SKIN MANIFESTATIONS OF INTERNAL DISEASE

EXTENSIVE VULVAR CUTANEOUS MASTOCYTOSIS IN AN ADULT WITH NEUTROPENIA

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Background: Mastocytosis has been classified into cutaneous and systemic disease according to a World Health Organization consensus. Another classification scheme divides cases into pediatric and adult onset disease. In adults, cutaneous disease is usually accompanied by evidence of mastocytosis in other organs, and most experts agree that a bone marrow biopsy is recommended. We present an unusual case of vulvar cutaneous mastocytosis (CM) in an adult with progressively worsening neutropenia. Vulvar involvement in mastocytosis is unusual with a total of 15 cases reported worldwide. To our knowledge, this is only the second case of vulvar mastocytosis reported in an adult, and the first case with hematologic abnormalities.

Observation: A woman in her 60s was seen for a pruritic vulvar and perianal eruption for 6 months. She had been followed by hematology for neutropenia for 5 years. Examination revealed extensive firm skin to brown colored papules on the vulvar and perianal skin. Biopsy showed a dense dermal mast cell infiltrate that stained positive for c-KIT and tryptase. Her serum tryptase level was 29 ng/ml (normal < 11.5 ng/ml). Complete blood count demonstrated leucopenia and neutropenia. C-KIT D816V mutation was detected in the peripheral blood. Bone marrow biopsy was non-specific with <5% blasts, no mast cells, and normal cytogenetics. She was diagnosed by hematology to have clonal cytopenia of uncertain significance given that she failed to meet criteria for systemic mastocytosis. Treatment with potent topical steroids for 3 weeks resulted in near complete resolution of cutaneous lesions.

Key message: 1. Vulvar involvement is extremely rare in mastocytosis. It should be included in the differential diagnosis of firm vulvar papules in children and adults.
2. Adults with CM should have a workup to exclude systemic involvement including a hematologic assessment.