47TH ANNUAL SOUTHEASTERN CONSORTIUM FOR DERMATOLOGY CONFERENCE

OCTOBER 4-6, 2024—BOAR'S HEAD RESORT, CHARLOTTESVILLE, VA

ATTENDEE GUIDE

Hosted by

UNIVERSITY of VIRGINIA

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47TH ANNUAL SOUTHEASTERN CONSORTIUM FOR DERMATOLOGY CONFERENCE

Dear Colleagues,

Welcome to lovely Charlottesville, Virginia and the 47th Annual Meeting of the Southeastern Consortium for Dermatology (SEC). Our focus is diseases of the old and the young, with a special emphasis on scientific output from UVA alumni. This year marks UVA Dermatology's 100th year, founded in 1924 as the first dermatology department in the South. Our SEC planning team, the Board of Directors, and our meeting managers have worked tirelessly to bring you an exciting and educational program. We are thrilled to have in-person patient viewing for the first time since the COVID-19 pandemic; we hope to see you Sunday morning for that session.
We are also particularly grateful to our Lund lecturer, Dr. Kelly Cordoro (a UVA alumnus!), and our EP Cawley lecturer, Dr. Brittany Craiglow. For our alumni joining us, we look forward to celebrating with you at the alumni reception Friday evening.
Welcome to Charlottesville and enjoy the weekend!

Sincerely,

R Hal Flowers MD
UVA Dermatology
SEC 2024 Meeting Chair

UNIVERSITY of VIRGINIA

ABOUT

CHARLOTTESVILLE, VA

Welcome to Charlottesville, nestled at the foothills of the Blue Ridge Mountains. The city was founded in 1762 and is well known as the home of Thomas Jefferson and his UNESCO World Heritage estate Monticello as well as his university, UVA. The city itself features the historic downtown area, a charming brick-paved pedestrian mall, with opportunities for dining, shopping and entertainment. Check out some of our recommendations for some of the best eats in the area.

The surrounding area offers numerous opportunities for hiking, biking, exploring and wine and beer tasting. Nearby Shenandoah National Park and Skyline Drive are popular destinations for visitors, providing stunning vistas and outdoor adventures throughout the year. Consider Humpback Rock for a challenging but brief hike with a stunning overlook.

We hope you enjoy your stay!

UVA DERMATOLOGY

As UVA Dermatology celebrates 100 years as a department, we reflect on the changes and growth that have happened over the past century. Founded initially as the Department of Syphilology and Dermatology in 1924 by Dr. Dudley Crofford Smith, UVA was the first dermatology department in the South. Initial efforts were focused on public health, particularly around detecting and treating syphilis. Following Dr. Smith's passing in 1950, the department was re-vitalized with a renewed emphasis on research by Drs. EP Cawley and Clayton Wheeler. The second half of the 20th century saw growth in the number of clinical faculty and an emphasis on clinical services and education, which remain the cornerstones of UVA Dermatology. Chairs and leaders like Drs. Peyton Weary, Kenneth Greer, Tom Cropley, Barbara Wilson, Art Saavedra and Barrett Zlotoff have grown our department, increased the number of residents, expanded fellowship and subspecialty programs and strengthened the reach of influence in the community. Dr. Lu Le now serves as the Kenneth Greer Endowed Chair of the department and brings a vision of sustained excellence, research expansion and departmental growth to UVA Dermatology.

As we round out our first century as a department, the University of Virginia Dermatology remains committed to distinction in resident career development and mentorship, patient care, research, and community service. Thank you for joining us to celebrate this occasion.

ABOUT

THE SOUTHEASTERN CONSORTIUM FOR DERMAOLOGY

The first meeting of the Southeastern Consortium for CME in Dermatology (SEC) was in 1977. The educational goal of the annual meeting is an update in one or two areas of clinical dermatology selected from a core curriculum. The meeting rotates every year among the participating institutions. The participating Institutions include:

- Duke University
- Emory University
- University of North Carolina at Chapel Hill
- University of Alabama at Birmingham
- University of Virginia at Charlottesville
- Wake Forest University
- Virginia Commonwealth University
- Medical College of Georgia
- Medical University of South Carolina
- Eastern Virginia Medical School

SCHEDULE

The meeting agenda & more can also be found on our website.

Friday, October 4, 2024	
Time	**Description**
11:25 am – 12:35 pm	Registration Open & Lunch *Pavilion Lobby* Visit with Exhibitors *Ballroom (Main Inn)* Poster Presentations *Ednam Hall*
12:35 pm – 12:40 pm	Welcome *Pavilion* *Hal Flowers, MD*
Lecture Session 1 - UVA Dermatology	*Pavilion*
12:40 pm – 1:00 pm	Why Does Hair Turn Gray and How Can We Reverse It? *Lu Le, MD, PhD*
1:00 pm – 1:20 pm	The Importance of Mentorship: Cross Sections in Career Development and Personal Life *Art Saavedra, MD, PhD, MBA*
1:20 pm – 1:40 pm	Surgical Hacks for the General Dermatologist *Darren Guffey, MD*
1:40 pm – 2:00 pm	Melanoma: Increases in Surveillance Intensity and Diagnosis. Bug or Feature? *Robert Swerlick, MD*
2:00 pm – 2:10 pm	Panel Discussion
2:10 pm – 2:40 pm	Break & Visit with Exhibitors *Ballroom (Main Inn)* Poster Presentations *Ednam Hall*
Lecture Session 2 - Geriatric Dermatology	*Pavilion*
2:40 pm – 3:00 pm	Geriatric Dermatology: Taking the Best Care of Your Oldest Patients *Nikki Levin, MD, PhD*
3:00 pm – 3:20 pm	Managing Hidradenitis Suppurativa in Older Adults *Christopher Sayed, MD*
3:20 pm - 3:40 pm	Vulvar Disease in the Elderly: Assessing an Unmet Need *Jodi Ganz, MD, FAAD*
3:40 pm - 3:50 pm	Panel Discussion
3:50 pm - 4:05 pm	Break & Visit with Exhibitors *Ballroom (Main Inn)* Poster Presentations *Ednam Hall*
Lecture Session 3 - UVA Dermatology	*Pavilion*
4:05 pm – 4:25 pm	History of UVA Dermatology *Tom Cropley, MD*
4:25 pm - 4:45 pm	Then and Now: 1971-2024 *Ken Greer, MD*
4:45 pm - 5:05 pm	Skin Cancer in the Young and Old: 2024 Updates *Mike Marchetti, MD*
5:05 pm - 5:15 pm	Panel Discussion
5:15 pm - 6:00 pm	**EP Cawley Lecture -** Pediatric Alopecia Areata *Brittany Craiglow, MD*
6:00 pm – 7:00 pm	**Welcome Reception** *Ballroom (Main Inn)* *This event is available to Meeting Attendees only. No guests.*
6:00 pm – 7:00 pm	**UVA Alumni Celebration** *Albemarle, Blue Ridge & Commonwealth (Main Inn)* *This event is available to UVA Alumni & Guests only.*

SCHEDULE

The meeting agenda & more can also be found on our website.

Saturday, October 5, 2024	
Time	**Description**
7:00 am – 7:55 am	Breakfast *Pavilion Lobby* Visit with Exhibitors *Ballroom (Main Inn)* Poster Presentations *Ednam Hall*
Lecture Session 4 - Pediatric Dermatology	*Pavilion*
8:00 am - 8:20 am	Alpha Gal Syndrome, History & Clinical Overview *Jeff Wilson, MD, PhD*
8:20 am - 8:40 am	Pediatric Cutaneous Lymphoma Updates *Jakki Junkins-Hopkins, MD*
8:40 am - 9:00 am	Gender & Sexual Diversity Across the Lifespan: A Primer for Dermatologists *Klint Peebles, MD, FAAD*
9:00 am - 9:20 am	When It's Not Atopic Dermatitis *Leslie Lawley, MD*
9:20 am - 9:30 am	Panel Discussion
9:30 am - 10:00 am	Break & Visit with Exhibitors *Ballroom (Main Inn)* Poster Presentations *Ednam Hall*
Lecture Session 5 – Geriatric Dermatology	*Pavilion*
10:00 am - 10:20 am	Bullous Pemphigoid and Aging: Pathogenesis and Therapeutic Challenges *Russell Hall, MD*
10:20 am – 10:40 am	Pruritus in the Elderly *Shawn Kwatra, MD*
10:40 am – 11:00 am	Lessons Learned on the Inpatient Dermatology Consult Service *Paul Schneiderman, MD*
11:00 am – 11:10 am	Panel Discussion
11:15 am – 12:10 pm	**Lund Lecture – Beauty and the Beast: Challenging Nevi in Children** *Kelly Cordoro, MD*
12:10 pm – 1:10 pm	**Board of Directors Lunch** *Albemarle & Blue Ridge (Main Inn)* *This event is available to SEC Board of Directors only.*
6:00 pm – 8:00 pm	**Faculty Dinner** *Albemarle, Blue Ridge, & Commonwealth (Main Inn)* *This event is available to Faculty, Speakers, and SEC Board of Directors only.*
6:00 pm – 8:00 pm	**Resident's Reception** *Kardinal Hall* *This event is available to Residents only. Residents from all institutions are welcome.*

Sunday, October 6, 2024	
Time	**Description**
7:00 am – 7:55 am	Breakfast *Pavilion Lobby*
7:55 am – 9:45 am	Live Patient Viewing *UVA Health Orthopedic Center Ivy Road* *Buses for Live Patient Viewing will leave from outside of Ballroom at 7:45am and will continue on a loop.*
10:00 am – 12:00 pm	Case Presentations & Discussions *Pavilion*

LOCATIONS & EVENTS

Conference Venue

Boar's Head Resort
200 Ednam Dr., Charlottesville, VA 22903
October 4-6, 2024

REGISTRATION DESK
Pavilion *(Across from Main Inn)*

Registration Desk Hours	
Friday:	11:25 am – 6:00 pm
Saturday:	7:00 am – 1:00 pm
Sunday:	7:00 am – 12:00 pm

GENERAL SESSION
Pavilion *(Across from Main Inn)*

POSTER PRESENTATIONS
Ednam Hall *(Across from Main Inn)*

EXHIBIT HALL
Ballroom *(Main Inn)*
Exhibit Hall closes at 1pm on Saturday

WELCOME RECEPTION
Ballroom *(Main Inn)*
Friday at 6pm

UVA ALUMNI EVENT
Albemarle, Blue Ridge & Commonwealth
(Main Inn, upstairs from Ballroom)
Friday at 6pm

RESIDENTS RECEPTION
Kardinal Hall
722 Preston Ave #101, Charlottesville, VA 22903
Saturday at 6 pm

LIVE PATIENT VIEWING
UVA Health Orthopedic Center
2280 Ivy Road,
Charlottesville, VA 22903
Sunday from 7:55 - 9:45am

Meeting Information

COMPLIMENTARY WI-FI

Wireless Network: Boar's Head Guest Wifi
No Password Needed

PARTICIPATE IN THE EXHIBITOR ACTIVITY!
Complete the exhibitor activity card by having a conversation with each exhibitor and receiving a signature on each box. Once completed, turn into the registration desk by 1:00 pm on Saturday for the chance to win a DERMATOSCOPE! Winner will be announced during Sunday's general session.

CLAIMING CME CREDIT
An attendance and evaluation survey will be sent via Survey Monkey following the conference. In order to receive CME credits, you must complete the survey. Once the survey is complete, you will receive an electronic certificate. If you have any questions regarding the credit process, please contact the CME office at 757-446-6140.

SPEAKERS

Thank you to this year's speakers for their time and presentations.

** indicates UVA Alumnus*

Kelly Cordoro, MD*
Professor of Dermatology and Pediatrics
Division Chief and Fellowship Director,
Pediatric Dermatology
McCalmont Family Endowed Professor
in Pediatric Dermatology
University of California, San Francisco

Brittany Craiglow, MD
Associate Adjunct Professor
Yale School of Medicine

Tom Cropley, MD*
Professor of Dermatology and
Chair Emeritus
University of Virginia School of Medicine,
Department of Dermatology

Jodi Ganz, MD, FAAD*
Dermatologist
Olansky Dermatology and Aesthetics,
Atlanta GA

Ken Greer, MD*
Emeritus Professor; Rick A. Moore
Professor of Dermatology
Former Chair of Dermatology
University of Virginia School of Medicine,
Department of Dermatology

Darren Guffey, MD*
Director of Mohs Surgery
Commonwealth Dermatology

Russell Hall, MD
J Lamar Callaway Professor of Dermatology
Duke University School of Medicine,
Department of Dermatology

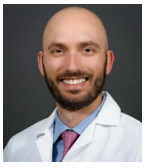
Jakki Junkins-Hopkins, MD*
Dermatologist; Dermatopathologist
Consultant in Cutaneous Lymphoma
Geisinger Medical Center

Shawn Kwatra, MD
Dr. Joseph W. Burnett
Professor and Chair
University of Maryland School of
Medicine, Department of Dermatology

Leslie Lawley, MD
Associate Professor of Dermatology
and Pediatrics
Emory University

Lu Le, MD, PhD*
Kenneth E. Greer, M.D. Endowed
Professor and Chair
University of Virginia School of
Medicine, Department of Dermatology

Nikki Levin, MD, PhD*
Professor of Dermatology
University of Massachusetts
Chan Medical School



Mike Marchetti, MD*
Dermatologist
Skagit Regional Health

Klint Peebles, MD, FAAD
Dermatologist
Kaiser Permanente, Mid-Atlantic
Permanente Medical Group

SPEAKERS

Thank you to this year's speakers for their time and presentations.

** indicates UVA Alumnus*

Art Saavedra, MD, PhD, MBA*
Dean of the School of Medicine,
Executive Vice President for
Medical Affairs
Virginia Commonwealth
University, School of Medicine

Christopher Sayed, MD
Professor of Dermatology
University of North Carolina at
Chapel Hill Department of Dermatology

Paul Schneiderman, MD*
Clinical Professor of Dermatology, Retired
Columbia University Medical School

Robert Swerlick, MD*
Professor & Alicia Stonecipher Chair of
Dermatology, Chairman Emeritus
Emory University

Jeff Wilson, MD, PhD*
Assistant Professor of Medicine,
Division of Allergy and Immunology
University of Virginia School of Medicine

Onsite Materials

Access Digital Attendee Guide & Speaker Talks Here!

CME INFORMATION

TARGET AUDIENCE

This program is designed for dermatologists, physician assistants, residents, and other healthcare providers specializing or interested in dermatology.

OBJECTIVES

- Analyze hair graying and potential reversal techniques, understanding the biological mechanisms and exploring emerging treatments that may reverse or slow down this process.
- Describe surgical techniques that can be applied in a general dermatology practice, including innovative "surgical hacks".
- Manage common dermatological conditions in elderly patients, focusing on comprehensive care approaches.
- Explore the latest updates in pediatric dermatology, including the management of pediatric alopecia areata and cutaneous lymphoma.
- Discuss the unique dermatological needs across different populations, including gender and sexual diversity and the challenges faced in geriatric and pediatric care.

ACCREDITATION STATEMENT

This activity has been planned and implemented in accordance with the accreditation requirements and policies of the Accreditation Council for Continuing Medical Education (ACCME) through the joint providership of Macon and Joan Brock Virginia Health Sciences at Old Dominion University and Southeastern Consortium for Dermatology. Macon and Joan Brock Virginia Health Sciences at Old Dominion University is accredited by the ACCME to provide continuing medical education for physicians.

DESIGNATION STATEMENT

Macon and Joan Brock Virginia Health Sciences at Old Dominion University designates this live activity for a maximum of ***12.25 AMA PRA Category 1 Credits™***. Physicians should claim only the credit commensurate with the extent of their participation in the activity.

HOW TO CLAIM YOUR CME CREDIT AND CERTIFICATE

An attendance and evaluation survey will be sent via Survey Monkey following the conference. In order to receive CME credits, you must complete the survey. Once the survey is complete, you will receive an electronic certificate. If you have any questions regarding the credit process, please contact the CME office at 757-446-6140.

SUMMARY OF FACULTY DISCLOSURES

The following presenters have disclosed the following relevant financial relationships with ineligible companies:

- Brittany Craiglow, MD - Consultant for Abbvie, Dermavant, Incyte, Lilly, Pfizer, Regeneron, Sanofi/Genzyme, and Sun Pharmaceuticals. Speaker for Abbvie, Incyte, Lilly, Pfizer, Regeneron, and Sanofi/Genzyme. Investigator for Lilly.
- Russell Hall, MD - Consultant for Akari Pharmaceuticals. Data Safety Monitoring Board for Oasis Pharmaceuticals. Prior Consultant and Investigator for Argenx and Principia.
- Leslie Lawley, MD - Consultant for Bristol Myers Squibb.
- Christopher Sayed, MD - Honoraria for Consulting/Speaking for Abbvie, Incyte, Novartis, and UCB. Investigator for Incyte, Novartis, and UCB.
- Jeff Wilson, MD, PhD – Receives support for Laboratory Assays from Thermo-Fisher/Phadia

All other presenters and/or planners have no relevant financial relationships with ineligible companies to disclose.

All relevant financial relationships with ineligible companies have been mitigated.

2024 POSTER PRESENTATIONS

Research posters are displayed onsite during Friday and Saturday meeting breaks in Ednam Hall. Half of the research posters will have live presentations on Friday, and half will be held on Saturday during meeting breaks. Clinical posters are displayed electronically during Friday and Saturday sessions in Ednam Hall.

Poster Award Categories:

Best Clinical Research Poster, Runner-Up Clinical Research Poster,

Best Basic Science Research Poster, Runner-Up Basic Research Science Poster,

Clinical People's Choice Poster *(clinical category only)*

Winners will be announced at 12pm on Sunday

Be sure to vote for the Clinical People's Choice Poster!

