Adult onset urticaria pigmentosa: A case report

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SUMMARY

Urticaria pigmentosa is the most common form of cutaneous mastocytosis, but the diagnosis is often missed due to the rarity. We report an adult-onset urticaria pigmentosa on a male with typical lesions, proven by Giemsa and CD117-immunocytochemistry staining. The patient responds well with appropriate oral H1 antihistamine and topical steroids.

INTRODUCTION

Urticaria pigmentosa (UP) is caused by abnormal mast cell accumulation in the skin, which might also accompany systemic symptoms such as flushing, itching, blisters, diarrhoea, abdominal pain, nausea, hypotension, headache and bone pain.[1] Systemic mastocytosis (SM) involves at least one extra-cutaneous organ, almost always involving bone marrow.[2] Urticaria pigmentosa is characterised by itchy hyperpigmented macules or papules usually found on the trunk but rarely on the scalp, palms of the hands and soles.[3] The incidence of UP is estimated at 3 to 7 per 1,000,000 population/year worldwide, and the prevalence in the population is difficult to determine because many cases are isolated and often undiagnosed.[1] Based on studies at the Karolinska Teaching Hospital and the Mayo Clinic, adult-onset cutaneous mastocytosis (CM) is an indolent variant of SM.[4] Based on patient visit data from the Allergy and Immunology Division of the Dermatology and Venereology Polyclinic, RSUP Prof. Dr. IGNG Ngoerah from January 2019 to January 2023 found only two cases of mastocytosis, all of which were CM.[5] The diagnosis of CM is made based on the clinical picture and histopathological examination.[4] Identification of SM needs to be done as early as possible to provide appropriate management and understanding of the risk of osteoporosis and anaphylactic reactions that may occur.[1,4]

CASE PRESENTATION

A 36-year-old man presented with red-brown patches on his face, chest, back and feet persisting for 6 months. The patches became itchy and thickened after being scratched or exposed to heat. There was no history of any systemic disease during childhood, and systemic symptoms were denied. Multiple well-defined, reddish-brown patches with varying sizes between 0.3 × 0.5 cm to 1 × 2 cm were found on the face, chest and back. A wheal develops after rubbing on an existing lesion (Darier's sign was positive) (Figure 1a). Dermoscopy examination (Figure 1b) shows a light-brownish blot (blue arrow) and pigment network (black arrow). We provisionally made a diagnosis of UP. We planned for a complete blood count, blood chemistry and skin biopsy. The complete blood count and blood chemistry are within normal limit. Histopathologic examination with hematoxylin and eosin (HE) stain revealed basketweave keratin pattern, an increased number of perivascular mast cells, and a preponderance of ovoid-spindle-round cells suggestive of mast cells (Figure 1c-f). Giemsa stain revealed positive granules (Figure 1g), and the CD117 stain revealed an increase of mast cells in perivascular at 400x magnification, shown as copper granules (Figure 1h).

He was treated with oral H1 antihistamine (cetirizine 10 mg daily) and topical steroid (0.25% desoximetasone cream twice daily) for 2 weeks, and he has achieved good control of his wheals and itch (Figure 2).

DISCUSSION

Mastocytosis is characterised by the expansion and accumulation of mast cells in one or more organ systems. The pathogenesis involved a c-kit mutation at codon 816 that causes amino acid substitution and excessive mast cell production.[6] Based on organ involvement, the World Health Organisation (WHO) differentiates mastocytosis into CM and SM. In CM, mast cell proliferation occurs in the skin, while SM involves at least one extra-cutaneous organ. It is typically observed that bone marrow involvement is strongly linked with SM.[4] Some triggering factors are food, exercise, heat, Hymenoptera and animal stings, skin trauma, alcohol and drugs.[7] In this case report, the exact trigger is unknown.

Diagnosis of UP is based on the clinical history, physical examination and supporting examination. Multiple macules and patches on the trunk and extremities with Darier's sign are the pathognomonic signs of this disease.3 Darier's sign is an urtication with erythematous halo that produced by rubbing on the lesions. A dermacoscopic examination shows light brown blots, a pigment network and a vascular pattern.[8] Light-brownish blot and pigment network seen with dermoscopy indicate hyperpigmentation of basal keratinocytes or papillary dermis caused by a high concentration of mast cell growth factor that stimulates melanocyte proliferation and melanogenesis. The classical histopathologic findings of UP are ovoid-spindle-round cells, indicating mast cells. In patients with cutaneous involvement, the only confirmatory diagnosis is skin biopsy, whereas SM is confirmed by bone marrow biopsy. Plasma

