1. **Adolescent with Deforming Facial Swelling and Fatal Course**
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3. **CDSN Mutation in Siblings with Inflammatory Peeling Skin Syndrome with Systemic Manifestations**
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4. **Bilateral Blastomycosis-like Pyoderma Exacerbated by DKA**
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5  Febrile ulceronecrotic Mucha-Habermann disease in a patient with T-cell ALL
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7  Proliferative Nodule Absent Underlying Congenital Melanocytic Nevus: An Unusual Neonatal Neoplasm
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   Dan Miller, MD – University of Minnesota
   Andrew Larson, DO – University of Minnesota
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8  Annular Lipoatrophy of the Ankles
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   Ruth Ann Vleugels, MD, MPH – Brigham & Women’s Department of Dermatology
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9  Menkes disease presenting as possible child abuse
   Jillian Rork, MD, Karen Wiss, MD, Leah Belazarian, MD – University of Massachusetts Medical School
Blood, Tears, But No Sweat: An Extraordinary Case of Hematohidrosis
Amy Thorne, DO – University of New Mexico School of Medicine
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**Title of Presentation**

Adolescent with Deforming Facial Swelling and Fatal Course

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**Case Summary**

We report a 17 year-old Hispanic male with a five year history of deforming facial swelling with crusting. Initial review of systems was negative and studies including CBC, CMP, ACE, C3, C4, C1inh, type III hereditary angioedema testing, herpes DFA, and chest x-ray were within normal limits. MRI/MRA of head and neck demonstrated only soft tissue swelling. Three biopsies of the face and mucosal lip were interpreted to have mild granulomatous perivascular infiltrates associated with marked dermal edema, felt consistent with orofacial granulomatosis (OFG). Oral doxycycline, minocycline, and ibuprofen had minimal effect on swelling. Further therapy was deferred until he completed workup to assess for inflammatory bowel disease (eventually negative) and granulomatous contact dermatitis (never completed).

Thirteen months ago, the patient developed fever, malaise, hepatitis, myositis, and petechial patches x 4 on the extremities and buttock, with episodes of difficult swallowing and oral/airway swelling. Skin biopsy showed interface dermatitis, but muscle and liver biopsy showed nonspecific inflammation. We considered possible connective tissue disorder instead of OFG, while rheumatology considered unusual sarcoidosis, and a trial of oral prednisone was initiated with near resolution of systemic symptoms. He had notable partial improvement in his facial swelling with this combined with monthly infliximab. However, four months ago, he developed a cribiform indurated nodule at the right forearm that became necrotic, requiring debridement and antibiotics, with poor healing over three months. Oral cyclosporine was added for potential pyoderma gangrenosum. Several weeks later, he presented with two other nodules on the right upper extremity and rapid deterioration with multiorgan failure. RSV testing was positive and bone marrow biopsy revealed hemophagocytic lymphohistiocytosis (HLH), likely secondary to EBV infection in the setting of immunosuppression. Meanwhile, re-review of his right forearm lesion showed extranasal NK/T-cell lymphoma. He was started on etoposide, decadron, IVIG and gangciclovir but unfortunately ultimately died from HLH. We present this case given his unusual presentation and to discuss the potential role of EBV.

**References**


Atypical mycobacterium infection in three sisters

Case Summary

Three sisters, ages 12, 15, and 17 years old, presented with similar violaceous and erythematous fluctuant nodules scattered on the trunk and extremities. The lesions were first noted on the two younger sisters approximately 4-5 months before presentation to a pediatric dermatologist in February 2015. Many of the 12 year old child’s (Patient 1) lesions developed in areas previously treated with liquid nitrogen for molluscum contagiosum. The 15 year old child (Patient 2) described her lesions as small, erythematous papules that became scabbed and then rapidly increased in size, became tender, and drained purulent material. In March, the oldest sister (Patient 3) had two excisions performed on benign lesions, a nevus and a lipoma. By the end of April, she developed purulent drainage within one excision site. The family denied contact with fish tanks or having pedicures in a salon. They do have a hot tub and a whirlpool bath. Examination of Patients 1 and 2 revealed fluctuant, violaceous, asymptomatic nodules up to 2 cm in diameter and red to purple thin papules and macules (Figures 1 to 4).