2024 POSTER PRESENTATIONS

Research Posters

#	Name	Title	Affiliation
1	Abukhadra, Sabine	Spesolimab for the Treatment of Generalized Pustular Psoriasis (GPP) Flares	Bowman Gray School of Medicine of Wake Forest University
2	Andrews, Laura	Characterizing Non-Melanoma Skin Cancer in the Hispanic Population	MUSC Dermatology Department
3	Bateganya, Lisa	Understanding Resident Perspectives: Key Factors for Matching and Excelling in Dermatology Residency	Medical College of Georgia
4	Batheja, Aashish	Evaluating Adverse Effects of Oral Tranexamic Acid for Treatment of Melasma with a Global, Federated Network	Virginia Commonwealth University School of Medicine
5	Burns, Meredith	Community Perceptions of Clinical Trials and the Relevance for Dermatology	UAB Heersink School of Medicine
6	Carmichael, Anna	Cicatricial Alopecia in the Pediatric Population: A Case Series and Review of the Literature	Wake Forest University, School of Medicine
7	Chime-Eze, Chinecherem	Variation in Hospital Use of Adjuvant Immunotherapy for Stage III Cutaneous Melanoma	University of North Carolina at Chapel Hill
8	Foushee, Raisa	Barriers to Accessing Dermatologic Care and Potential Solutions: Patient Perspectives in Richmond, Virginia	Virginia Commonwealth University, School of Medicine
9	Gadomski, Stephen	Compliance of Dermatology Screening Visits among Patients with Skin Cancer-Predisposing Mutations	Medical University of South Carolina, College of Medicine
10	Gantz, Hannah	Is It True that Psoriasis Skin Lesions Tend to Occur before Arthritic Involvement?	Wake Forest University School of Medicine
11	Greenzaid, Jonathan	Surgical Management of Acral Lentiginous Melanoma and Nail Unit Melanoma at an Academic Medical Center	Wake Forest University, School of Medicine
12	Guirguis, Christopher	The Protective Effects of Biologics on Skin Cancer in Patients with Psoriasis: A Database Study	Georgetown University School of Medicine
13	Guo, Robyn	Drug Survival of Biologics in Pediatric Patients with Hidradenitis Suppurativa Seen at Duke	Duke University School of Medicine
14	Gupta, Anahita	Analyzing Patients' Comprehension of Written Patient Education Materials at Various Reading Grade Levels: A Cross-Sectional Study	University of North Carolina at Chapel Hill/School of Medicine
15	Hagander, Maya	The Availability of In-Patient and Consultative Dermatology in Virginia Hospitals: A Needs Assessment	University of Virginia School of Medicine
16	Heilenman, Abigail	Alleviating Skin Cancer Misconceptions in Homeless Populations through Community-Based Health Education	Wake Forest University, School of Medicine
17	Money, Silas	Incidence of Bullous Pemphigoid after Scabies Diagnosis in Dialysis patients: A Retrospective Cohort Study	Medical College of Georgia

2024 POSTER PRESENTATIONS

18	Hrin, Matthew	Mohs Micrographic Surgery Vs Wide Local Excision for Primary Dermatofibrosarcoma Protuberans	Wake Forest University School of Medicine
19	Jiminez, Victoria	Surgical Management of Hidradenitis Suppurativa in Patients with Human Immunodeficiency Virus	Heersink School of Medicine, University of Alabama at Birmingham
20	Johnson, Chandler	Utility of Human Papillomavirus Vaccination for Keratinocyte Carcinomas: A Literature Review	Medical College of Georgia
21	Lyles, Elliott	Matched Case Control Study Assessing Onset of Skin Cancer in Patients with Gilbert Syndrome	Medical University of South Carolina - College of Graduate Studies
22	Lyons, Catherine	Utilization of Herbal Medicine in the Appalachian Region	University of Virginia School of Medicine Department of Dermatology
23	Marcelletti, Anthony	Impact of Anchoring on Acne Treatment Preference	University of Kentucky, College of Medicine
24	Martin, Taylor	Patient Preference for Safety Vs Efficacy in the Treatment of Psoriasis: A Literature Review	East Tennessee State University, James H. Quillen College of Medicine
25	McGrath, Lauren	Opioid Prescribing by Mohs Vs Non-Mohs Dermatologists for Medicare Part D Patients in 2022	Wake Forest University, School of Medicine
26	McRae, Charlotte	Enhancing Dermatology Practice through Art: Impact on Clinical Proficiency and Physician Well-Being	UAB Heersink School of Medicine
27	Moran, Shannon	A Novel Skin Classification System Utilizing AI-Driven Facial Analysis for Enhanced Skincare Recommendations	Center for Research at Wake Forest Department of Dermatology
28	Nichols, Deaquan	Augmentation of COVID-19's Neurological Impact in Patients with Subacute Cutaneous Lupus Erythematosus	Virginia Commonwealth University School of Medicine
29	Ormaza Vera, Ana	Increased Incidence of Major Adverse Cardiovascular Events among Psoriasis Patients Aged 40-75, without of type-2 Diabetes mellitus, Treated with Moderate-High Intensity Statins per ACC/AHA guidelines, Compared to Those Not on a Statin: A Retrospective Cohort Study of 2,587 Psoriasis Patients.	Eastern Virginia Medical School
30	Palmer, Victoria	The Current State of Patient Adherence to Lab Monitoring Guidelines for Jak Inhibitors	Wake Forest University, School of Medicine
31	Parkinson, Sierra	Patch Test Sensitization During the COVID-19 Pandemic and Comparison Between Two Propolis Vendors	University of North Carolina at Chapel Hill/School of Medicine
32	Patel, Heli	The Effectiveness of an Educational Intervention to Introduce Adolescents from Marginalized Backgrounds to dermatology: A Pilot.	Jefferson Medical College of Thomas Jefferson University
33	Razler, Dillon	Histopathologic Reporting of Non-Melanoma Skin Cancers and Implications for Treatment	Wake Forest University, School of Medicine

2024 POSTER PRESENTATIONS

34	Ruley, Ainsley	Utilization of p16, p21, Ki67, HMB-45, and Prame Immunohistochemistry in Melanocytic Lesions	Wake Forest University, School of Medicine
35	Sagut, Pelin	A Retrospective Review of Direct Immunofluorescence Patterns in Lupus Erythematosus	Medical University of South Carolina
36	Schwarz, Taylor	Skin Carotenoid Levels and Dermatologic Diseases	Georgia Skin and Cancer Clinic
37	Seely, Mason	Comparing Disease Severity, Management, and Clinical Outcomes across Racial/Ethnic groups, a Retrospective Analysis of Pemphigus Patients Seen at Duke University Hospital	University of Florida Health
38	Shah, Shailey	The Use and Impact of Patient Support Groups in Hidradenitis Suppurativa Patients	Wake Forest University School of Medicine
39	Shams, Rayad	Prevalence of Adulthood Secondhand Cigarette, Cannabis Smoke, and Electronic Cigarette Vapor Exposure in Hidradenitis Suppurativa Patients	University of North Carolina School of Medicine
40	Shan, Divya	The Impact of Financial Toxicity on Quality of Life in Hidradenitis Suppurativa Patients: A Single-Center Cross-Sectional Survey Study	Virginia Commonwealth University, School of Medicine
41	Sluder, Isaac	Examining the Attitudes of Gender Affirming Care Providers Toward Incorporating Dermatologists into Gender Affirming Care Teams: A quantitative and qualitative study	UNC School of Medicine
42	Smith, Aaron	The Effect of Anchoring on Patients' Likeliness to Take an Injection Treatment for Hidradenitis Suppurativa	University of Virginia
43	Suen-Wallach, Alisa	Assessing Changes in Dermatology Resident Proficiency and Confidence following an Online Educational Module on Evaluation and Interpretation of Direct Immunofluorescence (DIF).	UNC Chapel Hill School of Medicine
44	Sunkara, Meghana Devi	The Role of Artificial Intelligence in Explaining Graft Vs Host Disease to Children: A Comparison of Chatgpt and Gemini for Readability and Image Accuracy	Eastern Virginia Medical School
45	Swain, Elizabeth	A Pilot Study of Revian Red All Led Cap as a Novel Treatment for Central Centrifugal Cicatricial Alopecia	Wake Forest University, School of Medicine
46	Swift, Alexis	Allergic Disease and the Risk of Rosacea and psoriasis: A Population-Based Cohort Study Using a Large U.S. Database	Virginia Commonwealth University School of Medicine
47	Thigpen, Bradley	An Evaluation of Infusion Reactions Using Infliximab Vs. Infliximab Biosimilars	University of Alabama Heersink School of Medicine
48	Vescovacci, Nashali	Exploring the Cutaneous Associations of Crohn's Disease: A Retrospective Study of 108 Patients	Wake Forest University, School of Medicine
49	Yi, Robin	The Natural Progression of Molluscum Contagiosum	Wake Forest University, School of Medicine

2024 POSTER PRESENTATIONS

Clinical Posters

#	Name	Title	Affiliation
1	Agner, Morgan	Clinical features of concurrent vulvar lichen sclerosus and vitiligo: a case series	Wake Forest University, School of Medicine
2	Ali, Rowanne	Significant Improvement of Treatment-Resistant Dermatomyositis with Anifrolumab Treatment	Emory University Department of Dermatology
3	Attari, Sara	Eruptive Keratoacanthomas following 5-Fluorouracil Topical Therapy	Medical College of Georgia
4	Blanchard, Kaitlyn	Demodex Folliculitis in a Pediatric Patient with a NOD2 Mutation	MCG
5	Choudhary, Fatima	Multiple Asymptomatic Papules Arising within a Birthmark	University of Virginia School of Medicine
6	Clawson, Rebecca	Lichen Planus Pemphigoides in an Adolescent Male with Trisomy 21	VCU Dermatology
7	Collette, Sydney	Acrodermatitis Enteropathica-Like Dermatitis in Pediatric Patient with Biotin Deficiency	Emory University School of Medicine - - Atlanta, GA
8	Collins, Maya	Toxic erythema of chemotherapy induced by enfortumab vedotin	Emory University School of Medicine
9	Cushman, Courtny	Superficial granulomatous pyoderma gangrenosum in a pediatric patient: A rare diagnosis	Eastern Virginia Medical School
10	Daly, Kathleen	Utility of Ultraviolet-Induced Fluorescence Dermoscopy in Diagnosing Scalp Disorders	Medical College of Georgia, School of Medicine
11	Dar, Nakul	Use of Sirolimus for Refractory Cutaneous Dermatomyositis: A Retrospective Case Series	University of Virginia School of Medicine
12	Drohan, Alex	Unconventional Hues: An Atypical Presentation of Erythropoietic Protoporphyria	MUSC
13	Duchesne, Gabriela	Non-Scrapable Yeast: A Case of Chronic Hyperplastic Candida	Medical College of Georgia
14	Dyson, Taylor	Don't Sweat It: A Case of Managing Gustatory Hyperhidrosis with Botulinum Toxin	Eastern Virignia Medical School
15	Echuri, Harika	Leukemia Cutis in a Patient with Chronic Myelomonocytic Leukemia (CMML)	Emory Universtiy Department of Dermatology
16	Faraz, Khushnood	Pacemaker Lead Erosion Presenting as a Scaly Papule with Electric Shock Sensations	Duke University
17	Gangal, Ameya	Metastatic angiosarcoma mimicking lymphedema	Emory University School of Medicine
18	Gaurav, Ahana	A Case of Lichen Planus-Like Features in a Young Male with Dysphagia	Medical College of Georgia, School of Medicine
19	Himed, Sonia	Consideration of Porphyria Cutanea Tarda as a Rare Differential in the Setting of Liver Dysfunction	Emory Dermatology Residency
20	Huang, Chenan	Formalin Safety in the Outpatient Setting: An Update of Safety Precautions	Wake Forest University, School of Medicine
21	Jowdy, Peter	Sle Presenting as an Acute Bullous Eruption	UVA Dermatology Resident

2024 POSTER PRESENTATIONS

22	Jung, Joo	Tinted Sunscreen for Camouflage and Sun Protection in Vitiligo Patients	Medical College of Georgia
23	Karatas, Turkan Banu	Atypical Serologic and Immunofluorescence Findings in Granulomatosis with Polyangiitis (GPA)	Emory University School of Medicine
24	Leath, Mary	Vexed by VEXAS: A Case Report	Medical University of South Carolina
25	Lopes Almeida Gomes, Lais	Pemphigus Vulgaris Complicated by Acyclovir-Resistant Herpes Simplex infection: A Case Report	Hospital of the University of Pennsylvania
26	McGrath, Lauren	Efficacy of Deucravacitinib and Guselkumab in a Psoriasis Patient with Multiple Biologic Failures: A Case Report	Wake Forest University, School of Medicine
27	McPherson, Gillian	Intralesional Adalimumab for the Treatment of Refractory Cutaneous Sarcoidosis	MUSC
28	McPherson, Gillian	A Novel Case of Risperidone-Induced Pseudoporphyria Affecting Non-Sun-Exposed Skin	Medical College of Georgia
29	Momin, Rushan	The Recombinant Zoster Vaccine in Immunocompromised Patients	Medical College of Georgia, School of Medicine
30	Moore, Kaila	Cutaneous Herpes Outbreak following Talimogene Laherparepvec	UNC School of Medicine
31	Moran, Shannon	Erythematotelangiectatic Rosacea: A Patient Journey Case Report	Center for Research at Wake Forest Department of Dermatology
32	Myers, Emma	Case of Multiple Pilar Cysts and Atypical Proliferating Pilar Tumors: Syndromic Presentation to be Distinguished from Cutaneous Squamous Cell Carcinoma	UNC School of Medicine
33	Myers, Hannah	Keep Your Hands Peeled: A Novel Plec Variant in Epidermolysis Bullosa Simplex	Medical College of Georgia
34	Neimy, Hannah	Acne Development Associated with GLP-1 Agonist Therapy: A Case Report	Medical University of South Carolina
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CASE 1: MHC Class I deficiency/TAP2 mutation

Authors: Rebecca Hicks BA, Courtney Remington MD, Barrett Zlotoff MD

HISTORY

An 11-year-old female patient presented with chronic bilateral thigh ulcers. At age 2, she experienced Gianotti Crosti syndrome following MMR vaccination, which resolved spontaneously. Following her second MMR vaccination at age 5, she developed enlarging purple bumps on her right thigh associated with pain and pruritus. Despite an initial diagnosis of "picker's nodules," subsequent evaluation revealed necrotizing granulomatous inflammation in the left thigh biopsy. Further investigation indicated persistent rubella virus in the left leg ulcer and an immunodeficiency panel confirmed a TAP2 mutation. Family history includes consanguinity, with a similar vaccine-related reactions in a cousin.

PHYSICAL EXAMINATION

On the bilateral thighs were erythematous and violaceous plaques with indurated borders, satellite areas of ulceration as well as overlying granulation tissue along the periphery.

PATHOLOGY

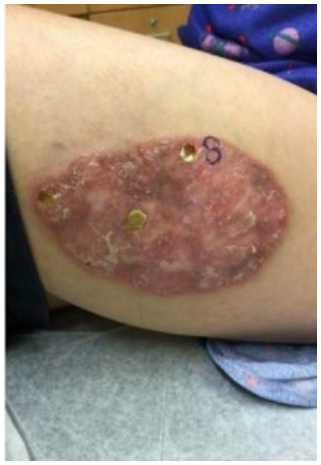
Punch biopsy from the left thigh demonstrated necrotizing granulomatous inflammation. A research test using double immunofluorescent staining with rubella (RuV) capsid mouse monoclonal antibody and CD206 M2 macrophage-specific rabbit polyclonal antibody was performed and positive for rubella P4+ (on a scale of 1+ to 4+).

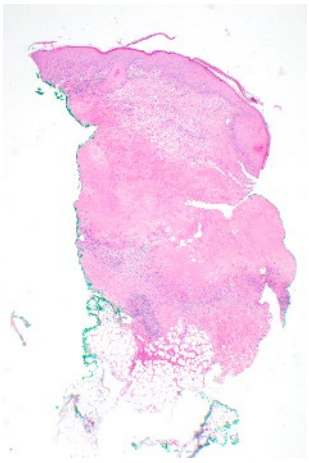
TREATMENT

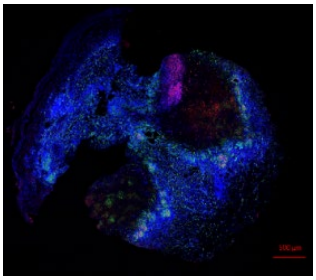
Treatments have been relatively unsuccessful, including oral doxycycline, itraconazole, and hydroxychloroquine combined with nitazoxanide (Alinia). She recently started topical imiquimod. She is not yet pursuing stem cell transplant. Ongoing management is focused on symptomatic relief and infection control.

DISCUSSION

Granulomatous skin lesions due to persistent rubella virus have been reported in both immunocompromised and immunocompetent patients.[1,2] Onset of these granulomas is variable and symptoms range from pruritus, pain, to asymptomatic. Though reported to occur at sites of prior vaccination, as in our patient, they may also affect non-vaccine sites. The initial presentation is concerning for infection, but further investigation will rule out an infectious source. A thorough patient and family history may help reveal clues pointing to an underlying IEI. Our patient's older cousin was reported to have similar skin findings and upon investigation by our clinic found to share the same TAP2 deficiency.







Double immunofluorescent staining with RuV capsid mouse monoclonal antibody and CD206 M2 macrophage-specific rabbit polyclonal antibody demonstrates positivity for rubella P4+.

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CASE 2: Pityriasis rubra pilaris

Authors: Hope Winfield BS, Courtney Remington MD, Barrett Zlotoff MD

HISTORY

A 45-year-old male with a history of hypertension and tobacco use presented with a diffuse rash for four weeks. This initially started on the scalp, with cephalocaudal progression to nearly 100% of body surface area (BSA), while sparing the mucous membranes and genitals. He denied recent medication changes.

PHYSICAL EXAMINATION

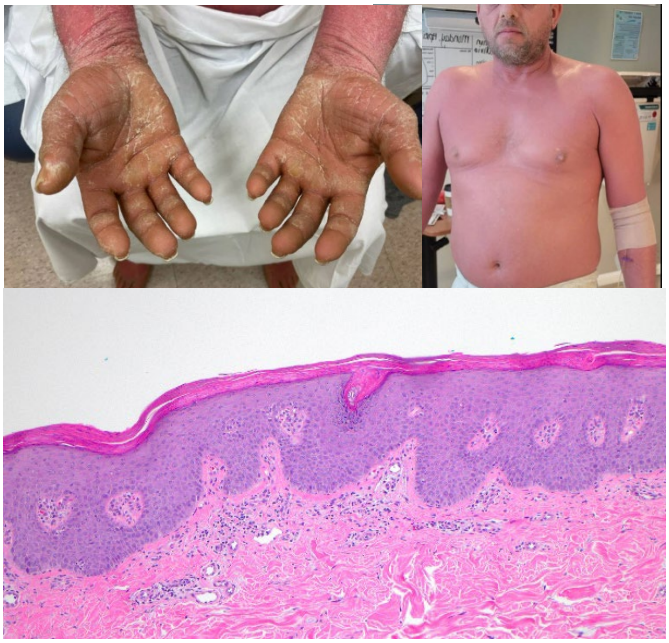
There was exfoliative erythroderma involving 100% of BSA with thick scale overlying the scalp, face, and extremities. He had marked edema of the hands with large sheets of scale peeling from the surface. Crusting was present within the bilateral conjunctiva. Islands of sparing were noted along the lower shins.

PATHOLOGY

Biopsy demonstrated thickening of the stratum corneum with parakeratotic foci between orthokeratosis oriented vertically and horizontally. Follicular plugging was also noted.

TREATMENT

Patient started cyclosporine 250 mg twice daily with topical triamcinolone 0.1% ointment, then transitioned to Ixekizumab, acitretin 10 mg daily, and methotrexate 10 mg weekly. After negligible improvement, acitretin and methotrexate were increased. Patient noted a slight increase in clearing skin; methotrexate was increased to 25 mg daily while acitretin and ixekizumab were continued as before. When he flared 5 months later, acitretin was increased to 50 mg daily. Given the lack of treatment response, methotrexate was discontinued and cyclosporine was restarted, while acitretin was continued at 50 mg daily.



DISCUSSION

Pityriasis rubra pilaris (PRP) is a rare inflammatory papulosquamous dermatosis of uncertain etiology marked by palmoplantar keratoderma and hyperkeratotic follicular papules merging into well-demarcated red-orange plaques with non-adherent scale.[1] Six clinical subtypes have been proposed; five defined on the basis of age of onset, disease extent, and sclerodermoid versus ichthyosiform characteristics, while the sixth is associated with HIV infection.[1] Recent open-label studies targeting the IL-17 pathway with ixekizumab and secukinumab highlight those as therapeutic options, with brodalumab, bimekizumab, IL-23 inhibitors and TNF-alpha inhibitors as other options. This case of largely treatment-refractory erythrodermic PRP underscores the challenges in managing this rare condition. In light of its profound impact on patients' quality of life, providers should also query about affected individuals' mental health.

