

P1104

Umbilical cord blood transplantation can cure pruritic eczematous eruption in a hypohidrotic ectodermal dysplasia with immunodeficiency patient

Satoko Minakawa, MD, PhD, Department of Dermatology, Hirosaki University Graduate School of Medicine, Hirosaki, Aomori, Japan; Daisuke Sawamura, MD, PhD, Department of Dermatology, Hirosaki University Graduate School of Medicine, Hirosaki, Aomori, Japan; Eturou Ito, MD, PhD, Department of Pediatrics, Hirosaki University Graduate School of Medicine, Hirosaki, Aomori, Japan; Hajime Nakano, MD, PhD, Department of Dermatology, Hirosaki University Graduate School of Medicine, Hirosaki, Aomori, Japan; Hajime Takeda, MD, PhD, Department of Dermatology, Hirosaki University Graduate School of Medicine, Hirosaki, Aomori, Japan

Anhidrotic ectodermal dysplasia and immunodeficiency (EDA-ID) is an X-linked recessive genodermatosis characterized by severe eczematous eruption, hypohidrosis, dental anomalies, alopecia, and immunodeficiency. A 4-month-old Japanese boy with pruritic eruption on his entire body since birth was referred to our clinic. Examination revealed diffuse erythema and reddish papules, with scratching evident on most of the body, which was compatible with atopic dermatitis. A skin biopsy specimen revealed spongiotic change in the epidermis and perivascular lymphoid and eosinophilic infiltration in the superficial dermis. Results of appropriate laboratory tests confirmed deficient cellular immunity. A pathogenic mutation c.1167insC was identified in the nuclear factor-kappaB (NF- κ B) essential modulator (NEMO) genes in both the patient and his mother. His mother had been diagnosed with incontinentia pigmenti by skin biopsy a few months after her birth. His maternal uncle died from a disorder of unknown etiology when he was 1 month old. His maternal grandmother and grandmother's sister were suspected to have had incontinentia pigmenti from their histories. He was diagnosed as EDA-ID from clinical features, family history, and mutation analysis of the NEMO gene. The patient was treated with hydrocortisone cream and a moisturizing agent, which resulted in minimal improvement of eczema and intense pruritus. When he was 3 years old, he received an umbilical cord blood transplantation. After the transplantation, his itchy eruption was gradually getting better. Nonpurpuric erythema developed on the trunk at day +35, and the biopsy specimen showed liquefaction in the dermoepidermal interface with satellite cell necrosis, compatible with acute GVHD. His eczematous eruption had almost disappeared, leaving slight xerosis a year after the transplantation. The responsible gene for EDA-ID is the NEMO gene, which is critical for the activation of NF- κ B signaling involved in inflammation, immune responses, and cell survival.

In male patients, the hypomorphic mutations, in which NEMO function is decreased but not abolished, generally result in EDA-ID, while larger frameshift and deletion mutations which completely impair NEMO function cause a lethal condition in male fetuses. This family had one nucleotide insertion mutation of c.1167insC, which might sustain some function of the NEMO gene, as previously reported.

Commercial support: None identified.

P1105

Scleromyxedema with neurologic involvement: Therapy with intravenous immunoglobulin

Filipa Ventura, Dermatology and Venereology Department, Hospital de São Marcos, Braga, Portugal; Celeste Brito, Dermatology and Venereology Department, Hospital de São Marcos, Braga, Portugal; Margarida Rodrigues, Neurology Department, Hospital de São Marcos, Braga, Portugal; Maria da Luz Duarte, Dermatology and Venereology Department, Hospital de São Marcos, Braga, Portugal

Scleromyxedema is a rare idiopathic disorder characterized by dermal mucin deposition with fibrosis associated with monoclonal gammopathy and systemic manifestations. Despite some reports of success with numerous agents, there is not a completely satisfactory therapeutic approach to the scleromyxedema. A 69-year-old male, previously healthy, presented in a coma without fever or meningeal signs to the emergency department. He underwent a full neurologic workup and the EEG showed slow and irregular background activity. The lumbar puncture, MRI and laboratory studies were normal. He was prescribed valproate, cefotaxime, ampicillin, acyclovir, and dexamethasone with a slow improvement. He was discharged without complaints on the fourteenth day. Six months later, the patient presented a widespread symmetric eruption of tiny waxy monomorphic papules predominating in the forehead, forearms, and hands. The physical examination also revealed a diffuse erythema and induration of the skin, leading to decrease the motility especially of the mouth and the hands. A skin biopsy specimen revealed a diffuse mucin deposit in the dermis and a marked fibroblastic proliferation. The most relevant results of the hematologic investigation were IgG monoclonal gammopathy with λ light chains and absence of thyroid disease. The diagnosis of scleromyxedema was made. The patient was treated with thalidomide 100 mg/day. After 6 months, there was a discrete improvement, but the treatment had to be stopped because of neurologic complaints. In the meantime, he suddenly had a loss of consciousness and he came back to the hospital in a comatose state. This episode was similar to the previous one. A 5-day course of intravenous immunoglobulin (IVIG; 400 mg/kg/d) was started and his encephalopathy improved steadily. After completing 6 cycles of treatment, there was a dramatic skin lesions improvement. We present this case because (apart of being rare) this case reports an encephalopathy associated with scleromyxedema and the effectiveness of IVIG in treatment.