Patients 1 and 2 were initially referred to a pediatric surgeon for excisional biopsy in April. Some of the biopsies revealed necrotizing granulomatous inflammation (Figures 5-10). Tissue cultures for bacteria and fungi were negative, and acid fast bacilli (AFB) stains were negative. AFB culture of Patient 2 was positive at 13 days, Patient 1 was positive at 18 days, and Patient 3 is still pending. Speciation of their isolates demonstrated Mycobacterium fortuitum. AFB cultures of their home bath and shower were positive at 1 day with speciation pending.

M. fortuitum is a ubiquitous organism present in soil and water. It is an opportunistic pathogen that is often associated with trauma, surgical procedures, and immunodeficiency, but has been reported in immunocompetent individuals (1). Outbreaks of infections have been reported after cosmetic procedures (2-3). Single cases have been reported after amateur tattooing, leg waxing, various medical procedures, and unknown exposures (4-8). To our knowledge, there have been no reported cases secondary to likely antecedent trauma and home water supply contamination.

In summary, we present three sisters who presented with multiple violaceous nodules which eventually grew the atypical mycobacterium M. fortuitum. We assert that the source was contaminated home water supply.

References

Title of Presentation | CDSN Mutation in Siblings with Inflammatory Peeling Skin Syndrome with Systemic Manifestations | Case Number | 3

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Case Summary
We present siblings with inflammatory peeling skin syndrome (IPSS) due to a homozygous CDSN mutation, highlighting features of this multi-system disorder.

Case 1
The older of the two siblings, an 8 year old female presented with severe AD/generalized ichthyosis, eosinophilic esophagitis, failure to thrive requiring gastric-tube placement, iron-deficiency anemia, multiple food allergies and dilated cardiomyopathy. She is the product of consanguineous union (first cousins) from the Republic of New Guinea, without a known family history of skin disease. She has an unaffected brother. She was born erythrodermic. Laboratory evaluation has been remarkable for persistent eosinophilia (27%) and IgE of 1,700. Bone marrow biopsy was normal and skin histology showed epidermal thickening and mild, thickening of the stratum corneum. Eyebrow hair mount was normal.

Case 2
The younger sister, 5 years old, presented with erythroderma at birth and developed severe atopic dermatitis, localized ichthyosis, eosinophilic esophagitis, multiple food allergies, and gastric-tube dependence. Laboratory evaluation has been remarkable for persistent eosinophilia (19%), IgE of 86,900. BM biopsy was normal. Skin biopsy showed basket weave orthokeratosis and focal parakeratosis. Eyebrow hair mount was normal.

Initial genetic evaluation revealed expected long contiguous stretches of homozygosity in multiple chromosomes given consanguinity, and targeted analysis of SPINK5 revealed heterozygous single nucleotide variant in both sisters. Whole exome sequencing revealed a novel homozygous c.85G>C, p.29G>A mutation in CDSN in both sisters, consistent with IPSS.

Discussion:
Peeling skin syndrome is a rare group of clinically and genetically heterogeneous cornification disorders. It is characterized by shedding of the stratum corneum at birth or early infancy and is classified as generalized or acral. Generalized PSS is further classified into non-inflammatory (type A) and inflammatory (type B). Type B [OMIM #270300] is due to a recessive mutation in CDSN which resulting in loss of corneodesmin function. It manifests as an ichthysiform erythroderma with lifelong patchy peeling of the skin that presents at birth or in the first weeks of life. Additional clinical features include severe AD and pruritus, food allergies, elevated IgE levels, eosinophilia, failure to thrive, recurrent skin infections, recurrent angioedema, urticaria and asthma. There is a strong phenotypic resemblance between PSS type B and Netherton’s syndrome, due to shared pathobiology featuring excessive desquamation. Several treatment modalities have been utilized for with limited efficacy.