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CASE 3: Addison's disease

Authors: Courtney Remington MD, Bridget Bryer MD, Lynne Wold NP

HISTORY

A 41-year-old woman presented to the dermatology clinic for evaluation of an approximately four-year history of gradually worsening discoloration of the oral mucosa. Her past medical history included a diagnosis of Sjogren's syndrome in 2022. With prompting, patient reported fatigue, heartburn, nausea, vomiting, and a 20-pound unintentional weight loss.

PHYSICAL EXAMINATION

Clinical examination revealed smooth, non-tender hyperpigmented patches on the tongue, bilateral buccal mucosa, hard palate, gingiva, and mucosal lips.

LABORATORY DATA

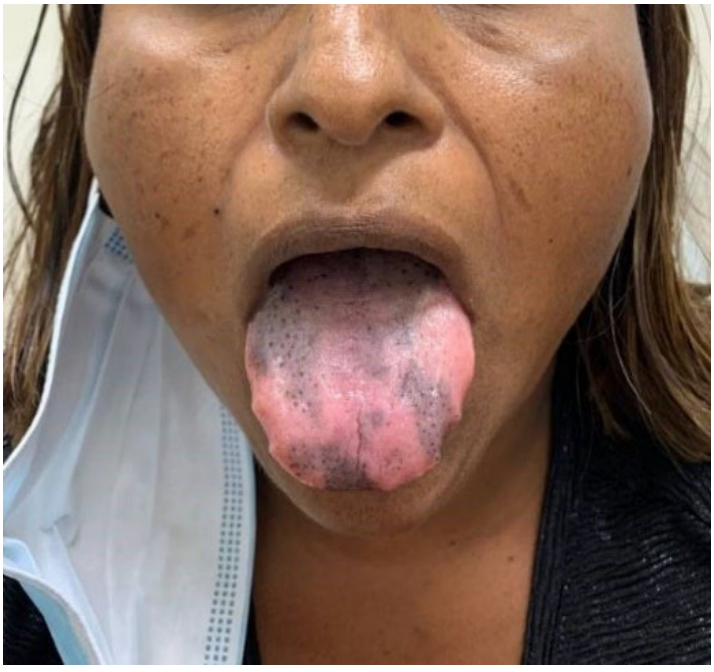
An endocrinology workup revealed elevated adrenocorticotropic hormone (ACTH) and plasma renin activity (PRA) levels which were consistent with primary adrenal insufficiency. A diagnosis of Addison's disease was confirmed by 21-hydroxylase autoantibody assay.

TREATMENT

The patient was referred to Endocrinology and started on 0.1 mg fludrocortisone and 20 mg hydrocortisone daily. Within a few days, her gastrointestinal symptoms and energy levels improved significantly.

DISCUSSION

Addison's disease is a rare condition characterized by insufficient glucocorticoid production by the adrenal cortex. Common presentations include hypotension, hyponatremia, hyperkalemia, and skin hyperpigmentation, though other nonspecific symptoms may make for a challenging diagnosis. Hyperpigmentation in Addison's disease is caused by induction of the melanocortin-1 receptor in the skin and mucosa by increased circulating corticotropin levels and usually involves sun-exposed areas, palmar creases, and nail beds.[1]



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CASE 4: Acrodermatitis Continua of Hallopeau treated with Ixekizumab

Authors: Diego Gomez BS, Courtney Remington MD, R. Hal Flowers MD

HISTORY

A 50-year-old female with no reported past medical history presented with a 3-week history of a painful pustular rash affecting 9/10 distal fingers, with a similar pustular rash on the plantar feet and medial ankles.

PHYSICAL EXAMINATION

Hyperkeratotic, scaling plaques with pustules were noted on the bilateral distal fingers with associated dystrophic and full loss of numerous fingernails. Scaly plaques are present on the bilateral plantar feet and medial ankles.

PATHOLOGY/RADIOLOGY

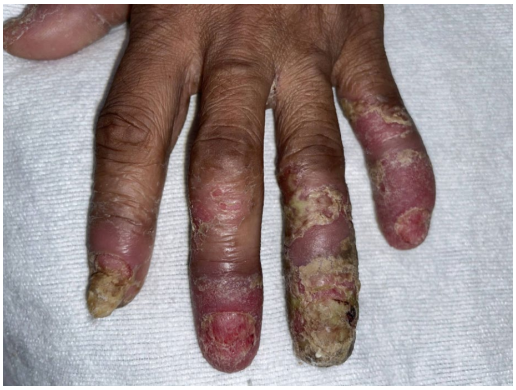
Biopsies of the left calf and left thumb showed a psoriasiform dermatitis. The left thumb biopsy demonstrated a large central subcorneal pustule . Neutrophils were present in the parakeratotic stratum corneum of both biopsies. Hand X-rays demonstrated no evidence of inflammatory arthritis or other notable changes.

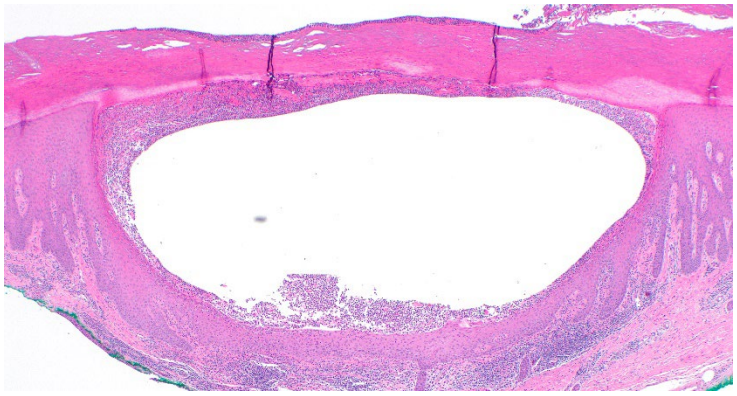
TREATMENT

Patient was diagnosed with acrodermatitis continua of Hallopeau (ACH). She was started on cyclosporine 100 mg twice daily and cefadroxil 500 mg twice daily, with significant improvement noted within 3 weeks. The patient was transitioned to ixekizumab while remaining on cyclosporine 100 mg BID. This regimen was later supplemented with acitretin 10 mg daily, and the patient noted significant improvement 8 months after initial presentation.

DISCUSSION

Clinical and histologic findings favored a psoriasiform process over infection. Importantly, significant involvement of the nails and distal fingers favored a diagnosis of ACH.[1] Due to the rare nature of ACH there are no standard treatment regimens, and most therapies are described through case reports. Ixekizumab was chosen as our primary treatment due to high efficacy of anti-IL-17 biologics in treating psoriatic skin disease and multiple reports of successful treatment of ACH with this class of medication.[2] While the patient remained on cyclosporine throughout the course of her treatment, she experienced the most improvement with ixekizumab maintenance dosing.





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CASE 5: Tufted Angioma

Authors: Courtney Remington MD, Barrett Zlotoff MD

HISTORY

An 8-year-old male presented to the hospital for worsening pain, swelling, and redness overlying a "birthmark" with concurrent fever. The "birthmark" on his left chest developed at 9 months old and remained unchanged until a similar spot appeared on the underside of his chin when he was 2 years old. He recently developed a similar spot on the right chest.

PHYSICAL EXAMINATION

On the submental chin, left anterior chest, and right anterior chest there are multiple firm, violaceous, arcuate plaques with surrounding erythematous patches and streaking noted.

PATHOLOGY

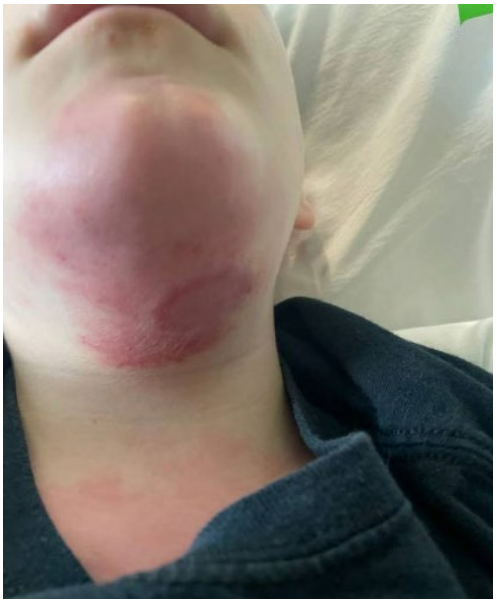
Punch biopsy from the central chest demonstrated clusters of glomeruloid capillary proliferations separated by normal dermis.

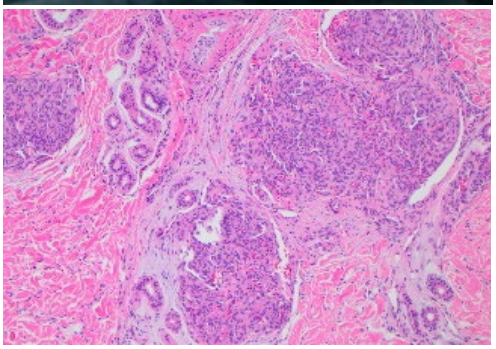
TREATMENT

Patient was admitted to the hospital due to concern of erysipelas on his neck and chest. An ultrasound of the chest wall was negative for a high flow lesion. A 4 mm punch biopsy was performed on the chest and was interpreted as a tufted angioma. He was seen in the dermatology outpatient clinic and elected to treat topically with sirolimus as well as PDL laser.

DISCUSSION

Tufted angiomas are rare tumors usually present at birth or within the first five years of life (50%).[1] They can enlarge rapidly when introduced to a trigger, such as an infection, which can be a signal of Kasabach-Merritt Phenomenon (KMP), a rare and potentially life-threatening consumptive coagulopathy with a microangiopathic hemolytic anemia. KMP occurs when excessive platelet trapping results in severe thrombocytopenia, anemia, elevated fibrin split products, and low fibrinogen.[2,3] Sirolimus +/- prednisone is generally used as first line treatment for true KMP. Once the threat of KMP has subsided, definitive treatment of the tufted angioma can be obtained via surgical resection, though radiation therapy, embolization, and laser therapy have also been utilized.





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CASE 6: Eosinophilic Annular Erythema Treated with Dupilumab

Authors: Divya M. Shan BA, Courtney Remington MD, R. Hal Flowers MD

HISTORY

A 66-year-old female with a past medical history of type 2 diabetes mellitus, hypothyroidism, hypertension, and Crohn's disease presented with a 50-year history of waxing and waning pruritic rash primarily affecting her extremities and face, especially following insect bites. She had previously been diagnosed with idiopathic eosinophilic annular centrifugum and showed minimal response to multiple treatments, including clobetasol ointment, antihistamines, prednisone, and azathioprine.

PHYSICAL EXAMINATION

Numerous urticarial annular and polycyclic pink to red plaques on the bilateral upper and lower extremities.

PATHOLOGY

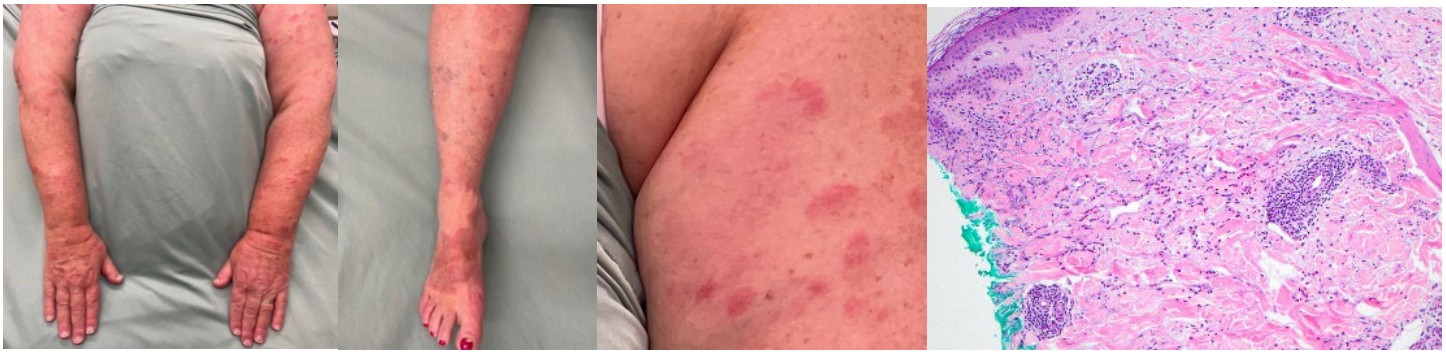
Biopsy of a recent lesion on the chest revealed significant predominantly perivascular infiltrate with numerous eosinophils and occasional lymphocytes.

TREATMENT

Based on the morphology, history and pathology, patient was diagnosed with eosinophilic annular erythema (EAE). She was started on dupilumab, starting with a 600 mg subcutaneous injection, followed by 300 mg every other week. After five days of treatment, she reported dramatic symptomatic improvement.

DISCUSSION

EAE is a rare eosinophilic dermatosis with ongoing debate regarding whether it is a distinct clinical entity or subtype of Well's syndrome.[1] EAE features a superficial and deep perivascular and interstitial inflammatory pattern with abundant eosinophils and occasional flame figures. It is characterized by a chronic, relapsing-remitting course and is often refractory to treatment. Standard therapeutic options are limited and include systemic steroids, hydroxychloroquine, and dapsone. Recently, dupilumab, which blocks the IL-4 and IL-13 signaling pathways involved in the activation and recruitment of eosinophils, has emerged as an efficacious therapy.[2]



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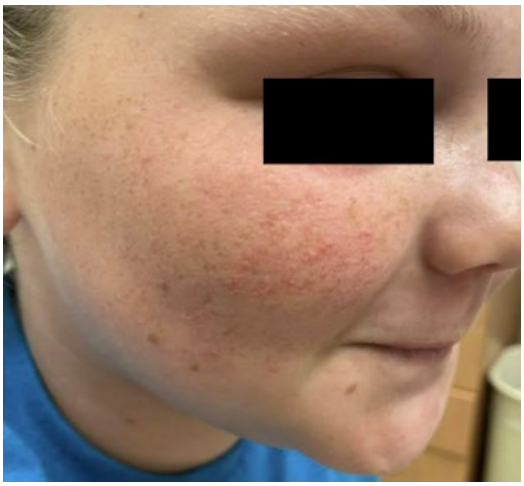
CASE 7: Elastosis perforans serpiginosa

Authors: Laura I. Ortiz-López BS, Gabrielle Schwartzman MD, Barrett Zlotoff MD

HISTORY

A 10-year-old female presented with an eight-month history of a pruritic, painful, and scaly rash on the antecubital fossa with a similar scaly rash on her bilateral cheeks for five years. These lesions were more noticeable during the winter. The patient had no known medical conditions. Patient also had a history of a Salter-Harris II fracture of the distal radius.

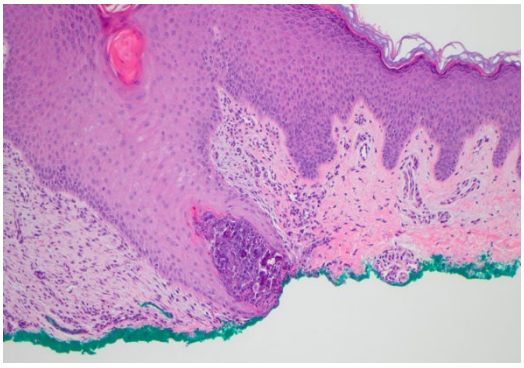


PHYSICAL EXAMINATION

One cm erythematous papules in an annular configuration with surface scaling on the right antecubital fossa. Small, scaly, and erythematous papules involving the bilateral cheeks with background pitted scarring.

PATHOLOGY

A shave biopsy of the right antecubital fossa revealed transepidermal elimination of a keratinous plug through the dilated infundibulum and acanthotic epidermis. The extruded material contained brightly eosinophilic fibers, keratinous debris, and inflammatory cells. Van Gieson's stain highlighted elastic fibers within the extruded material and increased elastic fibers in the papillary dermis.



TREATMENT

The patient was diagnosed with idiopathic elastosis perforans serpiginosa (EPS) and started on a tretinoin 0.025% cream nightly.

DISCUSSION

Elastosis perforans serpiginosa (EPS) is a rare perforating It is often associated with underlying disorders such as osteogenesis imperfecta, Marfan syndrome, Ehlers-Danlos syndrome, and Down syndrome. There are also reports of idiopathic and drug-induced EPS, particularly related to D-penicillamine. To our knowledge, EPS involving the face has only been documented twice in the literature. Spontaneous resolution of EPS is not uncommon. However, the intense pruritus and pain can be distressing to patients. Various treatments, such as photodynamic therapy, pulsed dye and carbon dioxide lasers, topical steroids, and retinoids, have been reported. However, none of these treatments can fully prevent recurrence.

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CASE 8: Giant cell arteritis with alopecia

Authors: Laura Ortiz-López BS, Gabrielle Schwartzman MD, Mary Margaret Noland MD

HISTORY

An 81-year-old female with a past medical history of an aortic aneurysm repair, coronary artery disease, anemia, hypertension, peripheral vascular disease, and type two diabetes mellitus presented with scabs on the scalp, a bruise-like rash on her neck, and hair-loss. These symptoms started two days after her aneurysm repair and have persisted for the past two months. Since her procedure, she also experienced severe headache, neck pain, jaw pain and fatigue. Her head CT was negative for acute pathology.

PHYSICAL EXAMINATION

Prominent vessels on the bilateral temples with nodularity to palpation, with violaceous retiform purpura with ulceration on the posterior neck. Stellate hemorrhagic ulcers were present on the right paramedian scalp with adherent scab and alopecia at areas of involvement.

PATHOLOGY

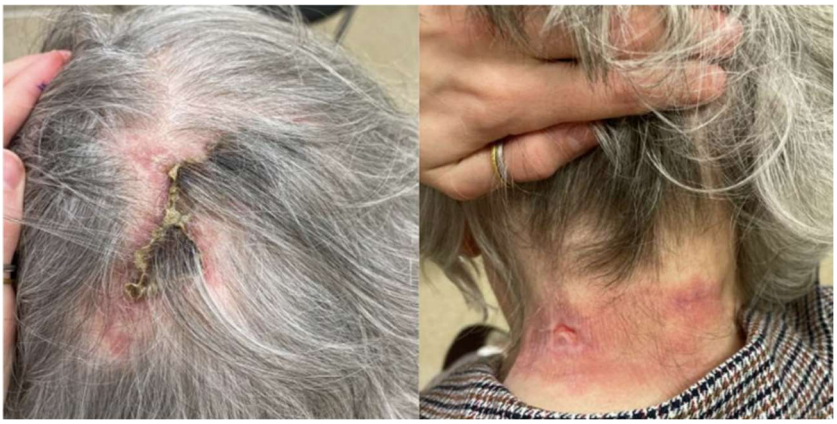
Temporal artery biopsy showed marked intimal fibrosis and significant luminal stenosis, consistent with giant cell arteritis.

