A Bald Spot on an Infant's Scalp

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What's Your Diagnosis?

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Introduction. A healthy 4-month-old infant girl presented to the clinic with her mother after the mother noticed the infant scratching an itchy rash on her torso.

History. The infant was born full term with no complications. During the physical examination, a scalp lesion (Figure 1) was incidentally noted. The lesion was found to have a patch of excess hair surrounding the site. There were no other similar lesions on the skin. A papulosquamous eruption was seen on the trunk and treated as atopic dermatitis. The itchy rash was clinically consistent with atopic dermatitis and treated with a topical steroid. No atopic changes were noted on the scalp. The patient's mother stated that she was aware of the lesion on the scalp, which she was told was just a scar from a scalp monitor during labor.

Figure 1. *A lesion on the scalp of a 4-month-old infant is shown.*

Diagnostic testing. An ultrasound of the head did not reveal any bony defects or underlying malformations.

Correct answer: B. Aplasia cutis congenita

Aplasia cutis congenita (ACC) is a congenital defect defined by lack of the dermis, epidermis, and subcutaneous tissue primarily on the scalp vertex.[1] The diagnosis, and exclusion of differentials in this case, is primarily based on the infant's physical examination, which revealed excess hair surrounding the site. This "hair collar sign" is a potential indication of underlying malformation that warrants imaging, particularly ultrasound in those 4-6 months of age whose skull has not full developed. Although an MRI could have been performed, such imaging is more invasive and is generally performed for patients in their late teens or early 20s, when bones are more fully developed.

Differential diagnoses. The differential diagnoses in this case include alopecia areata (AA), congenital triangular alopecia, meningocele, which also clinically present as hairless plaques, as well as nevus sebaceous.

AA is an autoimmune dermatologic disorder that manifests as a non-scarring alopecia4 and primarily manifests early in life, with the majority of cases presenting before the age of 20.[4] The condition, which is exceptionally rare in neonates, manifests as sharply demarcated, localized patches of alopecia, but may be severe enough to involve the entire scalp or body (eg, alopecia areata universalis.)[4] Treatment for AA in pediatric patients includes topical, intralesional, or oral corticosteroids, minoxidil, light therapies, and, if needed, systemic therapies such as Janus kinase inhibitors.[4] A key clinical feature of AA is the presence of "exclamation point" hairs near the edges of bald patches [4] . This diagnosis was ruled out due to the presence of a hair collar sign, which is not seen in AA, and because AA is exceedingly rare in newborns [4,5]

CTA is a non-scarring, nonprogressive form of alopecia of unclear etiology, typically manifesting in early childhood as a triangular or spear-shaped patch of complete hair loss on the frontotemporal region of the scalp [5]. It is generally asymptomatic and remains stable over time [5]. While it may resemble other forms of alopecia, CTA lacks signs of inflammation or scarring [5]. We excluded CTA due to the presence of a hair collar sign and the round shape of the lesion, which differs from the characteristic triangular pattern of CTA, and is more characteristic of ACC [5].

The diagnosis of meningocele of the scalp was also considered. A meningocele is a type of neural tube defect that involves the protrusion of the meninges and cerebrospinal fluid through a spinal defect into the subcutaneous tissue.[3] The exact etiology of meningocele remains complex.[3] Physical examination typically reveals a visible midline swelling that transilluminates.[3] It often appears as a midline swelling on the scalp or spine that transilluminates and may be detected prenatally or at birth. Ultrasound, MRI, or X-ray are used to confirm the diagnosis by identifying the bony defect and meningeal protrusion [3]. A meningocele diagnosis would require immediate follow-up. Associated complications are,

namely, tethered cord syndrome, and rarely, hydrocephalus.[3] In our case, we ruled out meningocele because we did not find any bony defects or protrusions in the patient's ultrasound.

Nevus sebaceous is a rare congenital hamartoma that involves hyperplasia of epithelial, sebaceous, follicular, and apocrine structures.[2] It is typically diagnosed clinically due to its distinct appearance on physical examination, which involves plaque with partial or full alopecia, as well as an orange or hyperpigmented color generally found on the scalp or face.[2] Biopsy and histopathological evaluation is rarely done, but can reveal epidermal and adnexal changes such as epidermal hyperplasia and sebaceous gland hyperplasia.[2] Despite debate over optimal management, full thickness excision is often chosen for aesthetic concerns and to minimize rare risks of malignancy, which occur in approximately 3% of cases[2]. We excluded this condition due to the presence of a hair collar sign, which nevus sebaceous does not exhibit.

Treatment and management. The ultrasound showed no underlying malformation and the patient's mother received reassurance that further management was not indicated. The patient's mother was informed that an elective surgical excision of the area may be done in the pre-teen years if they desire removal for cosmetic reasons.

Outcome and follow-up. The favorable prognosis of the area, given the lack of underlying malformation, did not necessitate a follow-up appointment. Midline scalp lesions, the presence of a "hair collar sign", and vascular stains, can indicate underlying cranial or central nervous system involvement such as neural tube defects, cortical dysplasia, and intracranial arteriovenous malformations[2].

Discussion. ACC is a congenital dermatologic disorder marked by the absence of skin at birth, with subtypes categorized by lesion location, inheritance patterns, and associated malformations.[1] The condition most frequently presents as a solitary scalp lesion and can be linked to various abnormalities, including cranial or central nervous system involvement.[1] Although many ACC lesions heal spontaneously, certain features—such as a midline location, hair collar sign, or vascular markings—suggest possible soft tissue or life-threatening anomalies that warrant further investigation.[1] The hair collar sign, a patch of dark, thick hair in a ring like pattern around a scalp lesion, is a significant indicator of underlying conditions such as encephaloceles or meningocele, especially when observed with midline scalp lesions[7]. A previous study found that infants displaying this sign are at a high risk for central nervous system anomalies, including meningeal heterotopia, venous malformations, and midline malformations [7]. Cranial imaging, such as ultrasound for those 4-6 months old and MRI for those older, is recommended to evaluate and facilitate referral to appropriate pediatric specialists for comprehensive management.[8]

In this case, the mother brought the infant in due to concern over itching from what was later identified as atopic dermatitis. The scalp lesion was discovered incidentally during the exam. Most patients with ACC are asymptomatic, and the lesion may not be recognized until several months after birth.[1] Diagnosis typically relies on clinical findings alone to avoid unnecessary biopsy, which can risk complications if myelomeningocele is present. However, due to ACC's variable presentations, imaging techniques such as ultrasound or MRI can be useful to rule out associated bony, vascular, or neurological abnormalities when necessary.[6] Once ACC is diagnosed, management depends on lesion size and characteristics. Lesions under approximately 4 cm at birth generally heal through scar formation and normal wound healing, while larger lesions or those with underlying abnormalities may require surgical repair with skin grafting or flap techniques.[1] Antibacterial treatment can help prevent infection if the lesion remains open.

Conclusion. A 4-month-old healthy term girl presented with a clinically apparent hair collar sign. ACC was strongly considered and diagnosed through physical examination. Ultrasound imaging ruled out underlying malformations, yet it remains crucial to recognize the hair collar sign as an indicator of potential central nervous system anomalies, such as encephaloceles or meningoceles. Lesions located at the midline vertex or posterior occiput, particularly when accompanied by the hair collar sign, warrant careful evaluation.[7] Cranial and cerebral MRI in older kids, and ultrasound in those 4-6 months of age, is indicated in such cases to ensure early identification and appropriate management of any underlying neuroectodermal anomalies.[8] Clinicians should maintain a high index of suspicion for ACC with these features to prevent missing significant underlying abnormalities.

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