References
**Title of Presentation**  
Bilateral Blastomycosis-like Pyoderma Exacerbated by DKA

**Case Number**  
4

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**Case Summary**

HPI: We present a 15 year old African American female with a history of type 1 Diabetes Mellitus, admitted for diabetic ketoacidosis at our Children’s hospital. Dermatology was consulted to evaluate bilateral pretibial lesions. The lesions had been present for 6 years, and started shortly after swimming in a public pool with possible trauma. Since then, they have grown slowly. These lesions were pruritic, and occasionally tender. Of note, with episodes of hyperglycemia, the lesions would develop purulent drainage. She had been afebrile and remaining review of systems was negative.

Exam: On exam she was a young female in no acute distress. On the right pretibial plateau was a 15cm x 5cm gray-brown hyperpigmented verrucous plaque with purulent drainage and overlying crust. Though initially nontender, reinspection in 1 hour revealed exquisite tenderness, followed by copious purulent drainage and ulceration of the lesion.

Labs: Initial labs were significant for a WBC count of 41.84, an initial blood glucose of 586, and HbA1C% of 11.7%. Biopsy was performed for H&E and tissue culture. Tissue and wound cultures showed S. aureus, and group B strep with no fungal or acid-fast bacilli growth. Blood cultures were drawn and found negative. Imaging was negative for osteomyelitis. Biopsy showed a markedly inflamed ruptured epidermal cyst and follicles with a dense dermal lymphoepithelial infiltrate and overlying pseudoepitheliomatous hyperplasia. PAS, GMS, AFB and Giemsa stains were negative. No granules consistent with a Splendore-Hoeppli phenomenon were observed.

These findings were consistent with a diagnosis of blastomycosis-like pyoderma (pyoderma vegetans). Patient was placed on trimethoprim-sulfamethoxazole and discharged home with scheduled follow-up.

Discussion/Review: Blastomycosis-like Pyoderma, also known as Pyoderma Vegetans, is a rare cutaneous bacterial infection that often masquerades as other fungal, inflammatory or neoplastic disorders. There are fewer than 70 reported cases. It is most commonly due to infection with S. aureus and can occur on the face, scalp, axilla, trunk, and distal extremities. Predisposing factors include immunosuppressed states such as diabetes mellitus, poor nutrition, HIV, malignancy and alcoholism. Lesions are typically verrucous hyperkeratotic purulent plaques with raised borders. Histological examination reveals pseudoepitheliomatous hyperplasia with neutrophilic abscesses. Though caused by bacteria, response to systemic antibiotics is variable. Other treatment modalities include dapsone, systemic and intralesional corticosteroids, retinoids, carbon dioxide laser and excision.

**References**

Febrile ulceronecrotic Mucha-Habermann disease in a patient with T-cell ALL

A 5-year-old girl with T-cell ALL was transferred for a progressive, ulcerative, necrotic rash involving 80% of her body. The rash began 10 months prior as faint pink patches that “looked like bruises.” T-cell ALL was diagnosed 5 weeks later. The skin cleared with induction therapy (AALL0434: cytarabine, vincristine (VCR), daunorubicin, prednisone, pegaspargase, and MTX). The eruption fluctuated during subsequent chemotherapy, then acutely worsened after its completion. Prior to transfer, she was febrile did not respond to steroids, cyclophosphamide, and weekly IVIG 2g/kg x 3 weeks. Skin biopsies revealed a perivascular and interface dermatitis composed of predominantly small mature lymphocytes that was negative for blasts. The most recent biopsy showed an infiltrate of CD3+, CD8+, TCR-beta+, TdT- T-cells. Clonal T-cell gene rearrangement and stains for EBV, VZV, and PAS were negative. The biopsies were consistent with pityriasis lichenoides et varioliformis acuta, favoring a diagnosis of febrile ulceronecrotic Mucha-Habermann disease (FUMHD). One biopsy showed clonal TCR gene rearrangement which could represent infiltration by leukemic cells. Infectious workup was negative. On transfer, low-intensity chemotherapy for ALL and FUMHD treatment was started (MTX 15 mg/week, dexamethasone, mercaptopurine, VCR, and azithromycin). IVIG 1g/kg x 2 was added but the skin did not improve. A mediastinal mass was noted, and biopsy confirmed relapsed ALL. The risk of infection due to skin breakdown prevented chemotherapy intensification. The soluble IL-2 receptor level was markedly elevated and basiliximab (12 mg/m2/week x 4 weeks), a chimeric antibody against the α-chain of the IL2 receptor (CD25), was added. The ulcers began healing gradually, chemotherapy was advanced, and she is awaiting bone marrow transplant.