Commercial support: None identified.

P1106

Eruptive vellus hair cysts treated with lactic acid: Case report and review of the literature

Bryant Tran, Wake Forest University School of Medicine, Winston Salem, NC, United States; Andrew Lee, MD, Wake Forest University School of Medicine, Winston Salem, NC, United States; Ashley Curtis, MD, Wake Forest University School of Medicine, Winston Salem, NC, United States; Gil Yosipovitch, MD, Wake Forest University School of Medicine, Winston Salem, NC, United States

Because eruptive vellus hair cysts were first identified 3 decades ago, treatment for this condition remains unsatisfactory and time-consuming. We describe a 41-year-old African American female who presented with a 3-month history of a progressive, pruritic eruption on her upper chest and neck that was characterized by multiple dome-shaped, hyperpigmented papules. Skin biopsy of a lesion on her chest revealed cysts lined by squamous epithelium and containing laminated keratin and numerous vellus hair shafts. Oral antihistamines and topical corticosteroids did not relieve her pruritus nor affect the appearance of the lesions. A trial of topical 12% lactic acid resulted in modest improvement in the appearance of the lesions. Topical lactic acid is an effective option for eruptive vellus hair cysts in richly pigmented patients, because surgical options can produce scarring and dyspigmentation.

Commercial support: None identified.

P1107

A retrospective review of the use of colchicine in chronic idiopathic urticaria

Lana Pho, MD, University of Utah, Salt Lake City, UT, United States; Christopher Hull, MD, University of Utah, Salt Lake City, UT, United States; Doug Powell, MD, University of Utah, Salt Lake City, UT, United States; Mark Eliason, MD, University of Utah, Salt Lake City, UT, United States; Michelle Regruto, MD, University of Utah, Salt Lake City, UT, United States

Background: Chronic idiopathic urticaria (CIU) is a common condition that can be debilitating, difficult to treat, and sometimes life-threatening. The disease is common with a lifelong prevalence of 0.5% to 1%. Recent reports have shown that the disability suffered by CIU patients is similar to that of patients with coronary artery disease. The treatment of CIU patients can be frustrating, and for those who do not respond to antihistaminic treatment, other treatment modalities are needed. In many patients, immunosuppressant medications are required, but these have major adverse effects, such as renal dysfunction, liver function abnormalities, and anemia. A safer and more efficacious therapy is clearly needed for CIU.

Aims: To evaluate efficacy and side effects of colchicine in patients with CIU.

Methods: Chart review of patients with diagnosis of CIU based on history, physical examination, and skin biopsy at the University of Utah between 2002 and 2007.

Results: A total of 36 patients who met criteria for CIU and were treated with colchicine were identified. Twenty-eight (78%) were women and the mean \pm SD age was 46 ± 15 years. Mean \pm SD duration of the CIU was 41 ± 63 months. Fifty-eight percent had a history of angioedema. Maximum dose of colchicine achieved was 1.5 mg/day with the majority of patients taking 1.2 mg/day. Forty-seven percent were on treatment for at least 1 month and 42% maintained treatment from >1 to 22 months. Subjective clinical response to therapy reported as partial ($n = 5$) or complete ($n = 15$) were found in 55% of patients. Although 39% ($n = 14$) reported adverse effects (diarrhea being the most common), only 11 patients (31%) stopped treatment. Of the 15 patients who had complete clearance, eight patients continue to be asymptomatic, two patients had recurrence after finishing treatment and switched to other agents, three patients stopped treatment secondary to side effects, and two patients had recurrence but responded to a second course of colchicine.

Conclusions: Colchicine is an effective treatment in 55% of our patients. In patients who had complete response, eight (53%) patients remained clear after tapering of colchicine. Side effects were mild. These results support the use of colchicine for patients with CIU unresponsive to standard therapy. Larger, controlled trials will further investigate colchicine for use in CIU patients.

Commercial support: None identified.