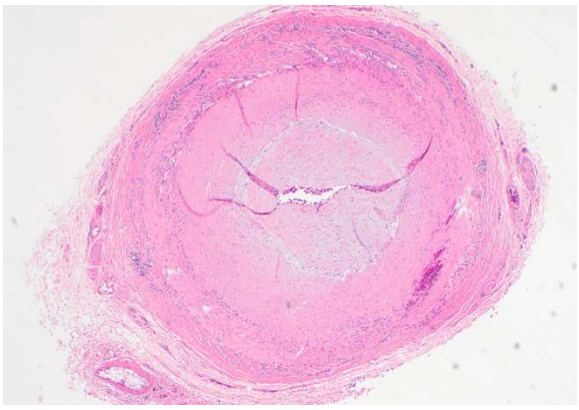
TREATMENT

Patient was started on oral prednisone 40mg daily and her temporal artery biopsy revealed giant cell arteritis. Her ESR and CRP were normal. The patient experienced adverse effects to prednisone, and it was later tapered to 20 mg daily. She was started on tocilizumab as a steroid-sparing agent. She remained in disease remission on tocilizumab monotherapy after 1 year, and subsequently stopped infusions without recurrence of symptoms. Patient represented for follow-up with exam findings showing stellate scars with erythematous reticulation of the paramedian scalp and violaceous scars of the posterior neck. Early hair regrowth was noted at the areas of involvement.

DISCUSSION

Giant cell arteritis (GCA) is a type of large and medium-sized vessel granulomatous vasculitis commonly seen in older adults. ESR and CRP tests are usually raised or normal, as seen in this case. Cutaneous manifestations of GCA are rare and appear later in the disease course but reflect a poor prognosis.[1] These include ischemic symptoms such as hair loss, ulceration and necrosis of the scalp, and indurated nodules. If clinical suspicion of GCA arises, glucocorticoid administration is crucial.





REFERENCE

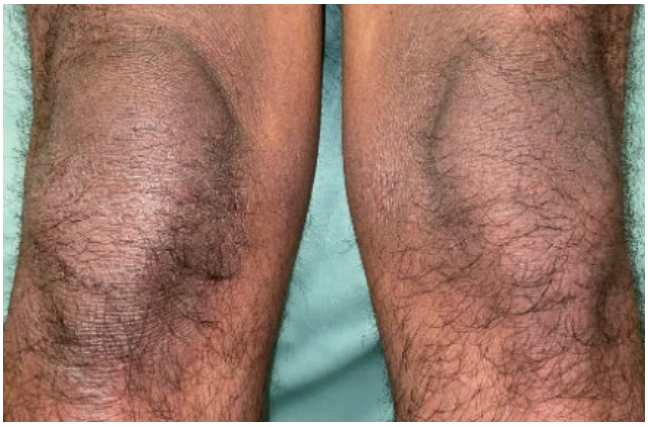
1. Prieto-Peña et al. “A Review of the Dermatological Complications of Giant Cell Arteritis.” *Clinical, cosmetic and investigational dermatology*;2021(14):303-312.

CASE 9: Wong-type Dermatomyositis

Authors: Mary Kathryn Howard BS, Gabrielle Schwartzman MD, R. Hal Flowers MD

HISTORY

30-year-old male with PM/Scl+ overlap connective tissue disease presented for skin thickening, fasciitis of the thigh on MRI, and rash of the extremities.

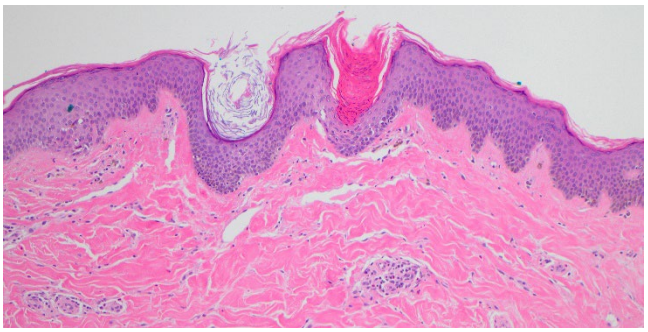


PHYSICAL EXAMINATION

Taught skin, firm, hyperkeratotic plaques and overlying follicular roughness on extensor upper extremities and knees with background erythema.

LABORATORY DATA/PATHOLOGY

Labs notable for elevated erythrocyte sedimentation rate, C reactive protein, creatine kinase, aldolase, and transaminases. He also had a high titer ANA of 1:640 in a nucleolar pattern with myositis panel positive for a PM/Scl and Ro 52 antibodies. Biopsies from left arm and left knee revealed vacuolar interface dermatitis with parakeratotic follicular plugs and no evidence of fasciitis. Immunofluorescent stains were positive for linear IgM, granular C5b9, and granular C3 at the basement membrane zone. Biopsies were consistent with Wong-type dermatomyositis (DM).



TREATMENT

He was previously treated with hydroxychloroquine (HCQ) without improvement, so he transitioned to mycophenolate mofetil (MMF) 1g BID. His myositis and arthralgias improved, but he has ongoing calcinosis cutis being treated with intralesional sodium thiosulfate injections. He was referred for baseline pulmonary and cardiac testing and CT abdomen/pelvis for malignancy workup.

DISCUSSION

Wong-type DM is the overlap of DM, an inflammatory myopathy with cutaneous features, and pityriasis rubra pilaris (PRP), a papulosquamous disorder with scaly plaques and follicular plugging and papules.[1] Fewer than 30 cases have been reported.[1] Keratotic follicular papules with compact orthokeratosis within follicular and non-follicular epidermal invaginations on histology, seen here, is a distinguishing feature. Two reports have similarly shown pseudocornoid lamellar changes, though both cases had clinical evidence of porokeratosis, not seen here.[1,2] Though rare, Wong-type DM should be suspected in patients with features DM like arthralgias, muscle weakness plus characteristic cutaneous findings and concurrent histopathologic findings of PRP.

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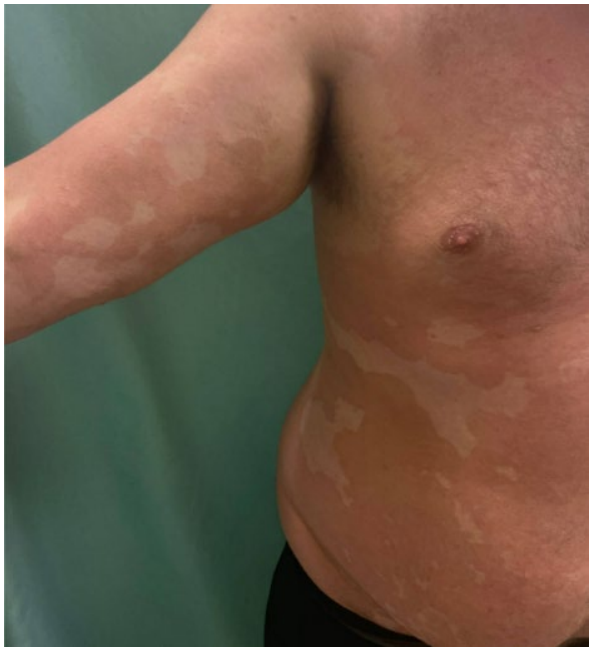
2. Lupton et al. An unusual presentation of dermatomyositis: the type Wong variant revisited. *JAAD.* 2000 Nov;43(5 Pt 2):908-12.

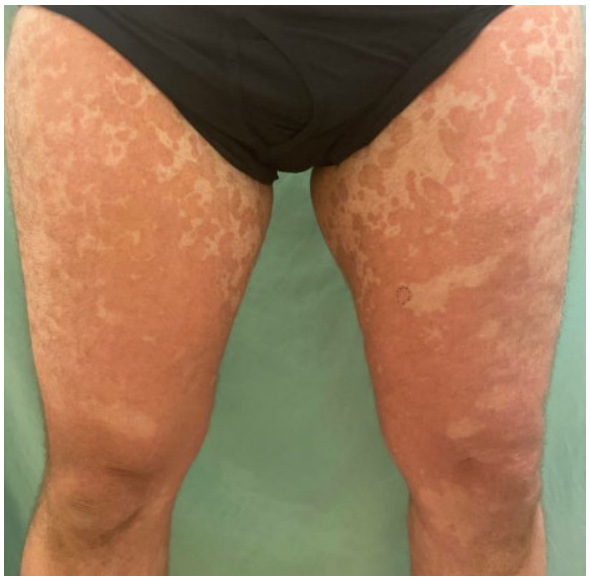
CASE 10: Tinea Versicolor Mimicking Pityriasis Ruba Pilaris

Authors: Mary Kathryn Howard BS, Gabrielle Schwartzman MD, Mary M Noland MD

HISTORY

41-year-old white male electrician with no significant past medical history presented with a five-year history of relapsing, remitting pruritic rash. The rash was intermittently scaly. Additionally, he endorsed associated hypohidrosis. At the time of presentation, he noted a severe flare, with the rash covering a significant body surface area. Primary care was concerned he had erythema multiforme and treated with a six-day course of prednisone and Benadryl without improvement. He denied mucosal or palmoplantar involvement, fever, night sweats, or unintentional weight loss.





PHYSICAL EXAMINATION

Diffuse erythematous, hyperpigmented patches and plaques with fine scale on the trunk, buttock, and bilateral upper and lower extremities. Areas of sparing, including bilateral axillae were present. No rash on the face, scalp, palms, or soles.

MICROSCOPY

KOH scrapings from multiple sites were diffusely positive for short hyphae and spores, consistent with tinea versicolor.

TREATMENT

He was treated with oral fluconazole 300mg once weekly and topical selenium sulfide 2.5% wash daily for 2 weeks. At one month, he reported 95% improvement of the rash and pruritis, with all areas except for his inner thighs clearing completely. He was switched to topical selenium sulfide every 3 days.

DISCUSSION

Given the extensive involvement with areas of sparing, there was initially concern for pityriasis rubra pilaris (PRP). However, the patient had no evidence of palmoplantar keratoderma classically associated with PRP. The diffusely positive KOH scrapings and rapid response to oral fluconazole were consistent with severe, extensive tinea versicolor. Providers should be aware that severe tinea versicolor can mimic PRP.[1] Two doses of fluconazole 300mg over two weeks in conjunction with selenium sulfide 2.5% lotion used as a wash was helpful in this case.

REFERENCE

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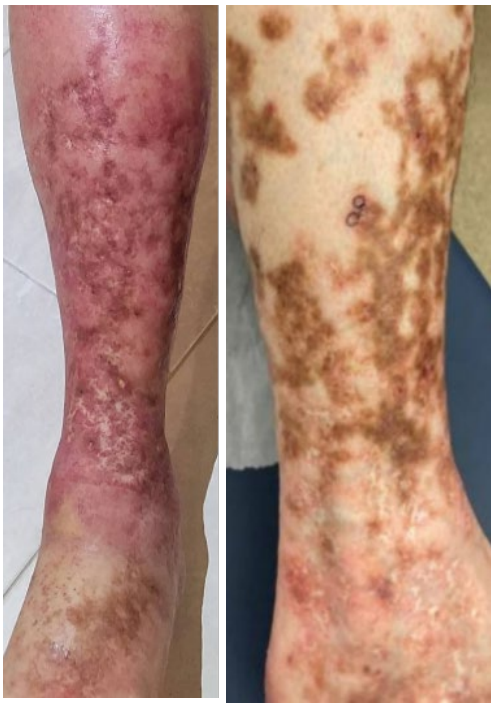
CASE 11: Livedoid vasculopathy-like changes treated with CHAP

Authors: Mary Kathryn Howard BA, Gabrielle Schwartzman MD, R Hal Flowers MD

HISTORY

64-year-old male with hypertension, hyperlipidemia, and osteoarthritis presented with 20-years relapsing, remitting ulcerations of his lower extremities. Remote outside biopsy of the lower extremity revealed ulcerated stasis dermatitis (SD). Venous duplex ultrasound at that time showed no abnormalities.

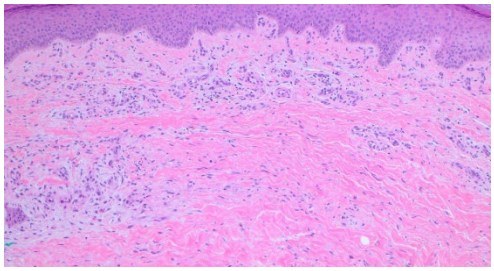


PHYSICAL EXAMINATION

Erythema with intermixed hyperpigmented induration and 1+ pitting edema of the left lower leg; reticulated, hyperpigmented scarred plaques on the right lower leg. White, stellate atrophic scars on bilateral ankles featured no active ulceration.

PATHOLOGY/LABORATOR DATA

Biopsy showed microvascular proliferation, dermal sclerosis, red blood cell extravasation, hemosiderin deposition, and superficial perivascular lymphocytic infiltrate, consistent with marked SD. Diffuse sclerosis of subcutaneous tissue and membranocystic changes, consistent with lipodermatosclerosis. Fibrinoid change in the superficial dermal vessels with fibrinogen and C3 deposition on direct immunofluorescence. He underwent an extensive work-up without evidence of autoimmune connective tissue disease (CTD) or hypercoagulable state.



TREATMENT

The patient was diagnosed with atrophie blanche/LV-like changes from severe SD. He was treated with pentoxifylline, aspirin (ASA) 325mg, and topical clobetasol without improvement. He was later treated with a combination of hydroxychloroquine 400mg, nifedipine 60mg, pentoxifylline 400 mg TID, ASA 325mg, and topical timolol. This regimen led to rapid and dramatic improvement. He successfully tapered off pentoxifylline, and ASA was decreased to 81mg. He developed blue-grey discoloration of his lower extremities, so hydroxychloroquine was also discontinued.

DISCUSSION

Livedoid vasculopathy (LV), a thrombo-occlusive vasculopathy associated with coagulopathy and CTD, can present with ulceration, atrophie blanche, and fibrinoid changes on histology.[1] Severe, ulcerative SD can appear clinically and histologically similar. Dermatologists should be aware of this overlap to avoid treatment delays, though LV-associated conditions may still need to be excluded. The patient's rapid and sustained improvement support recent data that the combination of a calcium channel blockade, hydroxychloroquine, aspirin, and pentoxifylline (CHAP) is a promising regimen for the treatment of LV-like changes from SD/LDS.[1]

REFERENCES

1. Coromilas & Micheletti. A novel combination ("CHAP") regimen for management of livedoid vasculopathy in 12 patients. *JAAD; 2023: 88*(3), 672–674.

CASE 12: Generalized Dowling-Degos Disease due to a Novel Mutation in *POFUT1*

Authors: Nidhi Kuchimanchi, Lauren Yi MD, Barrett Zlotoff MD

HISTORY

A 37-year-old male born in Honduras and his 8-year-old daughter presented with hyperpigmentation. The man, his mother, son, and daughter reported skin fragility and peeling from infancy and early childhood. His mother and son reportedly had no pigmentary alterations.

PHYSICAL EXAMINATION

The man had symmetrically distributed, reticular hyperpigmented macules and patches with areas of superficial desquamation over the face, neck, trunk, abdomen, and extremities. His daughter had lichenified plaques and scattered hyperpigmented macules and pits with patches of superficial desquamation over the flexures and dorsal hands.

PATHOLOGY/LABORATORY DATA

A punch biopsy taken from the left flank of the man revealed extensive hyperkeratosis and regular acanthosis with superficial epidermolysis. Whole exome sequencing on both patients identified a c.246+4 A>T variant in the *POFUT1* gene.

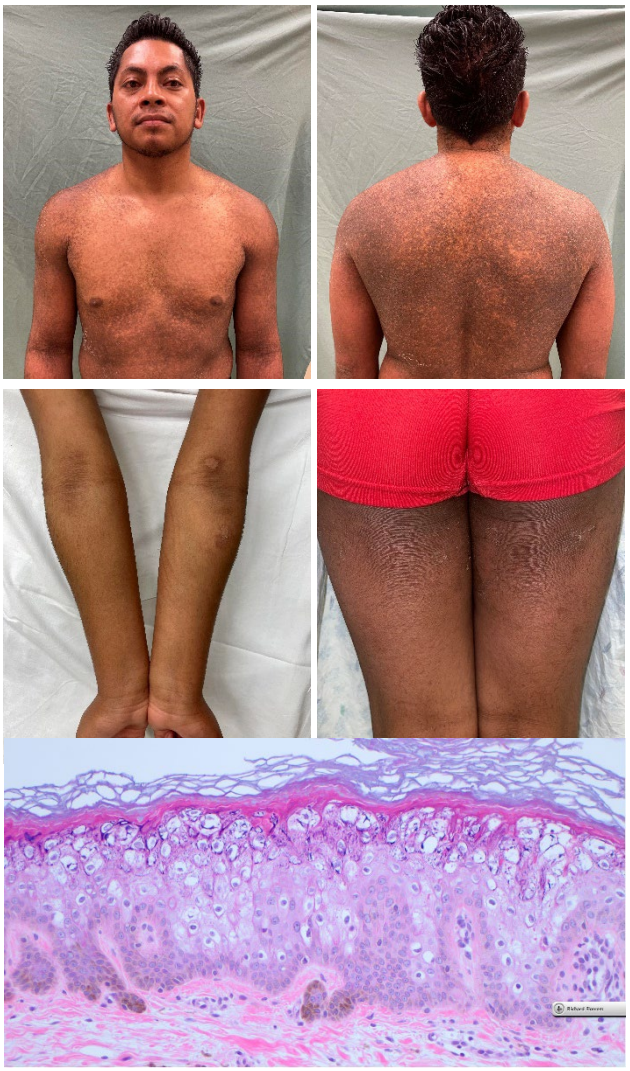
CLINICAL COURSE

Based on the clinical findings of skin fragility and peeling, absence of pigmentary alterations in first degree relatives with similar findings of skin fragility, and the histopathology showing epidermolysis, a concomitant diagnosis of superficial epidermolytic ichthyosis (SEI) and generalized DDD is favored. The pigmentary alterations have not improved with a trial of a combination of hydroquinone, kojic acid, niacinamide, and vitamin C cream.

DISCUSSION

DDD is an autosomal dominant condition with adolescent or adult-onset, characterized by symmetric, reticulated hyperpigmentation of the flexures.

The four genes implicated in varying etiologies of DDD are *KRT5, POFUT1,* and *PSENEN. POFUT1*, *POGLUT1*, and *PSENEN* encode proteins involved in Notch signaling, which are involved in melanocyte and keratinocyte proliferation and differentiation. Mutations in *POFUT1* are associated with generalized hyperpigmentation. *POFUT1* encodes protein O-fucosyltransferase 1 which adds O-fucose to proteins, including the Notch receptor.



One unusual feature of this case is the absence of a mutation in the coding region of *KRT2* which causes SEI. We hypothesize that the causative mutation may be in a non-coding region of *KRT2* or that the mutation in *POFUT1* may contribute to this family's phenotype. To our knowledge, there are no cases describing the specific *POFUT1* variant identified in our patients or prior reports of *POFUT1* variants causing a SEI-like phenotype.

REFERENCES

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2. Li et al. Mutations in POFUT1, encoding protein-O-fucosyltransferase 1, cause generalized Dowling-Degos disease. Am J Hum Genet. 2013;92(6):895-903.

CASE 13: Violaceous Papules on the Limbs of a 10-year-old girl

Authors: Celter Odango MS, Lauren Yi MD, Barrett Zlotoff MD

HISTORY

A 10-year-old girl presented with a one-year history of a pruritic, pustular rash on the legs. The rash started on her ankles and spread to involve her face, trunk, and arms towards her torso, face, and arms. The patient also reported six months of abdominal pain, hematochezia, and oral ulceration.

PHYSICAL EXAMINATION

Scattered violaceous papules with post-inflammatory hyperpigmentation and scarring were present on the patient's arms and legs. There was an ulceration with heaped-up mucosa on her left lower mucosal lip.