To our knowledge, this is the first reported case of FUMHD associated with ALL. FUMHD has been reported in <70 patients, mostly males <20 years old (1). Its etiology is unknown, but some cases have been associated with infection or monoclonal T-cell proliferations. Given its fluctuation mirroring ALL activity and the biopsy with monoclonal T-cells, this case may represent a paraneoplastic process. FUMHD presents with erythematous scaly papules that progress to necrotic ulcers and may be fatal (2). Systemic findings include fever, arthralgia, megaloblastic anemia, pancytopenia, DIC, pneumonia, myocarditis, and GI and CNS symptoms. There is no standard treatment, though MTX is often used (3). This patient’s improvement with basiliximab may suggest use of anti-CD25 drugs as a novel therapy for refractory cases of FUMHD.

References
### Title of Presentation

Psoriasiform necrotic circinate plaques, dysmoria and mid-line defects

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### Case Summary

We present a 21-month old female with multiple congenital malformations and a psoriasiform skin rash. At birth she was found to be dysmorphic with anorectal malformation with rectovestibular fistula, craniosynostosis and cleft palate. After surgery procedures at 6 months of age she developed a periorificial eczematous rash, diffuse alopecia and scarce eyebrows. The skin rash extended to form circinate psoriasiform plaques that disseminated to extremities and developed an advancing necrotic border and honey-colored keratotic crusts, (fig. 1,2). The skin biopsy reported a psoriasiform dermatitis with parakeratosis and superficial necrosis, as well as keratinocyte atypia of the lower two thirds of the epidermis. Keratinocytes were very large, pleomorphic, with several mitoses (fig. 3,4). We gave the possible diagnosis of biotinidase deficiency and requested a ketone and organic acid profile on urine, which were normal. Blood levels of biotinidase enzyme are still pending. The patient was started on biotin supplements. We believed this case might have a partial biotinidase deficiency, but in view of the little response to biotin she had, we would like to present it at SPD for other diagnostic and management options.

Biotinidase deficiency is one of the most rare congenital metabolic disorders. Dermatological manifestations included alopecia, loss of hair color, hypopigmentation, seborrheic dermatitis and eczematous and erythematous perioral and perianal papules. 2 Our patient had alopecia, seborrheic dermatitis and eczematous and erythematous perioral and disseminated patches that developed after a major surgery. Mock et al. suggest that marginal maternal biotin deficiency causes fetal biotin deficiency, and they speculate that the fetal malformations are primarily the consequence of fetal biotin deficiency. 3 This could explain our patient’s multiple congenital defects including craniosynostosis, cleft palate and anorectal malformation. When biotinidase deficiency is suspected, urine ketones and organic acids as well as blood biotinidase, carnitine and acylcarnitine profiles should be evaluated; however in patients with partial enzyme activity no abnormality may be found. 1 We believe our patient could be suffering from partial biotinidase deficiency, which confers the risk of developing the same manifestations as those in children with a profound deficiency. However, symptoms present in correlation with metabolic stressors, and until then, children may be asymptomatic. 4 We would like to present this case for orientation and advice from fellow pediatric dermatologists because so far she has not had a good response to biotin and we are not sure how to proceed.

### References

## Case Summary

A healthy 10-month-old female was referred to pediatric dermatology for evaluation of an expanding nodule on the left wrist. The nodule was asymptomatic, present at birth and had been suspected to represent an infantile hemangioma. Its initial appearance was of a small papulonodule, and reportedly it had grown in proportion to the patient. A full body skin examination was significant for a 3 x 2 centimeter, lilac-colored, telangiectatic, rubbery nodule on the left dorsal wrist. An MRI of the left upper extremity showed only cutaneous involvement without deeper extension of the tumor. Punch biopsy demonstrated a dermally-based tumor of small to medium sized ovoid cells with monomorphic nuclei. The nuclei had fine granular chromatin and prominent nucleoli. Tumor cell lineage was uncertain. S100 and Melan-A immunostains were diffusely positive, confirming a melanocytic origin. While no mitotic figures were identified on light microscopy or pHH3/Melan-A double stain, Ki-67 labeling was elevated, at 10-15% of lesional cells. p16 stain did not show significant loss of that tumor suppressor gene product. A diagnosis of benign proliferative nodule absent an identifiable conventional congenital nevus was favored. Excisional biopsy with 5mm surgical margins demonstrated identical findings and also identified a trace, secondary population of junctional melanocytes suggestive of congenital melanocytic nevus. Comparative genomic hybridization (CGH) results are pending.

