Denouement and Discussion

Urticaria Pigmentosa

Mastocytosis is a disorder characterized by mast cell accumulation in tissue most commonly in the skin, and it may also affect the bone marrow, gastrointestinal tract, skeletal system, liver, spleen, and lymph nodes. A World Health Organization consensus group[1] recently divided the disease into 7 forms: cutaneous mastocytosis, indolent systemic mastocytosis, systemic mastocytosis with an associated hematologic non–mast cell lineage, aggressive systemic mastocytosis, mast cell leukemia, mast cell sarcoma, and extracutaneous mastocytoma. Systemic mastocytosis is mostly an adult phenomenon.

CLINICAL FEATURES

Urticaria pigmentosa (UP), the most common type of mastocytosis, is defined by multiple red-brown hyperpigmented macules, papules, or nodules.[2] The most recent studies estimate its incidence to be 1 in 150 000.[1] In 55% of patients, UP begins before age 2 years, and in 10% of patients it occurs between ages 2 and 15 years. Onset for the remaining 35% of patients occurs between ages 20 and 40 years. Urticaria pigmentosa most often presents on the trunk, and when rubbed, these lesions have a tendency to urticate, forming a wheal and flare reaction known as the Darier sign, which is considered to be clinically diagnostic.[2]

Pruritus is the most frequent initial symptom in pediatric-onset mastocytosis.[1] Several years after the diagnosis of UP, the lesions may increase in number; however, pediatric-onset UP has a favorable prognosis, with spontaneous resolution occurring in 50% of children by adolescence.[3,4] The accumulation of mast cells in organs other than the skin may result in systemic symptoms, such as vomiting, diarrhea, nausea, bone pain, headache, dyspnea, flushing, irritability, or abdominal pain.[3,5]

The c-kit receptor has a central role in the biological development of mast cells in adult-onset disease. Only children with persistent or progressive versions of the disease express the c-kit mutation, suggesting a different and unknown pathogenesis for cases in children.

DIAGNOSIS

The diagnosis of UP is based primarily on the appearance of skin lesions together with a classic history of pruritus and the Darier sign. Although a skin biopsy was performed in this case, most cases do not need a biopsy to confirm the diagnosis. A skin biopsy specimen may be obtained showing hyperplasia of mast cells.[3] To rule out associated hematologic diseases and systemic involvement, a complete blood cell count and a peripheral smear may be considered.[1] Other possible laboratory measurements include the levels of serum tryptase, plasma histamine, and urinary histamine metabolites. Specific symptoms should guide further workup.[1] Physicians may mistake these cases for atypical bruises or café au lait spots. The differential diagnosis for UP also includes generalized eruptive histiocytoma, Langerhans cell histiocytosis, and non–Langerhans cell histiocytosis of childhood.

TREATMENT

In general, treatment of UP is conservative and aimed at symptomatic relief.[6] In some patients, topical corticosteroids may be applied to reduce pruritus and unsightliness of lesions, oral antihistamines may be used to reduce pruritus, and cromolyn sodium therapy may ameliorate gastrointestinal tract symptoms.[6] Parents and patients should be advised to avoid precipitating causes of mast cell degranulation (insect stings, sudden changes in temperature, certain medications [alcohol, aspirin, codeine, morphine, nonsteroidal anti-inflammatory drugs, and others], certain foods [alcohol, shellfish, some cheeses, spicy foods, and hot beverages], and friction).[1,2] Although not all patients will see histamine release with all of these triggers, it is helpful to educate families about these precipitating factors. Some patients and their parents worry about the theoretical risk of anaphylaxis in cases of significant mast cell degranulation. Although this is a rare occurrence, an epinephrine autoinjector (EpiPen Jr or EpiPen; Dey LP, Napa, California) can be prescribed as a precautionary measure.

This patient's lesions will likely remain active for another 5 years or so and will eventually fade, with perhaps some residual dyspigmentation. The patient's mother was informed of precipitating causes of degranulation and was given an epinephrine autoinjector (EpiPen Jr) and was instructed to follow up in approximately 6 months.

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