PATHOLOGY

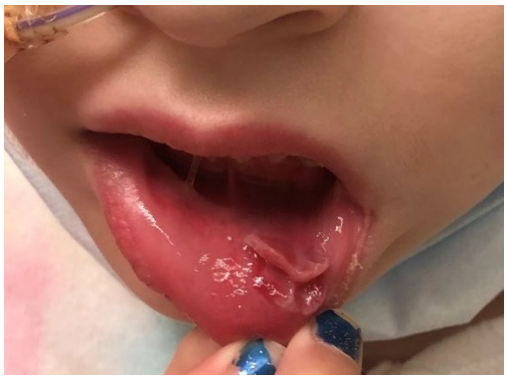
A biopsy from the patient's left leg revealed lichenoid interface dermatitis and granulomatous inflammation with epithelioid histiocytes and multinucleated giant cells within the superficial dermis.

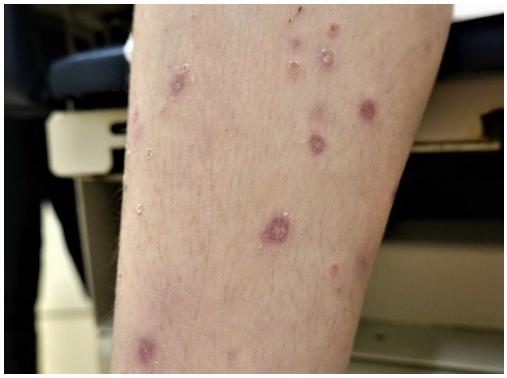
CLINICAL COURSE

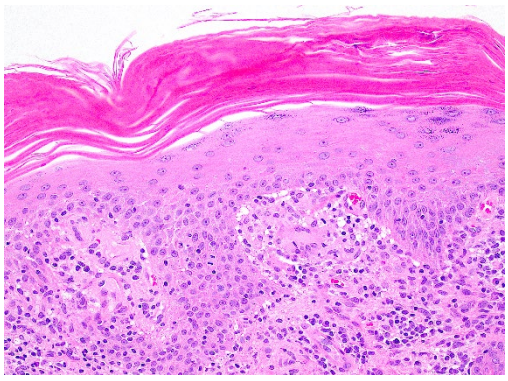
The patient underwent upper and lower endoscopy with small intestinal biopsies confirming a diagnosis of Crohn disease (CD). Her skin manifestations were favored to be cutaneous CD. She was started on systemic steroids and infliximab with resolution of rash and improvement in her gastrointestinal symptoms.

DISCUSSION

CD is a chronic granulomatous inflammatory disease of the gastrointestinal tract.[1] Extra-intestinal manifestations can be observed in some children, with cutaneous CD being the most common. Among the patterns of cutaneous involvement, metastatic CD (MCD) is the rarest form and is characterized by inflammation discontinuous with the gastrointestinal tract. Its cutaneous manifestations include erythematous and violaceous papules and plaques, edema, ulcerations, crusts, erosions, and perifollicular papules. The histopathologic findings are also highly variable, with granulomatous dermatitis being a consistent feature. Lichenoid interface dermatitis, as seen in this patient, is a less common feature seen in MCD. This case highlights the diverse clinical and histopathologic presentations of MCD.[2]







REFERENCES

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2. Yi et al. Violaceous papules on the limbs of a 10-year-old girl. *Pediatr Dermatol*. 2024;41(4):741-743.

CASE 14: Familial Reactive Perforating Collagenosis

Authors: Kristy Tefft MS, Lauren Yi MD, R. Hal Flowers MD

HISTORY

A 45-year-old Hispanic male with diabetes mellitus type 2 and hypertension presented for pruritic papules and nodules on the right forearm and right face. The lesions had developed after local superficial trauma. He endorsed numerous similar lesions occurring at the site of cutaneous trauma intermittently since childhood. He denied systemic symptoms, history of HIV, significant infections, or immunocompromise. Family history was notable for similar symptoms in two siblings.

PHYSICAL EXAMINATION

Three one-centimeter pink papules with central hyperkeratotic cores were present on the right forearm. A solitary one cm pink nodule with central keratotic core was noted on the right lateral oral commissure.

PATHOLOGY

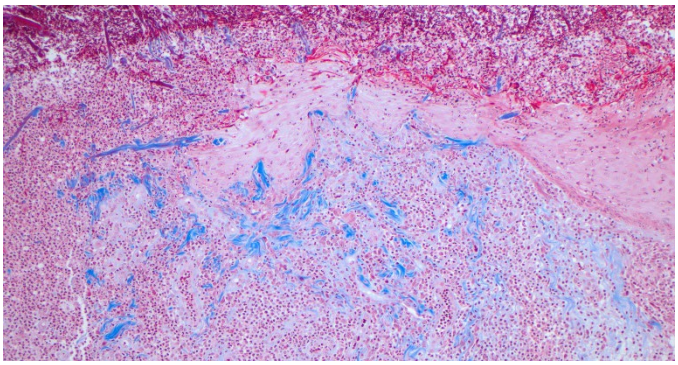
Shave biopsies from the right arm and right face demonstrated hyperkeratosis overlying a cup-shaped invagination of epidermis with focal ulceration. There was associated mixed inflammation with keratin debris. Marked dermal acute inflammation and abscess formation was observed in the specimen from the right oral commissure. Trichrome staining highlighted transepidermal elimination of collagen fibers in both biopsies.

CLINICAL COURSE

A trial of acitretin 10 mg daily was initiated. Patient is pending follow up.

DISCUSSION

Reactive perforating collagenosis (RPC) is characterized clinically by self-remitting papules with a central keratotic plug arising at the site of minor trauma. Lesions may be pruritic or cause scarring but are otherwise benign. The most common form of RPC is acquired in the setting of end stage renal disease and diabetes but more rarely may be inherited.[1] The exact etiology is unknown. Histopathology characteristically demonstrates a cup-shaped epidermal invagination and transepidermal extrusion of vertically aligned collagen fibers, which can be highlighted with trichrome stain to differentiate from elastosis perforans serpiginosa.



Treatment is challenging and aimed at minimizing recurrences and symptoms. Various approaches including oral and topical retinoids, antibiotics, and methotrexate have been trialed; however, evidence of efficacy is limited.[2]

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CASE 15: VEXAS with neutrophilic eccrine hidradenitis

Authors: Celter Odango MS, Lauren Yi MD, R. Hal Flowers MD

HISTORY

A 57-year-old male with hypertension presented with a one-year history of recurrent fevers and recurrent rash on the head, neck, and extremities. He had also recently been diagnosed with a B cell lymphoproliferative disorder.

PHYSICAL EXAMINATION

There were numerous edematous, annular red papules, plaques, and nodules on the head, neck, and chest.

PATHOLOGY

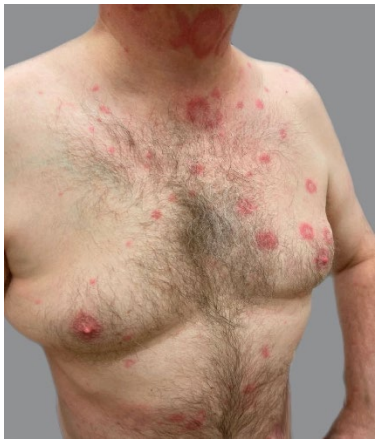
Multiple punch biopsies were performed throughout the course of the patient's condition which revealed dermal edema, neutrophilic inflammation extending into eccrine glandular epithelium, and leukocytoclasia consistent with neutrophilic dermatosis with neutrophilic eccrine hidradenitis (NEH).

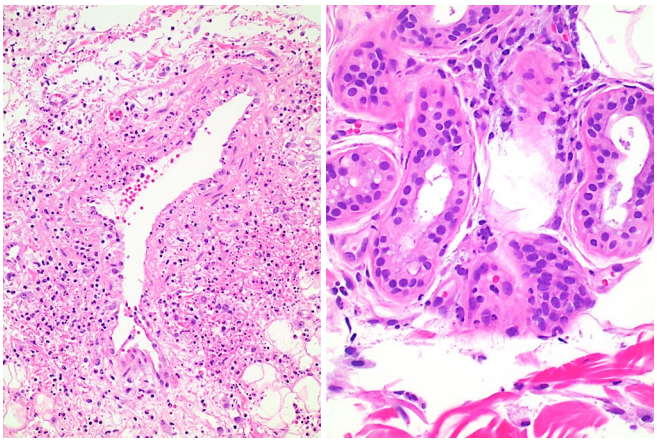
TREATMENT

The patient was initially treated with systemic prednisone with clearance of rash and resolution of fevers. Due to the recalcitrant nature of his condition and failure of numerous agents including but not limited to dapsone, colchicine, potassium iodide, and anakinra, serum genetic testing was conducted which identified a pathogenic variant in the *UBA1* gene, confirming VEXAS (vacuoles, E1 enzyme, X-linked, auto-inflammatory) syndrome. The patient was transitioned to adalimumab, which resulted in improvement in both the severity and frequency of his flares.

DISCUSSION

VEXAS syndrome is an autoinflammatory condition caused by an acquired *UBA1* gene mutation, leading to multi-organ involvement.[1] Skin manifestations, including neutrophilic dermatoses, occur in most patients. Vasculitis and periorbital edema are also common. Hematological conditions such as cytopenias and myeloid neoplasms are more commonly reported than lymphoid malignancies and lymphoproliferative disorders. Rheumatologic features typically include chondritis, arthritis, and scleritis. Diagnosis is confirmed through molecular sequencing identifying the *UBA1* mutation. The majority of histopathologic examinations in VEXAS syndrome typically show neutrophilic dermatosis, leukocytoclastic vasculitis, and neutrophilic urticarial dermatosis. Neutrophilic eccrine hidradenitis has not been previously described but likely represents an extension of neutrophilic inflammation.[2]





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CASE 16: *PTCH1* mutation associated encephalocraniocutaneous lipomatosis-like phenotype

Authors: Heli Patel MD, Lauren Yi MD, Barrett Zlotoff MD

HISTORY

An eleven-day-old male born at 37 weeks via cesarean presented for evaluation of nodules on the back that were present since birth. Pregnancy was complicated by gestational diabetes. Past medical history was notable for bilateral undescended testes.

PHYSICAL EXAMINATION

There was a smooth, lobulated hypopigmented plaque over the right temporal scalp with associated alopecia consistent with either nevus sebaceous or nevus psiloliparis. There were skin-colored papules over the right lower eyelid, right upper lip, and right lower vermillion border. Numerous ovoid blue patches with overlying epidermal atrophy and palpable nodules within were noted over the back.

IMAGING/PATHOLOGY/LABORATORY DATA

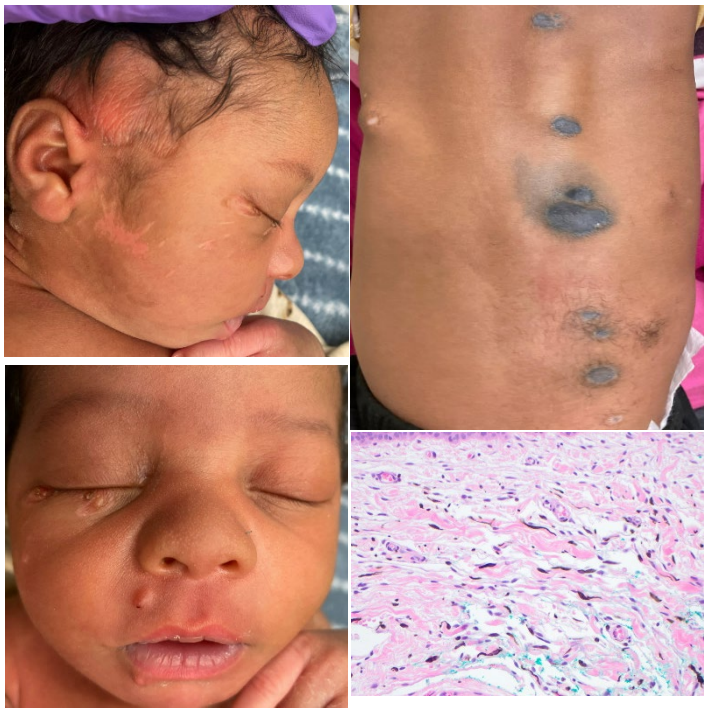
X-ray of the patient's back identified calcification within the back nodules. A punch biopsy from a blue patch on the right upper back showed deeply pigmented melanocytes and melanophages dissecting through bundles of collagen in reticular dermis consistent with dermal melanocytosis. Pathology from the excision of the papule on the right lower eyelid and right lower lip showed epidermal acanthosis. Whole exome sequencing from buccal swab identified no pathogenic variants. Sequencing of tissue from the right lower eyelid identified two pathogenic variants in *PTCH1*. There was insufficient tissue to perform genetic testing on the melanocytic lesion.

CLINICAL COURSE

The patient was diagnosed with congenital right sided hemihypertrophy, global developmental delay and an asymmetric brainstorm malformation. He has been referred to Pediatric Surgery for excision of painful nodules on his back.

DISCUSSION

The differential diagnosis for his presentation includes a segmental form of encephalocraniocutaneous lipomatosis (ECCL), SCALP syndrome (sebaceous nevi, central nervous system malformations, aplasia cutis congenita, limbal dermoid, and pigmented nevi/giant congenital melanocytic nevi), and epidermal nevus syndrome. All are multisystem disorders due to underlying mutations in the RAS signaling pathway.



Inactivating *PTCH1* mutations result in constitutive activation of the hedgehog pathway and are classically associated with basal cell nevus syndrome and basal cell carcinoma. However, there is a single case report of epidermal nevus syndrome due to an underlying germline *PTCH1* mutation.[1] How this mutation could result in this phenotype is unclear, but hedgehog signaling is hypothesized to cooperate with the RAS/MAPK signaling pathway.[2] To our knowledge, this is the first report of this constellation of findings in association with a *PTCH1* mutation. Confirmatory genetic testing on additional tissue is planned.

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CASE 17: PHACE syndrome complicated by stroke following propranolol

Authors: Emily Tocco BS, Margaret Ann Kreher MD, Barrett Zlotoff MD

HISTORY

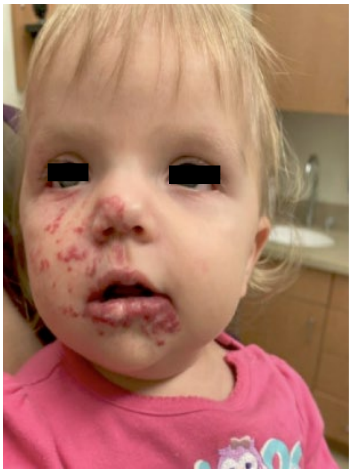
A 4-week female with agenesis of the corpus callosum, ventriculomegaly, ectatic right internal carotid artery, and extensive cerebrovascular malformations (discovered prenatally) presented to dermatology clinic for possible PHACE syndrome. She had a faint red patch one week after birth extending over the right cheek, orbit, lip, scalp and posterior neck that darkened and thickened.

PHYSICAL EXAMINATION

Examination revealed beefy red broken plaques over the S1-S3 and scalp distribution, consistent with segmental infantile hemangioma (IH). The IH extended onto the posterior scalp and neck with surrounding greyish discoloration concerning for impending ulceration.

RADIOLOGY

MRI revealed a tortuous right internal carotid artery and atretic left internal carotid artery with minimal intracranial flow signal. The right posterior cerebral artery appeared to terminate in a cluster of peri-mesencephalic vessels.



TREATMENT

The patient was started on a trial of propranolol and prednisolone for the IH, as well as aspirin for stroke risk reduction. Shortly after initiation, she was noted to have episodes of left gaze deviation with seizure activity on EEG, and repeat brain MRI/MRA showed new small parietal infarcts. Propranolol was immediately discontinued, and Keppra was started. Repeat brain MRI showed enlarged areas of diffusion restriction in the frontal and parietal lobes concerning for stroke vs post-ictal changes. She was stabilized prior to discharge, and propranolol was not restarted. Subsequently, at age 5 months, she underwent cerebral angiogram with embolization of the right superficial temporal artery resulting in reduction of the IH. She completed a long taper of prednisolone and on follow-up had significant flattening of the IH without rebound.

DISCUSSION

There have been no reported cases of oral propranolol causing stroke in patients with PHACE syndrome. The timing of propranolol in relation to symptoms and imaging changes is concerning in this case. Alternatively, the patient's compromised cerebral blood flow due to arterial abnormalities might have led to these complications independent of propranolol. The successful use of embolization in the debulking of the patient's segmental hemangioma underscores its value as a treatment when propranolol is contraindicated. Additional alternative treatments include vincristine, interferon alfa, and more recently, sirolimus, all of which have demonstrated efficacy with less favorable side effect profile than propranolol, atenolol or nadolol.

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CASE 18: CM-AVM Syndrome Secondary to RASA-1 Mutation

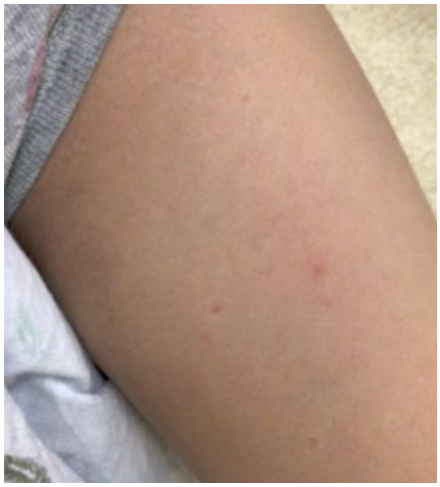
Authors: Emily Tocco BS, Margaret Ann Kreher MD, Barrett Zlotoff MD

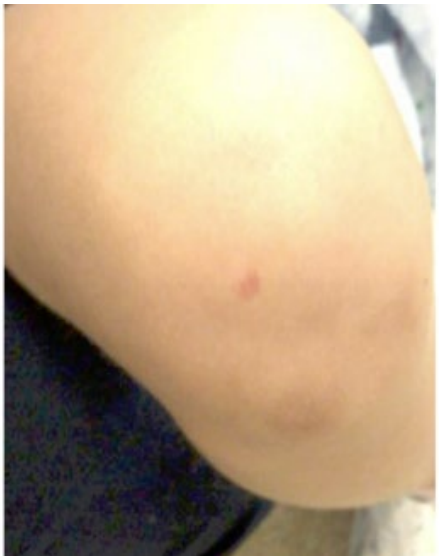
HISTORY

A 2-year-old female presented for management of previously diagnosed CM-AVM syndrome. At 19 months old, she had an episode of left arm swelling, pain, and decreased range of motion. Evaluation at that time showed an AVM in the triceps muscle and a small venous anomaly in the right cerebellum. Genetic testing was performed revealing RASA1 mutation, presumed to be de novo.

PHYSICAL EXAMINATION

Examination revealed a slightly hypertrophic left arm compared to the right arm (9 cm vs. 6 cm) with increased warmth. Thumbprint-type cutaneous capillary malformations were present on the cheek, chest, bilateral upper and lower extremities.





RADIOLOGY

MRI showed a small to moderate developmental venous anomaly in the right cerebellum and no vascular abnormalities in the spine. MRI of the left humerus showed several serpentine flow-voids primarily centered in the triceps muscle consistent with AVM. Echocardiogram was performed to aid in future monitoring for high-output heart failure; agitated saline study was suggestive of shunting and intra-pulmonary AVMs.

TREATMENT

The patient underwent successful arteriography and embolization of the left arm AVM without complication. Following the embolization, she experienced an improvement in size, warmth, and range of motion, and she has not required further intervention for the AVM of the arm. Treatment was not indicated for the apparent intra-pulmonary AVMs as the patient was asymptomatic.