Proliferative nodules (PNs) are secondary melanocytic proliferations which typically arise in large or giant congenital melanocytic nevi (CMN). We were unable to identify any previous reports in the literature of PN in the clinical absence of a congenital nevus. Differentiating PNs from congenital melanoma is often difficult as these two entities may resemble each other clinically and histopathologically. There are two classic clinical patterns of PN presentation: small dermal nodules (less than 1cm in diameter) that are often present at birth and large dermal nodules (greater than 1cm in diameter) with initial rapid growth and occasional ulceration.

In instances where the malignant potential of these secondary proliferations is in question, CGH can be a beneficial test which provides objective data regarding biologic behavior. In the case of PNs, the CGH profile may demonstrate gains or losses of whole chromosomes, whereas melanomas tend to have multiple partial chromosome abnormalities.

In regard to our patient, margins were focally positive on our excision, raising some concern for possible recurrence.

## References

Case Summary

Lobular panniculitis is rarely reported in children and can pose a diagnostic challenge. We report an 8-year-old boy with a history of alopecia universalis who presented with marked atrophy of the lower legs and right foot following the spontaneous development of lower extremity edema over the prior year. During the period of swelling, he endorsed pain, weight loss, and weekly fevers, with labs notable for elevated acute phase reactants and mild anemia. Examination revealed annular, coalescing indurated plaques with raised, erythematous borders on the bilateral lower legs and right plantar foot, as well as prominent lipoatrophy surrounding the calves and ankles. Biopsies from the right calf demonstrated a striking lobular panniculitis with a robust inflammatory infiltrate composed predominantly of foamy histiocytes, but also of scattered lymphocytes, neutrophils, and eosinophils as well as focal fat necrosis. Special stains were negative for organisms. Collectively, the clinical presentation and histology were consistent with annular lipoatrophy of the ankles. Also known as lipoatrophic panniculitis and lipophagic panniculitis of childhood, annular lipoatrophy of the ankles is a rare form of localized lipoatrophy, manifesting as tender, erythematous, radially enlarging annular plaques on the lower extremities. After the initial inflammatory stage resolves, residual areas of marked lipoatrophy remain, which can mimic various lipodystrophies. A histologic diagnosis of exclusion, the clinical differential includes trauma, foreign body reaction, infection, connective tissue disease, vasculitis, pancreatic panniculitis, alpha-1 antitrypsin deficiency, and panniculitis-like T-cell lymphoma. The few reported cases of annular lipoatrophy of the ankles suggest a strong association with autoimmune diseases, including thyroiditis, type 1 diabetes, and alopecia areata, as in our patient. While often self-limited, early treatment should be considered to minimize permanent end-stage lipoatrophy. Given the rarity of this entity, treatment data are scarce; reported therapies include systemic corticosteroids, antimalarials, potassium iodide, methotrexate, and dapsone. Our patient received methotrexate 25 mg SC weekly and prednisone 10 mg PO daily, tapered over two months, with rapid resolution of the active borders and no disease progression. Despite this success, impressive residual lower extremity lipoatrophy persists. Our case demonstrates that combining systemic corticosteroids and methotrexate may effectively treat the inflammatory stage of this rare entity, thereby halting irreversible lipoatrophy, and hence warrants prompt recognition and intervention.

References

Menkes disease presenting as possible child abuse

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Case Summary

A 5-month-old male infant presented to the emergency department for cough and shortness of breath. A chest x-ray was performed which revealed a healing right posterior seventh rib fracture. Child Protection Services was contacted and a skeletal survey showed irregularity of the bilateral distal radial, ulnar and femur metaphyses concerning for healing fractures. He was also found to have prominent wormian bones along the lambdoid sutures (Figure 1). An initial evaluation for metabolic bone disorders was negative. The infant was taken into state custody for concern of child abuse.

