Congenital urticaria pigmentosa mistaken for non-accidental injury

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Abstract

Awareness of cutaneous manifestations of non-accidental injury (NAI) is essential for healthcare professionals working with children. Some cutaneous conditions may present with bruise-like lesions that can mimic NAI. We present the case of a male infant referred for investigation of NAI, who was subsequently diagnosed with cutaneous mastocytosis.

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A previously well male infant presented to his general practitioner (GP) for his routine 6-week check. His parents reported the presence of tan-brown lesions since birth. The marks were uniform in color, were present on the trunk, scalp, and face, and were not overlying bony prominences. The GP was concerned that these lesions represented bruising as a manifestation of NAI, and arranged urgent review at our pediatric department. Following assessment by the pediatric team he was admitted for investigation of potential NAI, and dermatology consultation was sought. On examination, scattered reddish brown to tan macules and papules were noted on the torso and limbs, not overlying bony prominences (Figure 1A, 1B). Darier sign was negative, although some reactive cutis marmorata was appreciable on the trunk following rubbing of multiple lesions. A diagnosis of urticaria pigmentosa (UP), or cutaneous mastocytosis, was made clinically.

Skin biopsies demonstrated an infiltrate of spindled and round mast cells and scattered eosinophils within the superficial and mid dermis, with immunohistochemistry positive for CD117, consistent with UP (Figure 2A-2C). Blood tests during admission showed a normal complete blood count and coagulation screen (including von Willebrand factor), normal renal and liver function, normal lactate dehydrogenase, normal immunoglobulins, and normal mast cell tryptase. A cranial ultrasound and liver ultrasound were normal.

Mastocytosis is a spectrum of disease characterised by pathologic accumulation of mast cells in tissues, including skin, bone marrow and the gastrointestinal tract. The World Health Organisation has outlined three variants: cutaneous mastocytosis, systemic mastocytosis, and mast cell sarcoma. Cutaneous mastocytosis can be further classified as maculopapular cutaneous mastocytosis, also known as UP; diffuse cutaneous mastocytosis; and localised cutaneous mastocytomas.[1] Paediatric cutaneous mastocytosis is regarded as a benign condition limited to skin, most commonly presenting as UP, which usually spontaneously regresses in puberty. [2] In contrast, adult-onset mastocytosis is more often associated with systemic involvement, a persistent course, and activating mutations of the *KIT* gene. [1]

With UP, children typically present in the first two years of life with multiple reddish-brown macules, papules and/or plaques distributed anywhere on the body, most commonly the trunk. In darkly pigmented skin, lesions may be hyperpigmented and erythema less appreciable. Darier sign, a transient urticarial response upon gentle rubbing of the skin, is classic and supportive of diagnosis, though not always demonstrable. Treatment with antihistamines can be helpful. [3] UP has rarely been reported to be misdiagnosed as NAI. [4, 5]Other 'bruising' mimics include disorders of coagulation, Valsalva petechiae, vasculitis, acute haemorrhagic edema, incontinentia pigmenti, phytophotodermatitis, coin rubbing, spooning, cupping, Mongolian spots, morphea, neuroblastoma, and ink stains, some of which may be unfamiliar to pediatricians or dermatologists.

Dermatologists have a crucial role in recognising cutaneous manifestations of NAI in children, which commonly includes bruising, particularly in infants who are not yet mobile. [4]While consideration of NAI is important in all children presenting for medical care, this case highlights the importance of considering underlying dermatoses to avoid unwarranted distress and unnecessary investigations.

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Figures

Figure 1A. Subtle isolated red-brown to tan macules present on the abdomen.

Figure 1B. A red-brown plaque overlying the left temple.

Figure 2A. H&E at 10x magnification of a skin biopsy from the back with a cellular infiltrate in the dermis composed of mast cells and scattered eosinophils.

Figure 2B. Higher power (40x magnification) H&E of the mast cells and eosinophils within the dermis.

Figure 2C. Immunohistochemistry for CD117 which stains mast cells and highlights the mast cell infiltrate in the dermis (magnification 20x).