DISCUSSION

CM-AVM syndrome is an autosomal dominant disorder characterized by capillary malformations, arteriovenous malformations (AVMs), and/or arteriovenous fistulas (AVFs). Germline loss-of-function mutations in RASA1 have been linked to CM-AVM; there also appears to be a significant role for somatic "second hits" in the development of vascular anomalies in these patients, and mosaic mutations in RASA1 may cause clinically evident CM-AVM without a germline mutation.[1,2] In our case, hypertrophy of the patient's arm points to a more specific diagnosis of Parkes-Weber syndrome (PWS), which involves limb overgrowth in conjunction with vascular malformations and is also associated with RASA1 mutations. An interesting finding in this case was the associated pulmonary AVMs. There are no established guidelines for cardiopulmonary screening in CM-AVM, as pulmonary AVMs are uncommon. However, asymptomatic pulmonary AVMs may occur more frequently than previously thought, thus close monitoring for symptoms is warranted.

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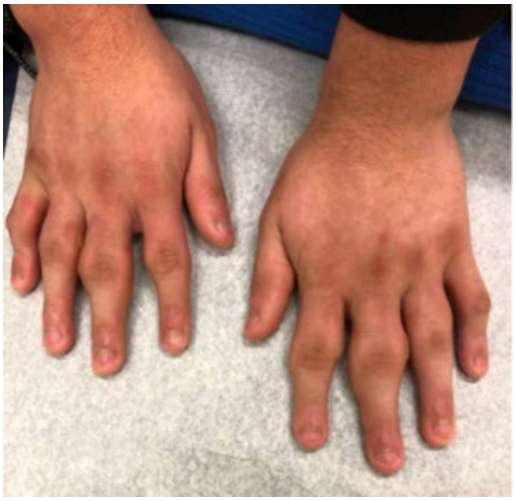
CASE 19: Pachydermodactyly

Authors: Olivia Lim BS, Margaret Ann Kreher MD, Barrett Zlotoff MD

HISTORY

An 11-year-old boy who had previously been diagnosed with psoriasis presented for swelling and scaling of the third, fourth, and fifth digits of his bilateral hands. He endorsed mild pruritis and intermittent pain in his proximal interphalangeal (PIP) joints, which he repetitively scratched and manipulated. He had no functional limitations due to joint stiffness or pain.



PHYSICAL EXAMINATION

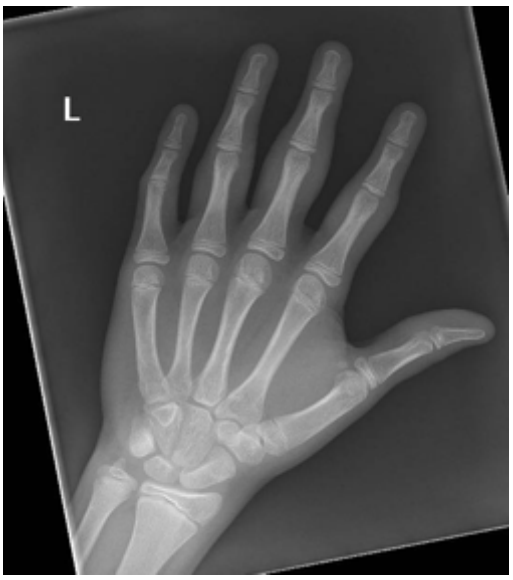
Examination showed symmetrical swelling of the PIP joints of the second, third, and fourth digits bilaterally with lichenified, erythematous, scaly plaques overlying the affected joints and tapering and sclerodactyly changes on the distal fingers.

RADIOLOGY

MRI and X-rays were negative for any psoriatic arthritis changes.

TREATMENT

The patient's presentation was consistent with pachydermodactyly complicated by lichen simplex chronicus, likely previously mistaken for psoriasis by an outside provider. Topical corticosteroids were first tried without improvement. Urea cream and pimecrolimus cream were next added with minimal benefit. Intralesional triamcinolone was attempted but limited by extensive fibrotic tissue. Ultimately, oral tranilast 300 mg daily was started, and follow-up is scheduled to assess response.



DISCUSSION

Pachydermodactyly is a rare, benign form of digital fibromatosis characterized by soft tissue swelling around the PIP joints without underlying bony changes on MRI or x-ray. It is often associated with repetitive mechanical trauma of the periarticular skin in adolescent males and may mimic psoriatic arthritis or juvenile idiopathic arthritis clinically. Thus, in a patient with enlarged PIP joints and exam findings suspicious for psoriatic arthritis, but without true arthritic pain or characteristic changes on imaging, pachydermodactyly should be considered. This condition is not rheumatic, and autoantibodies tend to be negative. Histopathologic examination typically reveals an increase in dermal collagen with fibroblast proliferation, variable overlying hyperkeratosis, and no inflammatory infiltration. Tranilast, an antiallergic drug which has been used in the treatment of keloids, may represent an effective therapeutic agent due to its inhibition of collagen synthesis. Further studies are needed to assess its efficacy in pachydermodactyly.

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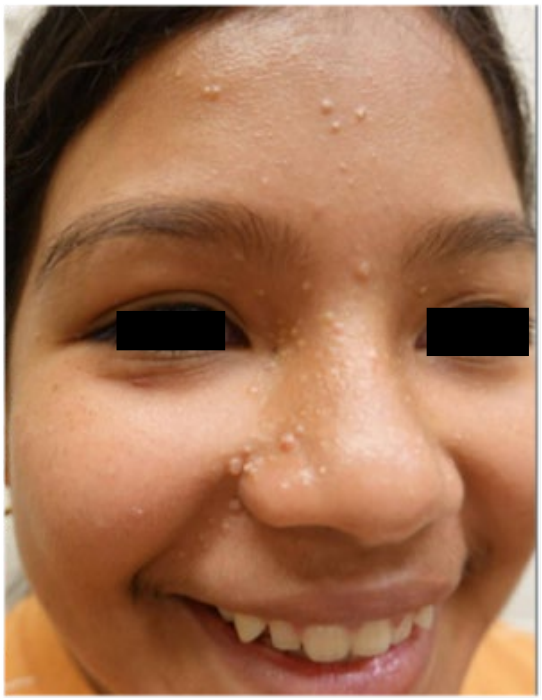
CASE 20: Multiple Familial Trichoepitheliomas

Authors: Olivia Lim BS, Margaret Ann Kreher MD, Barrett Zlotoff MD

HISTORY

A 16-year-old female with a past medical history of acne vulgaris presented for evaluation of bumps on the face. She had a family history of similar lesions in her father and sister. Review of systems was negative for seizure-like activity, other birthmarks, or nail involvement.

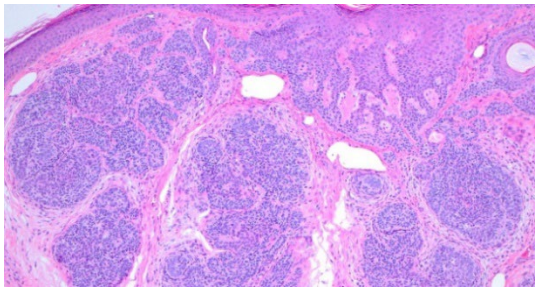


PHYSICAL EXAMINATION

Exam revealed multiple discrete, confluent, flesh-colored to white papules on the central face overlying the forehead, glabella, medial canthus, nasal bridge, and nasolabial folds. No cylindromas or spiradenomas were found on exam.

PATHOLOGY

Biopsy of a representative lesion revealed nodular, well-circumscribed growth of basaloid cells suspended within fibrous stroma, consistent with trichoepithelioma.



TREATMENT

Treatment with tretinoin 0.1% cream nightly and imiquimod 5% cream three times a week was prescribed in September 2020, with no improvement. Given the low risk of malignant transformation reported with familial trichoepitheliomas, the option for longitudinal surveillance was discussed.[1] In August 2023, the patient was started on sirolimus 1% BID for 9 months and was referred for ablative laser treatment. As of May 2024, the patient has had no response to the sirolimus and has not pursued laser treatment due to potential scarring.

DISCUSSION

Trichoepitheliomas are benign epithelial tumors composed of follicular germ cells that present as smooth, non-ulcerated skin-colored papules with occasional telangiectasias. The presence of numerous trichoepitheliomas should raise suspicion for Brooke-Spiegler Syndrome. The lack of cylindromas, spiradenomas, or systemic involvement makes the patient's diagnosis most consistent with multiple familial trichoepitheliomas (MFT1). These disorders are due to an autosomal dominant germline mutation in the cylindromatosis (*CYLD*) gene on chromosome 16q12.[1] *CYLD* mutations are detected in roughly 40-50% of patients with MFT1.[1] Malignant transformation can occur within 5-10% of affected persons. While destructive therapies are the mainstay of treatment, topical tretinoin, imiquimod, or sirolimus have also been used.[2] Monthly self-skin surveillance, decreased sun exposure, and frequent dermatology follow-ups are vital to minimize the potential for malignant transformation and disfiguration.

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CASE 21: Telangiectasia Macularis Eruptiva Perstans

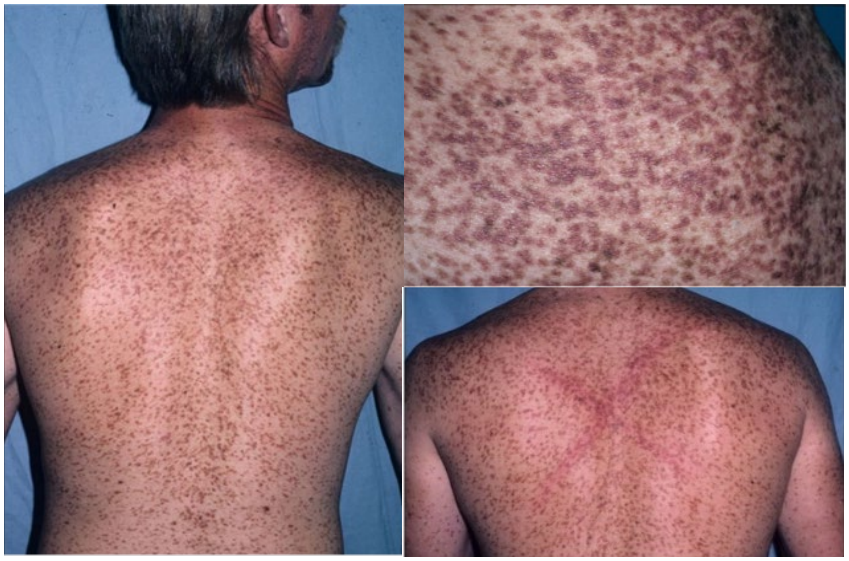
Authors: Max Stempel BS, Margaret Ann Kreher MD, Kenneth Greer MD

HISTORY

A 27-year-old male initially presented to dermatology clinic for evaluation of a persistent cutaneous eruption. He reported several months of an eruptive rash of erythematous macules which had become more diffuse and generalized. He had mild intermittent pruritis and noticed that the lesions became raised with pressure or trauma.

PHYSICAL EXAMINATION

Examination showed a diffuse eruption of irregular reddish-brown telangiectatic macules and thin papules, most prominently on the trunk and proximal extremities. Darier's sign was positive.



PATHOLOGY

Biopsy of a new erythematous macule on the chest revealed a patchy interstitial and superficial perivascular lymphocytic infiltrate. Immunohistochemical staining for CD117 was noncontributory. Leder stain showed an increased number of mast cells in the superficial dermis, consistent with urticaria pigmentosa (UP) or telangiectasia macularis eruptive perstans (TMEP).

TREATMENT

Treatment was aimed at reducing flares through avoidance of triggers such as excessive heat and alcohol. The patient used oral antihistamines regularly and mid-potency topical corticosteroids for flares. The macules persisted chronically with gradual improvement over time. At 20-year follow-up, scattered macules were still present that urticated with pressure. By 30-year follow-up, his skin was 95% clear.

DISCUSSION

TMEP is a rare variant of cutaneous mastocytosis (CM) characterized by reddish brown telangiectatic macules typically distributed on the trunk and proximal extremities. The lesions correspond to a subtle increase in mast cells in the superficial dermis and dilated superficial capillaries. TMEP is distinguished from other forms of CM by adulthood onset, telangiectatic morphology, and negative or less pronounced Darier's sign due to relatively less mast cell involvement. TMEP can be a diagnostic challenge due to the spectrum of clinical presentations described in the literature. Of note, TMEP frequently has some degree of overlap with UP, as seen in this case, there have been reports of associated systemic mastocytosis in cases of TMEP/UP overlap. Thus, close monitoring for systemic involvement is recommended. Further studies are warranted to differentiate TMEP from UP biologically and prognostically to help guide management and appropriate screening and counseling.

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CASE 22: Nevoid basal cell carcinoma syndrome with SUFU mutation

Authors: Catherine Lyons BS, Nicole Edmonds MD, R. Hal Flowers MD

HISTORY

A 30-year-old female patient with a history of medulloblastoma at age 2 months, status post resection and chemotherapy complicated by end stage renal disease requiring kidney transplant at age 13, ovarian fibrosis status post bilateral salpingectomy at age 9, meningioma status post excision at age 24, diabetes, and seizure disorder, presented for a routine annual skin check. She had been taking mycophenolate mofetil, tacrolimus, and prednisone for immunosuppression. The patient was adopted with no known family history.

PHYSICAL EXAMINATION

Numerous small flesh-colored papules with sharp vessels were noted in a periocular and perinasal distribution, with few subtle palmar pits.

PATHOLOGY/LABORATORY DATA

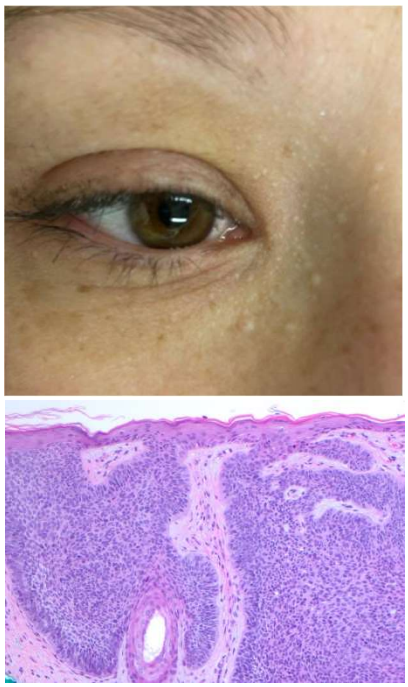
A right medial lower eyelid biopsy showed basal cell carcinoma (BCC), prompting additional biopsies of similar papules on her left vertex scalp, and central upper back which were all consistent with basal cell carcinoma. Serum testing confirmed a pathogenic SUFU mutation.

TREATMENT

The presence of the SUFU mutation, as well as the multiple basal cell carcinomas, medulloblastoma and ovarian fibrosis, led to the diagnosis of nevoid basal-cell carcinoma syndrome (NBCCS), or Gorlin's syndrome. The patient's numerous eyelid BCCs were treated with topical imiquimod then photodynamic therapy. The scalp BCC was treated with Mohs surgery and the back BCC was treated with shave and curettage. To minimize further cancer risk, the patient's dose of mycophenolate mofetil was reduced per her transplant team. She follows with dermatology every 4 months.

DISCUSSION

NBCCS occurs as a result of abnormal activation of the sonic hedgehog pathway.[1] The most commonly affected gene in NBCCS is PTCH1. Uncommonly, a suppressor of the SUFU gene, which is downstream from PTCH1, can cause NBCCS. NBCCS is associated with medulloblastoma (20 times more common in patients with SUFU mutations), meningiomas (which can cause seizures), BCC, and ovarian fibrosis, as seen in this patient.[1] Sonic Hedgehog (SHH) inhibitors like sonedigib and vismodegib would unlikely have any benefit in the setting of the SUFU mutation as it is downstream of the target SMO.[2] Other systemic treatment options include itraconazole which has been shown to have benefit in NBCCS irrespective of the mutation, though this was not a viable option in this case given the patient's seizure history requiring levetiracetam use. Aside from medulloblastomas, patients with SUFU mutations generally have a milder NBCCS phenotype, often lacking typical jaw cysts, and thereby making the diagnosis more challenging. Early recognition of this uncommon subtype can prevent diagnostic delay.



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CASE 23: Multi-treatment resistant PLEVA

Authors: Catherine Lyons BS, Nicole Edmonds MD, Thomas Cropley MD

HISTORY

An 18-year-old male presented to UVA ED with a 3-month history of a widespread papular rash with skin and mucosal involvement. He had been treated by various urgent care centers with steroids, oral antibiotics, and oral antivirals. His rash progressed from his trunk to involve his extremities and mucosal surfaces of the penis and mouth, prompting his admission to an outside hospital for IV antibiotics and antivirals. Following discharge, he initiated methotrexate 15 mg weekly and phototherapy, but his rash continued to progress, and he developed a fever of 100.6 F prompting his presentation to the UVA ED.

PHYSICAL EXAMINATION

On presentation, he was afebrile but tachycardic. Extensive, crusted, purpuric papules and papulovesicles were noted on exam predominantly on the neck, trunk, bilateral inguinal folds, groin, and face. There were no clear target lesions, but significant denuded and erosive areas were noted on the chest as well as the bilateral inguinal folds with superficial erosions along glans penis and penile shaft. There was also hemorrhagic crust on the vermillion border of upper and lower lips, with superficial ulcers on the hard palate.

PATHOLOGY

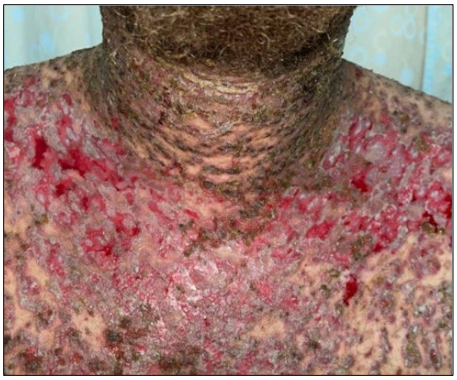
Prior biopsy showed a superficial perivascular dermatitis with a lymphocyte predominant dermal infiltrate with extravasated erythrocytes.

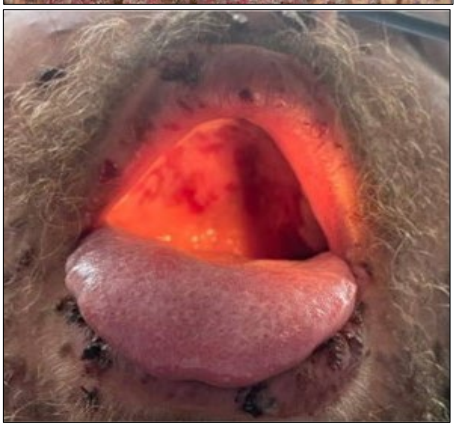
TREATMENT

Pathology findings as well as clinical evaluation supported a diagnosis of PLEVA, a subtype of pityriasis lichenoides. The causative agent of suspected PLEVA was investigated with rapid plasma regain, HIV antibody and antigen, and HSV serology, all of which were negative. His presentation also raised concern for progression to febrile ulceronecrotic Mucha-Habermann disease (FUMHD) given the acute worsening of the rash, fever, and tachycardia.[1,2] The patient was treated with ceftriaxone for the presumed superinfection and resumed methotrexate (MTX) therapy on discharge at a higher dose of 20 mg weekly. Within two months, the higher dose had significantly improved his condition (Figure D) and he continued to clear on this therapy with only minimal residual PIH on exam.