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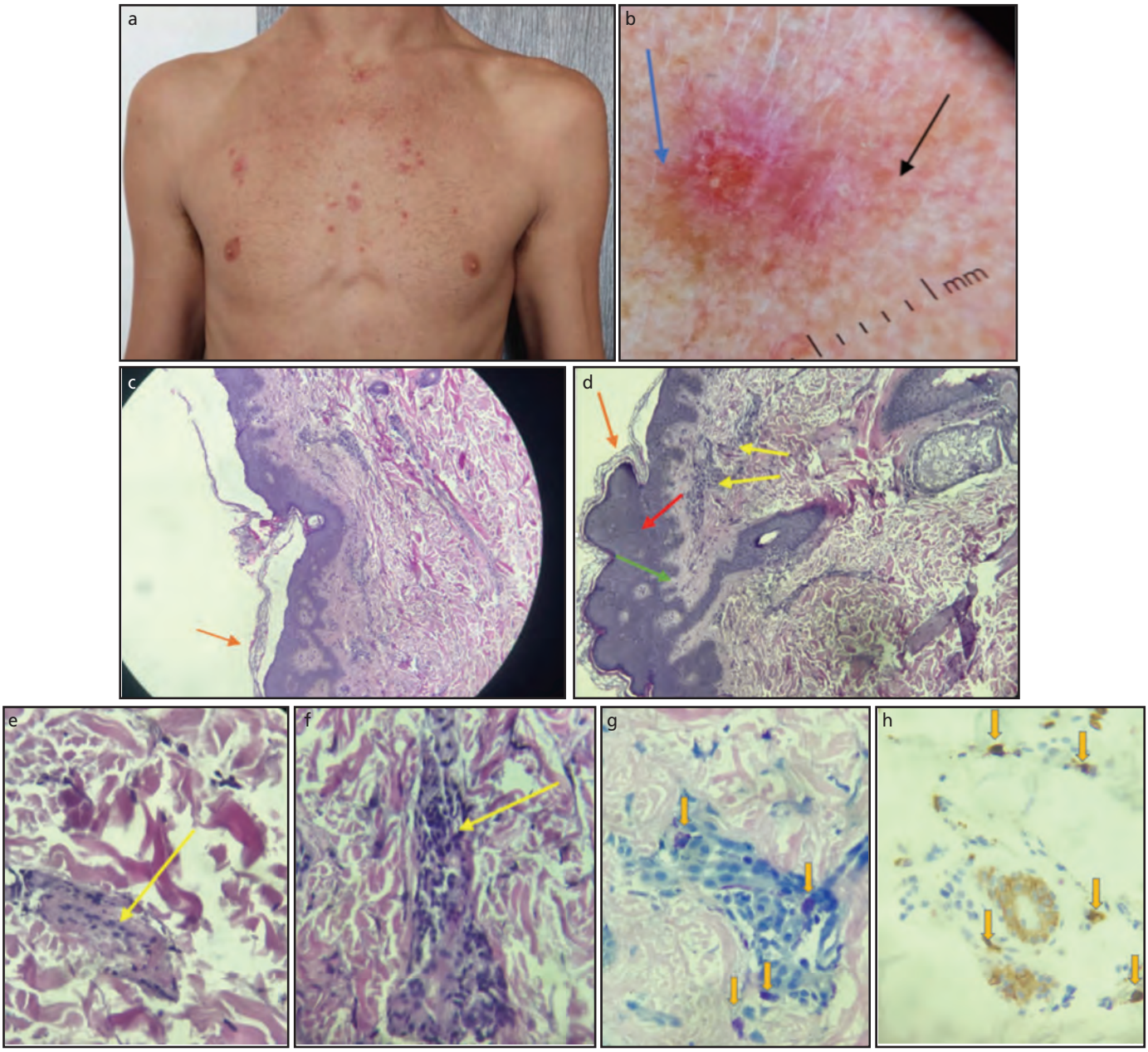


Fig. 1: a) Lesion on the chest. b) Dermoscopic finding. c-f) H & E stain indicates mast cell— g) Giemsa stain shows positive granules— h) CD117 stain shows copper granules.

tryptase examination is an alternative for those who reject bone marrow biopsy as it is a cell mast degranulation product. Total serum tryptase above >20 ng/mL becomes a minor criterion for SM. Skin biopsy followed with special stains such as Giemsa and CD 117 will reveal mast cells.[6] The diagnosis of UP in our case was based on the clinical picture and skin biopsy, however the possibility of SM could not be ruled out since tryptase examination is not available in Indonesia and bone marrow biopsy was not performed in this patient.

The main goal of CM management is to control symptoms related to mast cell mediators, which are based on histamine receptor antagonists.[3,6] The first-line treatment is the administration of first and second-generation H1 antihistamines.[2] If the patients have refractory symptoms, the H1 antihistamine dose may be increased, or other regimens such as H2 antihistamine, oral glucocorticoids, leukotriene inhibitors and monoclonal antibodies against immunoglobulin E can be used.[3,6] Oral disodium cromoglycate can also be given to treat gastrointestinal issues.[1,4] According to some studies, topical steroids can help improve skin lesions in UP. In this particular case, the patient was prescribed systemic AH-1 cetirizine 10 mg tablets, to be taken orally every 24 hours, along with topical anti-inflammatory 0.25% desoximetasone cream, to be applied topically every 12 hours. The treatment resulted in a good response.



Fig. 2: The lesion showed improvement after receiving treatment.

From a dermatological perspective, no ideal classification system can correlate CM lesion type with prognosis.[3] The outcome of UP is influenced by age, severity and clinical subtype. There is a lack of extensive data on the long-term monitoring of CM. The previous studies on CM showed that children had a favourable prognosis, with 50 to 60% improvement observed in adolescents. However, if CM persists into adulthood, there is a 10% risk of developing SM with a poor prognosis.[9] Our patient responded well to treatment. However, we are uncertain about their prognosis as we cannot rule out the possibility of SM.

CONCLUSION

Adult onset urticaria pigmentosa was diagnosed through history taking, physical examination and skin biopsy. Even though the treatment with H1 antihistamine and topical steroids has shown improvement, long-term observation is still needed.

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CONFLICT OF INTEREST

There is nothing to declare.

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