosis in this patient?

A. Spitz nevus

B. Pilomatricoma

C. Giant molluscum contagiosum

D. Amelanotic melanoma

E. Keratoacanthoma

Answers:

A. Spitz Nevus — Incorrect. Spitz nevi typically present in children as dome-shaped papules or nodules on the face and extremities, with colors ranging from pink, tan, to dark brown or black. Atypical spitz nevi can be very large with rapid growth, involvement of deep dermis and subcutis, and ulceration. Pathology demonstrates nests of epithelioid and/or spindled melanocytes.

B. Pilomatricoma — Correct. Pilomatricoma is a benign cutaneous adnexal tumor that originates from cells in the matrices of hair follicles. Typical lesions are round, mobile, firm, and surrounded by overlying skin. Pilomatricomas often exhibit calcification, causing them to feel hard or irregular upon palpation. Giant pilomatricomas are rare, greater than 5 cm in diameter, and are more common in females.[1] Histopathology shows characteristic findings of basaloid cells and eosinophilic "ghost cells."

C. Giant molluscum contagiosum — Incorrect. Molluscum contagiosum is a common childhood infection caused by poxvirus. Lesions typically present as multiple smooth papules with central umbilication. Giant molluscum is associated with HIV infection and immunosuppression. Henderson-Patterson bodies on histology are pathognomonic.

D. Amelanotic melanoma — Incorrect. Amelanotic melanomas typically appear skin-colored or erythematous and account for a significant proportion of childhood melanomas.[2] Pediatric melanoma is rare, and affected patients often have risk factors such as large/giant congenital nevi or genetic predisposition. Histopathology shows malignant melanocytes with decreased or complete absence of pigment.

E. Keratoacanthoma — Incorrect. Keratoacanthomas are cutaneous tumors which present as solitary nodules with a centralized keratin-filled crater. These tumors are more common in middle-aged or older adults but can present in childhood in the context of genetic disorders such as multiple self-healing squamous epitheliomas and xeroderma pigmentosum.

Question 2: What is the classical clinical sign for this lesion?

A. Darier's sign

B. Antenna sign

C. Tent sign

D. Auspitz sign

E. The ugly duckling sign

Answers:

A. Darier's sign — Incorrect. Darier's sign is seen in cutaneous mastocytosis. Positive Darier's sign is characterized by elicitation of urticaria on or around the affected area after scratching or rubbing the lesion, indicating the presence of mast cells.[3]

B. Antenna sign — Incorrect. Antenna sign is seen in keratosis pilaris. Upon examination with tangential lighting, lesions exhibit long strands of keratin resembling an antenna.[3]

C. Tent sign — Correct. Tent sign is classic for pilomatricomas and results from stretching of skin over calcification.[3]

D. Auspitz sign — Incorrect. Auspitz sign is seen in psoriasis. This sign is elicited when the scales are removed from a psoriatic plaque, resulting in pinpoint bleeding.[3]

E. The ugly duckling sign — Incorrect. The ugly duckling sign is used to describe cutaneous melanoma. Nevi on an individual tend to resemble one another (AKA "signature nevus"), and any atypical nevus raises suspicion for melanoma.[3]

Question 3: Genetic variation in which gene can be associated with this lesion?

A. PTCH1

B. HLA-B27

C. TYR

D. CARD14

E. CTNNB1

Answers:

A. PTCH1 — Incorrect. This is a tumor suppressor gene in the sonic hedgehog pathway. PTCH1 is a transmembrane protein that inhibits the release of smoothened. Germline mutations in PTCH1 are associated with nevoid basal cell carcinoma syndrome, and somatic mutations are associated with sporadic cases of basal cell carcinoma.

B. HLA-B27 — Incorrect. Genetic variation within this gene is associated with inflammatory conditions, including ankylosing spondylitis, inflammatory bowel disease, uveitis, pustular psoriasis, and psoriatic arthritis.

C. TYR — Incorrect. This gene encodes the enzyme tyrosinase which is pivotal in the synthesis of melanin. Mutations in this gene are associated with OCA1a and OCA1b forms of oculocutaneous albinism.

D. CARD14 — Incorrect. CARD14 is mainly expressed in keratinocytes, and gain of function mutations cause upregulation of proinflammatory genes that predispose an individual to the development of plaque and generalized pustular psoriasis. Mutations in this gene are also associated with familial pityriasis rubra pilaris.[4]

E. CTNNB1 — Correct. This gene encodes the protein beta-catenin, an important mediator in the Wnt/b-catenin signaling pathway. Somatic mutations in CTNNB1 are thought to cause unregulated proliferation of hair matrix cells, leading to the formation of pilomatricomas.[5]

Conflicts of interest

None disclosed.

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