DISCUSSION

The initial differential diagnosis included lymphocytic vasculitis secondary to systemic lupus or angioimmunoblastic T-cell lymphoma, PLEVA/ FUMHD, and SJS-TEN. ANA, anti-dsDNA, ANCAs, peripheral blood flow cytometry for B- and T-cell immunotyping, SPEP were all non-diagnostic. Treatment for PLEVA and FUMHD depends on the underlying cause, but great success has been achieved with oral antibiotics, systemic corticosteroids, and additional therapies such as methotrexate, IVIG, and NBUVB.





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CASE 24: Keratosis Lichenoides Chronica

Authors: Olivia Lim BS, Nicole Edmonds MD, Kenneth Greer MD

HISTORY

A 42-year-old woman with a past medical history of keratosis lichenoides chronica (KLC) diagnosed at 15 years old presents in clinic for ongoing KLC refractory to methotrexate, Accutane, acitretin, aldara, dovonex, topical steroids, and laser treatment. She notes that she has had improvement in the hyperkeratosis with topical tea tree oil. She reports that initial involvement began on acral surfaces and progressed to include her legs, arms, trunk, and face.

PHYSICAL EXAMINATION

On exam, purple hyperkeratotic papules and nodules arranged linearly with overlying excoriation were noted on the dorsal hands and flexor wrists. Additionally, reticular, violaceous plaques with overlying crust were present on her bilateral dorsal feet and toes with minimal involvement of the trunk, legs, upper arms, chest. Erythematous papules with overlying greasy scale were also present on the cheeks

PATHOLOGY

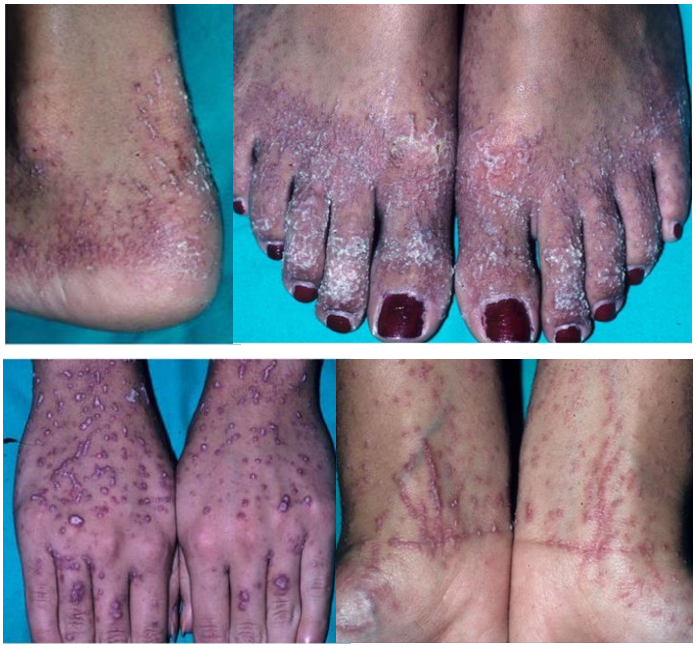
This patient had a biopsy as a child at an outside facility so we did not have access to the report. Typical KLC lesions are characterized by vacuolar degeneration of keratinocytes at the dermo-epidermal junction, numerous necrotic keratinocytes in the epidermis, chronic inflammatory infiltrate in the papillary dermis consisting of lymphocytes, histiocytes and plasma cells, and vascular dilatation.

TREATMENT

She has treated with dapsone 50 mg since January 2024 with improvement of her blepharitis and oral ulcers. She continues to have KLC on her arms, hands, and thighs. For blepharitis flares, she uses fluoromethalone 0.025% eye drops and lidocaine 2% mouthwash for recurrent aphthous ulcers.

DISCUSSION

Keratosis lichenoides chronica (KLC), also known as Nekham's disease, is an exceedingly rare skin condition characterized by linear and reticular scaly papules and thin plaques. It can be associated with blepharitis, a rash mimicking seborrheic dermatitis or rosacea on the face, palmoplantar keratoderma, ungual dystrophy, and mucocutaneous ulcers.[1] Some cases have been linked to germline NLRP1 mutation, which activate inflammatory cytokines.[2] KLC is extremely difficult to treat, but there are reports of improvement with PUVA and acitretin either alone or in combination.[1] Most recently, a case has been reported of substantial improvement with upadacitinib.[3] Nevertheless, complete resolution of KLC has never been reported and treatment is best aimed at its associated conditions.



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CASE 25: Cutaneous sarcoidosis mimicking squamous cell carcinoma

Authors: Catherine Lyons BS, Nicole Edmonds MD, R. Hal Flowers MD

HISTORY

A 60-year-old female patient with a history of multiple basal and squamous cell carcinomas (SCCs) on her face and body as well as pulmonary and ocular sarcoidosis presented for Mohs micrographic surgery of a SCC on the right dorsum of her nose.

PHYSICAL EXAMINATION

Prior to the procedure, the Mohs surgeon noted a reddish-brown indurated plaque of the right nasal sidewall and distal nose, distorting the architecture of the right nose. Due to concern that this lesion was an alternative diagnosis than SCC, the Mohs surgeon re biopsied the lesion.

PATHOLOGY/RADIOLOGY

The repeat biopsy showed pseudoepitheliomatous hyperplasia (PEH) and noncaseating granulomas, which based upon the clinical history, most likely represented cutaneous sarcoid. Review of chest CT findings from 2013 showed bilateral hilar and mediastinal lymphadenopathy, some of which demonstrate associated calcifications, as well as poorly defined ground glass centrilobular micronodules and scattered 2mm solid nodules in the right upper lobes, consistent with sarcoidosis.

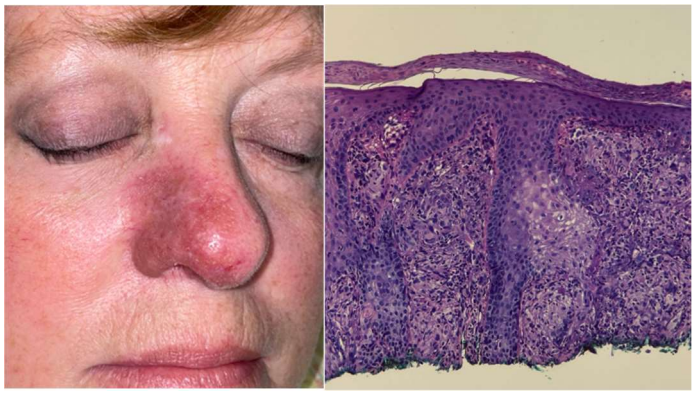
TREATMENT

The granulomatous inflammation in the initial biopsy was initially attributed to SCC. The accompanying PEH on repeat biopsy coupled with the clinical exam and history of pulmonary sarcoidosis and uveitis led to a re-diagnosis of cutaneous sarcoidosis. The patient was initially treated with topical steroids with no effect so was subsequently transitioned to hydroxychloroquine. She was also referred to pulmonology to re-establish care.

DISCUSSION

Pathology findings for sarcoidosis classically include noncaseating granulomas.[1] PEH is a benign, reactive proliferation of the epidermis that can mimic SCC due to its thickened, irregular growth of the epidermal layer with elongated rete ridges but is generally differentiated from SCC by the lack of cellular atypia.[2] PEH can be seen in various conditions often as a response to chronic irritation, trauma or infection. Though uncommon, PEH has been reported in cutaneous sarcoid.[2] Because PEH can be difficult to distinguish from SCC, there have been other cases in the literature of patients misdiagnosed with SCC in lieu of sarcoid.[1]

Cutaneous sarcoidosis commonly involves red-brown to purple-brown papules and plaques. Lupus pernio is a subtype of cutaneous sarcoidosis that involves violaceous, firm, indurated plaques most commonly on the face, as in this case.[1] Lupus pernio compared to other cutaneous sarcoid lesions more often have a more chronic course, are resistant to treatment, and result in scarring and disfigurement. Treatment of cutaneous sarcoid includes topical/intralesional/systemic steroids, systemic agents such as hydroxychloroquine or methotrexate, and TNF-alpha inhibitors.[1] Patients with lupus pernio often require more aggressive treatment with systemic agents to resolve the plaques. Methotrexate or adalimumab are being discussed for our patient currently.



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CASE 26: Diffuse xanthomas associated with lipoprotein X HLD

Authors: Maya Hagander BS, Nicole Edmonds MD, R. Hal Flowers MD

HISTORY

A 30-year-old woman with primary sclerosing cholangitis (PSC) and Crohn's disease presented with pruritic skin lesions on her shoulders and arms for three weeks. Her dermatologic history was notable for previously diagnosed reactive perforating collagenosis and granuloma annulare.

PHYSICAL EXAMINATION

Coalescing yellow to hypopigmented papules and small plaques were present, occasionally in annular configuration, on her neck, shoulders, and upper arms.

PATHOLOGY

Punch biopsy showed a brisk proliferation of xanthomatized histiocytes in papillary, mid, and reticular dermis with foci of necrobiosis and mucin deposition. Extracellular lipid deposition was present. The patient was diagnosed with cutaneous xanthomas with concomitant granuloma annulare (given the mucin deposition).

Lipid panel revealed cholesterol level of 876 mg/dL and low-density lipoprotein (LDL) of 840 mg/dL. TG were within normal limits.

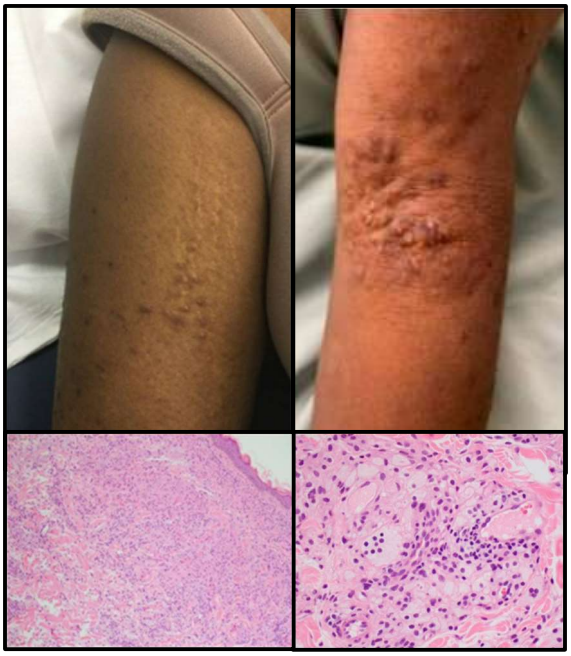
At follow up, the xanthomas had become larger and progressed to involve the eyelids, upper chest, extremities, and abdomen. Cholesterol and LDL had increased to 2011 mg/dL and 1938 mg/dL, respectively.

TREATMENT

Given the cutaneous/laboratory findings as well as history of PSC, the patient was diagnosed with cutaneous xanthomas in the setting of LpX mediated hyperlipidemia. Biweekly plasmapheresis decreased her total cholesterol levels and eventual liver transplant for PSC resolved her LpX-mediated hyperlipidemia and markedly improved her cutaneous xanthomas.

DISCUSSION

LpX mediated hyperlipidemia is a nonfamilial cause of hyperlipidemia primarily seen in liver dysfunction, notably in cholestasis related conditions such as primary sclerosing cholangitis (PSC). In these patients, cholestasis prevents the conversion of cholesterol to bile acids in the liver and results in plasma accumulation of low-density lipoprotein X, which is composed primarily of cholesterol and phospholipids. On laboratory evaluation, cholesterol levels are markedly elevated, TG levels are normal, and LDL can be artificially elevated due to similar density to lipoprotein X. Xanthomas are the major dermatologic manifestation of hyperlipidemia as excess lipoproteins cross dermal vasculature and are deposited in the dermis. Such plasma lipoproteins accumulate in dermal histiocytes where they are visible as "foam cells" on biopsy.[1] Treatment for xanthomas associated with LpX is focused on addressing the underlying cause. Six cases have been reported, which have resolved with pharmacologic management of dyslipidemia (1), plasmapheresis (2) and liver transplant (3).[2]



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CASE 27: Erythema Induratum with TB

Authors: Catherine Lyons BS, Nicole Edmonds MD, R Hal Flowers MD

HISTORY

A 51 yo female with a history of chronic anemia and recent positive Quantiferon-TB gold test presented with a 3-month history of a painful rash on the bilateral lower extremities. She also reported a nonproductive cough, low grade fevers, and recent weight loss.

PHYSICAL EXAMINATION

Numerous erythematous to hyperpigmented indurated subcutaneous nodules were scattered over bilateral lower legs, predominantly on the posterior calves. Areas of ulceration overlay several of the nodules.

RADIOLOGY/ PATHOLOGY

CXR was negative, computed tomography chest revealed nonspecific diffuse subtle ground glass interstitial haziness.

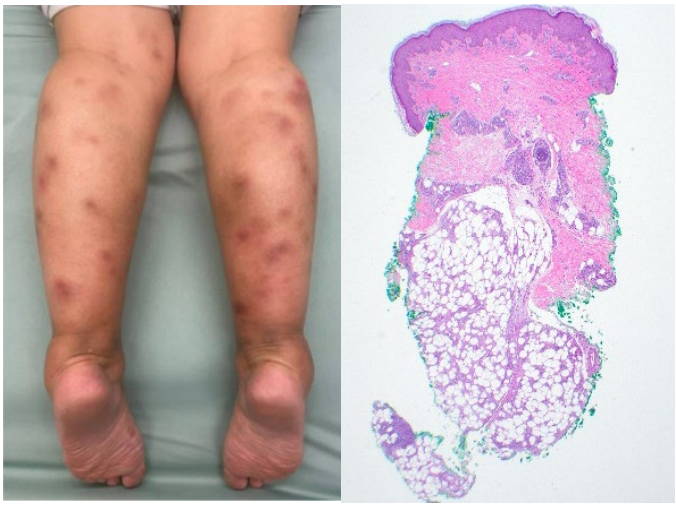
Biopsy of lesions showed sections with fat necrosis involving the subcutaneous fat lobules and rare granulomas noted at the periphery of a fat lobule. The septum was mildly thickened, by the biopsy showed predominantly lobular involvement, unlike the predominant septal involvement seen in erythema nodosum. There was a lymphocytic infiltrate with histocytes and eosinophils in the subcutis. No definitive vasculitis or caseating necrosis was identified making the diagnosis of polyarteritis nodosum and cutaneous tuberculosis, respectively, less likely. Infectious stains were negative.

TREATMENT

Despite AFB sputum smear being negative x 3 and negative CXR, the patient was treated as having active TB in the setting of cough, weight loss, low grade fevers, and nonspecific findings on chest CT. Infectious disease and the local health department guided treatment with isoniazid, rifampin, pyridoxine, ethambutol, and pyrazinamide for 2 months, followed by 4 months of only isoniazid and rifampin. Her EI resolved concurrently with TB.

DISCUSSION

EI is similar in clinical appearance to erythema nodosum (EN) but is histologically distinguished by the presence of predominantly lobular panniculitis (as opposed to septal predominance of EN) and positivity for TB. When TB is negative, the same presentation and histological findings have historically been referred to as nodular vasculitis, though more recently these terms have been used synonymously to describe a clinicopathologic entity with several possible causes, one of which is tuberculosis. This case is atypical as most EI features vasculitis on pathology. EI is often associated with active or latent TB and is considered a hypersensitivity reaction to mycobacterial antigens (“tuberculid”). The mainstay of treatment for TB associated EI is to treat the underlying tuberculosis. Clinicians should be aware of the association of EI with TB and should further investigate cutaneous lesions concerning for EI even in the absence of overt signs of active TB to ensure proper patient care.



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CASE 28: Neuroblastoma

Authors: Madelyn Reddan, Olivia G. Cohen MD MPH, Barrett Zlotoff MD

HISTORY

A 4-month-old male with history of polyhydramnios presented with multiple firm blue nodules. Lesions initially appeared 3 months prior on the abdomen and were assumed to be lipomas, but progressed to involve other areas, including the scalp and groin. No fatigue, weight loss, or fevers were reported. Laboratory values were significant for elevated lactate dehydrogenase and urine catecholamines (homovanillic acid, and vanillylmandelic acid).

PHYSICAL EXAMINATION

Irregular rubbery subcutaneous blue-hued nodules scattered on left periumbilical, inguinal, scrotal, chest, and vertex scalp in linear clusters demonstrating possible in transit distribution.

RADIOLOGY/PATHOLOGY

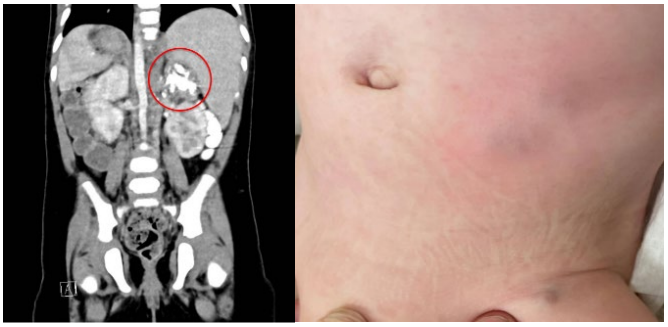
Ultrasound of the abdomen revealed a large heterogeneous mass with scattered internal calcifications areas in the right suprarenal space, favored to be neuroblastoma with multiple hepatic and subcutaneous soft tissue metastases. Biopsy of the abdominal wall mass showed poorly differentiated neuroblastoma. Bone marrow biopsy of the iliac crests revealed limited involvement by neuroblastoma and background of normocellular marrow and rare PHOX2B-positive cells.

TREATMENT

Clinical presentation, abdominal ultrasound, and elevated urine catecholamines supported a diagnosis of neuroblastoma with widespread metastasis on imaging; this was confirmed on biopsy of the abdominal lesion and bone marrow. Oncology initiated treatment for the patient with carboplastin/etoposide per protocol ANBL0531, which is ongoing.

DISCUSSION

Neuroblastoma originates from neural crest cells in the adrenal lineage, and is a common pediatric solid tumor. The PHOX2B gene encodes a transcription factor essential to development of the autonomous nervous system and proven to play a role in driving neuroblastoma, and children with high PHOX2B mutation burden are known to have high-risk disease. There is ongoing research into drug discovery and drug choice based on PHOXB2 expression and mutation burden for optimization of chemotherapeutic treatment in neuroblastoma patients. Our patient with a low PHOX2B positivity, was considered to have favorable genetic markers, however histology demonstrated poor differentiation prompting early chemotherapeutic treatment initiation. Although mistakable for benign soft tissue tumors, clinicians should be aware of this “blueberry muffin rash” presentation and consider underlying neoplastic disorders particularly if tumors grow, increase in number, or are associated with systemic symptoms. Imaging and additional testing should be done to confirm the diagnosis promptly.



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CASE 29: Multiple asymptomatic papules arising within a birthmark

Authors: Fatima Choudhary BS, Olivia G. Cohen MD MPH, Linglei Ma MD PhD, Barrett Zlotoff MD

HISTORY

A 7-year-old girl presented with asymptomatic papules arising within a birthmark on the trunk. The papules arose spontaneously and were increasing in number over several months. She endorsed occasional bleeding. She was otherwise healthy and denied skin trauma.

PHYSICAL EXAMINATION

Hyperpigmented brown segmental smooth bordered patch extending from the right flank to the left mid-back. Multiple scattered 1-2-mm red-purple vascular papules contained within the borders of the patch. Scar on the left mid-back at the site of previous biopsy.