Genetics was consulted to rule out a genetic condition given the multiple fractures and wormian bones. There was also concern for abnormal facies and developmental delay as the infant was not rolling over. Past medical history was significant for prematurity at 35 weeks gestation and prolonged hypothermia. He had no reported abnormal movements concerning for seizures.

On physical examination, his cheeks were pudgy with a cupid's bow of the upper lip and horizontal eyebrows. His hair was short, coarse, and white with darker distal tips. His skin was fair and lax with increased wrinkling on the abdomen and thighs (Figure 2). Pediatric Dermatology was consulted and light microscopy of the hair showed pili torti. The diagnosis of Menkes disease was proposed and confirmed by low levels of serum copper and ceruloplasmin. Custody was restored to his parents.

Menkes disease is an X-linked recessive neurodegenerative disorder of copper transport caused by mutations in ATP7A. Infants typically develop normally until 2-3 months of age and then gradually manifest with failure to thrive, developmental delay, hypothermia, hypotonia, and seizures. Milder forms have also been reported with similar cutaneous and musculoskeletal findings, but fewer neurologic disturbances (1). The presented patient showed developmental delay, but otherwise no neurodegenerative process, thus likely contributing to delayed diagnosis.

Since copper-dependent enzymes are pervasive, Menkes disease affects multiple organ systems. Several clinical findings can easily be misinterpreted as child abuse and may be the presenting features. These include subdural hematomas and musculoskeletal changes such as wormian bones, cervical spine defects, rib fractures, and spurring of the long bone metaphyses (2-5).

Fortunately, careful inspection of the hair and skin should facilitate correct diagnosis even with milder phenotypes and avoid undue accusations. Early diagnosis is also imperative since subcutaneous copper-histidine may be helpful if started before the onset of significant neurological symptoms.

References

Blood, Tears, But No Sweat: An Extraordinary Case of Hematohidrosis

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Case Summary

A 10-year-old girl presented with bloody discharge from the eyes and was treated for conjunctivitis. Over the next months, she had three to 35 “episodes” per day of blood-tinged discharge from her eyes, ears, axilla, mouth, rectum, vagina, urethra, scalp and different locations of the skin over the neck, trunk, and extremities. She reported occasional “disturbing” sensations in the sites prior to bleeding, including flushing, warmth, and fullness, but she was never in acute distress. The episodes occurred during both sleep and activity, with multiple adults and family members, and in multiple locations including home, school, and public places. The patient had a negative medical and family history. She was on no medications and was a high achiever in school. Numerous specialists were consulted including pediatric hematology, ophthalmology, otolaryngology, gynecology, neurology, cardiology, and dermatology.

Laboratory testing and imaging including a CT head and transabdominal ultrasound were normal. Hemostasis screen showed adequate intrinsic and extrinsic clotting factor and platelet activity. Blood-tinged fluid was collected during active expulsion while the patient was hospitalized. A forensic hematopathology analysis found that indeed the fluid contained the child’s blood. A surface culture of the skin was negative for Serratia marcescens. Skin biopsy immediately after an episode showed collections of erythrocytes surrounded by loose collagen fibers in the superficial dermis, not within a vessel or lymphatic space. A diagnosis of hematohidrosis was confirmed.

Hematohidrosis is an enigmatic condition in which a young girl exudes small amounts of watery, blood-tinged translucent fluid from the skin and mucous membranes. Accounts abound in art and literature, such as Homer’s Iliad, of persons “sweating blood,” as do historical and journalistic reports of religious stigmata appearing on a person’s skin. Only seven cases exist in the modern medical literature. Etiopathogenesis remains unclear; however, case authors agree that the fluid is not actually sweat but is “sweat-like” and that the purported cause is a gross derangement in the autonomic nervous system, but where the fluid originates and how it percolates to and exits the skin remains a mystery. Patients improved with use of diazepam and propranolol. Our patient improved on propranolol with a slow decrease in the number and frequency of episodes. In conclusion, we present the first case in the United States of both mucous membrane and skin hematohidrosis and the second biopsy demonstrating transient dermal defects with blood-filled spaces.

References