PATHOLOGY/LABORATORY DATA

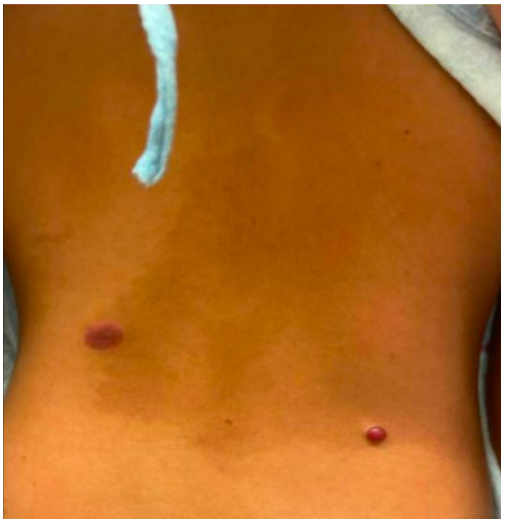
Shave biopsy of a papule demonstrated lobular proliferation of capillaries with focal areas of vascular ectasia, consistent with pyogenic granuloma. The background skin showed basilar hyperpigmentation. Dermoscopy was consistent with a cafe-au-lait spot. Tissue genetics of the vascular malformation was positive for NRAS Q61R missense mutation with Variant Allele Frequency (VAF) of 14.5%. Germline genetic analysis was negative.

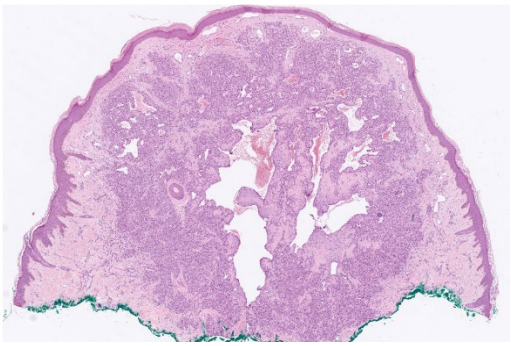
TREATMENT

Patient was diagnosed with pyogenic granulomas (PG) arising in a cafe-au-lait macule (CALM) due to a somatic mosaic NRAS mutation. She was started on oral propranolol without improvement and topical sirolimus, which was discontinued due to irritant dermatitis. She underwent shave removal of the larger pyogenic granulomas and pulsed dye laser of smaller lesions and is being monitored for recurrence.

DISCUSSION

Despite NRAS mutations being associated with CALM as well as PG in certain rasopathies, the occurrence of PG within a CALM has not been previously reported, either with or without underlying somatic NRAS mutation.[1,2] Treatments recommended for PG include surgical excision, electrocautery, cryotherapy, sclerotherapy, pulse-dyed laser, intra-lesional steroids and systemic treatments such as beta-blockers. Sirolimus, an mTOR inhibitor downstream in the RAS-MAPK pathway, has previously been shown to successfully reduce vascular tumors such as PG.[3] Our patient did not tolerate topical sirolimus and thus efficacy was indeterminate in this case. Shave removal and pulsed dye laser were effective second-line treatment options for our patient.





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CASE 30: Widespread Trichodysplasia Spinulosa Following Renal Transplant

Authors: Lucy Rose MA, Olivia G. Cohen MD MPH, Maggie Noland MD

HISTORY

A 58 y.o. female presented with a rash concentrated on her face that began spreading to her trunk and extremities. The rash initially presented on the central face and eyelids, and then spread diffusely over several months. She has a prior history of renal transplant with immunosuppressive therapy consisting of mycophenolate mofetil, tacrolimus, and prednisone. She has seen two outside dermatologists prior to referral.

PHYSICAL EXAMINATION

Numerous flesh colored to white spiny follicular papules on the face, neck, and trunk. Spines also extended into nasal vestibules and over the lips. Similar hyperpigmented papules and follicular spines were present on the upper trunk and arms.

RADIOLOGY/LABORATORY DATA/PATHOLOGY

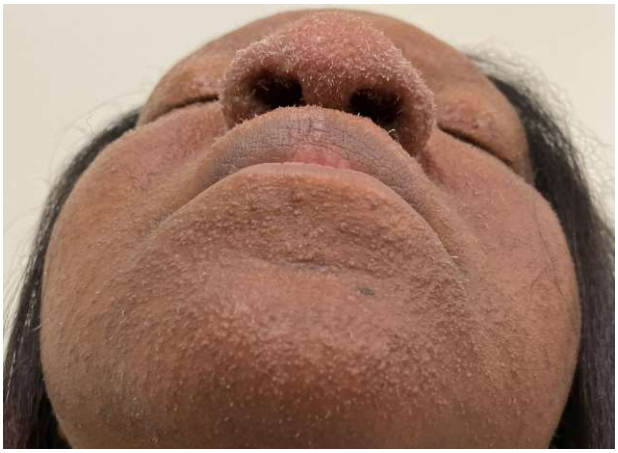
Biopsy of her left cheek demonstrated mildly dilated follicular ostia with a dystrophic inner root sheath. Many larger eosinophilic trichohyalin granules are noted in the inner root sheath. PAS staining was negative for fungal elements.

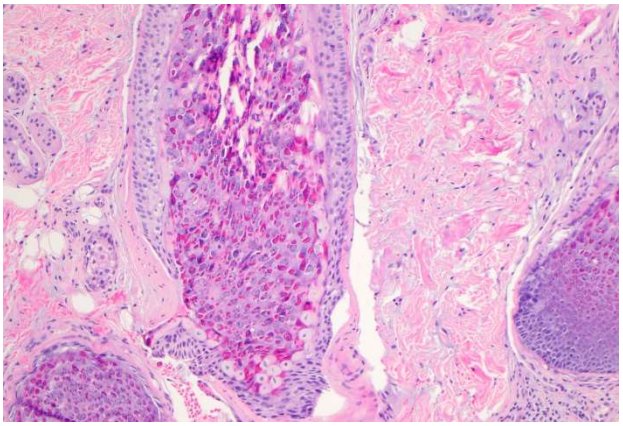
TREATMENT

A diagnosis of Trichodysplasia Spinulosa was made based on histopathology and clinical findings. Treatment was initiated with topical cidofovir 3% cream. However, plan was to increase to oral treatment if no significant improvement given the bothersome intranasal and oral follicular spines that interfered with comfortable respiration.

DISCUSSION

Trichodysplasia Spinulosa (TS) is caused by the Trichodysplasia Spinulosa-associated Polyomavirus (TSPyV), and is largely seen in patients who are immunosuppressed. The incidence is unknown. Clinically, TS is most frequently reported on the face, but cases extending to the trunk and extremities have been reported. Reducing immunosuppression usually leads to reduction in viral load and improvement in cutaneous findings; however, this is not always feasible. Topical cidofovir is an antiviral medication, however it can difficult to access and has not been used on larger surface areas.





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CASE 31: Nail-patella syndrome

Authors: Madelyn Reddan, Olivia G. Cohen MD MPH, Barrett Zlotoff MD

HISTORY

A 5-month-old female with a history of infantile hemangioma, atrial septal defect, cord drug screen positive for methamphetamines and amphetamines, preterm birth, nephrotic syndrome, presented to clinic regarding a recent diagnosis of nail-patella syndrome. Patient's mother shared that they have a strong family history of nail-patella syndrome, including the patient's mother, maternal grandmother, and older sister.

PHYSICAL EXAMINATION

Upon examination, the patient, her mother, and grandmother have triangular dysplastic thumbnails bilaterally. The patient has an abducted right leg. Patella were not palpable on exam.

RADIOLOGY

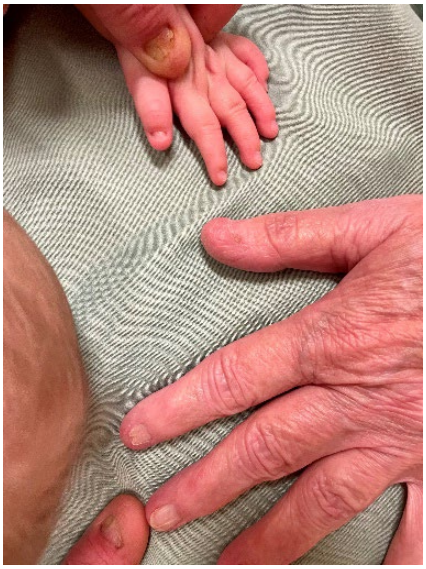
Annual renal ultrasound was reported as within normal limits. No radiographs of the long bones, nor patella were obtained.

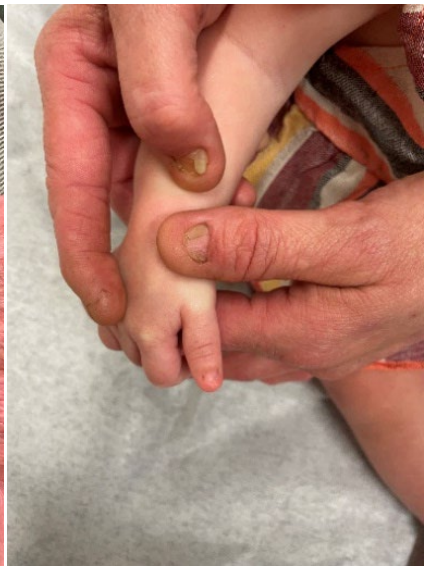
TREATMENT

Clinical presentation was consistent with previous diagnosis of nail patella syndrome (NPS). Patient had established care with pediatric nephrology, but had not established care with any other specialty providers. Patient was referred to ophthalmology for annual screening for glaucoma and ocular hypertension, to pediatric orthopedics for the patient's abducted right leg, and to pediatric genetics for further evaluation.

DISCUSSION

NPS is a rare autosomal dominant disorder due to mutation of the *LMXB1* gene on chromosome 9. This encodes a transcription factor involved in embryological limb dorsal-ventral patterning, renal and ocular development. Clinical features are variably expressed and include nail dysplasia, patellar aplasia or hypoplasia, iliac horns, radial dysplasia, glaucoma and nephropathy. Nail changes are most pronounced on the ulnar side of the thumbnail, and decrease as you reach the 5th fingernail. They include hypoplasia to absence of the nail plate, spooning, and fragility. Triangular or V-shaped lunula are pathognomonic for NPS. Radiographs of absent patella and radial heads dislocations may aid in diagnosis; however, patella do not ossify until age 2-6 years and therefore this type of imaging is not useful prior (as in our patient). Access to a multidisciplinary team of specialty providers can allow for detection of renal, ophthalmic, and neurological involvement. Early referral to genetics can lead to adequate screening and counseling for patients regarding comorbidities and mortality related to this diagnosis.





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CASE 32: Netherton Syndrome

Authors: Divya Shan BA, Olivia Cohen MD MPH, Kenneth Greer MD

HISTORY

A 55-year-old male was diagnosed at UVA Dermatology Clinic with congenital Netherton syndrome at age 8, in addition he has a history of gout and hypothyroidism. Since birth, he has experienced extensive skin erythema, scaling, and pruritus exacerbated by hot weather. He is prone to blistering. The patient has food allergies to nuts. His sister also has Netherton syndrome which has been refractory to Dupixent treatment.

PHYSICAL EXAMINATION

Widespread serpiginous erythematous plaques bordered by double-edged scales at the margins on the face, neck, abdomen, trunk, and upper extremities. Well-demarcated shiny erythematous plaques in the axilla and groin areas. Short fragile hair on the scalp and eyebrows with nodular hair-shaft deformities.

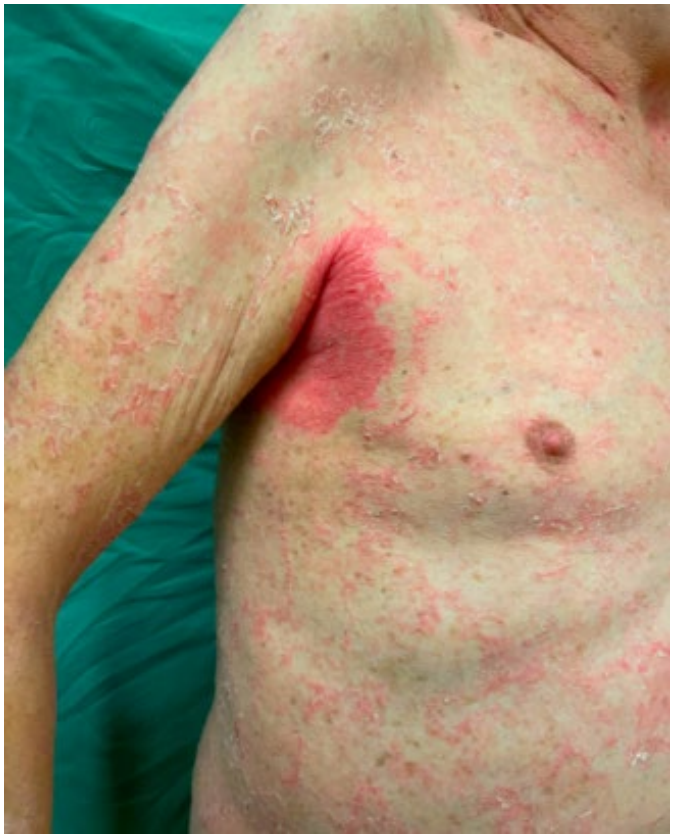
TREATMENT

He was initially treated with acitretin 10 mg daily, and experienced cutaneous flares of tender erythematous scaling areas with overlying pustules and headaches. Acitretin was stopped and the patient started a short course of Keflex 250 mg QID and steroid injections for subsequent severe flares. Other therapies included topical triamcinolone, oral hydrocortisone, and prednisone without success.

In 2020, the patient was invited back to clinic and started monthly Cosentyx (secukinumab) 300 mg injections based on new research findings. Currently managed with Cosentyx, he has experienced 50-75% improvement in ichthyosis symptoms, with resolution of pustules and cellulitis of the legs secondary to dry skin. He uses Eucerin as an emollient and no other topicals.

DISCUSSION

Netherton syndrome is a rare severe autosomal recessive disorder caused by mutations in serine protease inhibitor Kazal-type 5 (SPINK5). It classically presents with scaling erythroderma, hair shaft deformities (such as trichorrhexis invaginata or “bamboo hairs”), and atopic diathesis. The classical cutaneous presentation is ichthyosis linearis circumflexa, characterized by lesions that spontaneously change shape and size. Anti-IL-17 therapy, like secukinumab, has shown promise in managing Netherton syndrome, as observed in this case and a few other reported cases.



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Case 33: MEK inhibitor-induced Acneiform Eruption

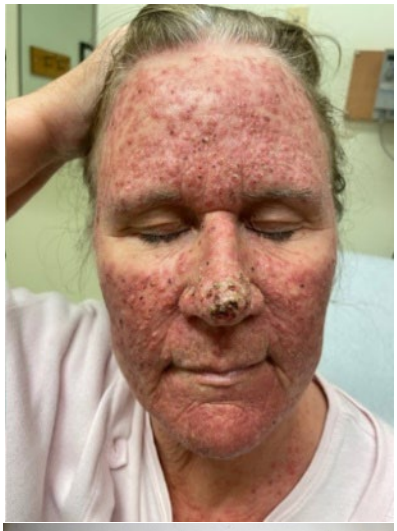
Authors: Nakul Dar BS, Peter Jowdy MD, Barrett Zlotoff MD

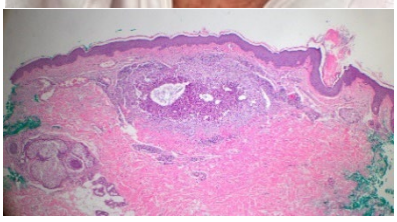
HISTORY

A 61-year-old woman presented with a painful, pruritic rash on her face and chest one week after starting trametinib for metastatic ovarian adenocarcinoma. She initially developed facial flushing which rapidly progressed to this eruptive morphology. Her metastatic ovarian adenocarcinoma was primarily treated two years ago. She had originally undergone treatment with neoadjuvant carboplatin/paclitaxel and debulking with total abdominal hysterectomy, bilateral salpingo-oophorectomy, appendectomy, liver resection, and splenectomy. For two years she had been on letrozole maintenance therapy but experienced disease progression prompting the use of trametinib.

PHYSICAL EXAMINATION

Robust, near confluent erythematous papules and pustules, some with overlying crust, most numerous on the face but also present on the chest, neck, and back.





PATHOLOGY

Punch biopsy of a lesion on the neck showed aggregates of neutrophils with nuclear debris in the upper dermis adjacent to a hair follicle.

TREATMENT

The patient was advised to stop trametinib and was prescribed triamcinolone cream without improvement. She was started on a methylprednisolone taper and oral doxycycline which resulted in improvement after one week. She is currently on a regimen of daily clindamycin lotion with hydrocortisone ointment as needed for flares, The patient has not resumed trametinib since the rash and is instead receiving anastrozole, a hormone-based chemotherapy.

DISCUSSION

The patient was diagnosed with an acneiform eruption and secondary bacterial infection due to trametinib, an oral MEK inhibitor. This is a common side effect of MEK inhibition estimated to occur in 74-89% of patients.[1] Dermatologic toxicity often begins within two weeks of oral MEK inhibitor use. Other cutaneous adverse effects have included palmar-plantar erythrodysesthesia syndrome, chronic paronychia, and eczematous dermatitis.[2] Oral MEK inhibitors are gaining attention for their efficacy in treating plexiform neurofibromas and low-grade gliomas in patients with Neurofibromatosis Type I (NF1). Topical formulations for the treatment of neurofibromas are currently being developed which may further increase the use of MEK inhibitor therapy. Awareness and medication counseling along with possible anticipatory management may reduce the severity of MEK inhibitor related reactions.[3] While there are no preventative guidelines in place, a concurrent course of doxycycline or prednisone when initiating MEK inhibitor therapy may limit adverse events. Dose reduction is recommended and tailored towards the severity and number of outbreaks.

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Case 34: Sjögren-Larsson Syndrome

Authors: Nakul Dar BS, Peter Jowdy MD, Barrett Zlotoff MD

HISTORY

A 12-year-old boy presented with congenital plate-like scaling, intellectual disability, and spastic diplegia of unknown etiology. A chromosomal microarray revealed increased regions of homozygosity suggestive of consanguinity, but no clear recessive genetic condition was identified. He had never been able to walk independently and had a history of developmental regression since age seven. Differential diagnosis at presentation included lamellar ichthyosis, X-linked ichthyosis, and Sjogren-Larsson syndrome.

PHYSICAL EXAMINATION

Patient is wheelchair bound. Skin examination revealed thick, brown, scaly, plate-like plaques on the face, extremities, and trunk.

LABORATORY DATA

Homozygous ALDH3A2 related fatty aldehyde deficiency identified.

TREATMENT

He had initially been treated with acitretin 20 mg daily with moderate response and triamcinolone 0.1% ointment as needed for pruritus. He had previously failed treatment with topical therapy alone including urea 20% cream and elta cream-menthol 0.25%. The patient was seen by ophthalmology and found to have no evidence of crystalline retinopathy or macular dystrophy. His acitretin dose was increased to 40 mg daily, and his topical regimen now includes triamcinolone 0.025% lotion, urea 40% cream, and ceramide-containing moisturizers.

DISCUSSION

Sjogren-Larsson syndrome (SLS) is a rare autosomal recessive disorder caused by mutations in the ALDH3A2 gene which plays a crucial role in fatty acid metabolism. Individuals with SLS typically present with a triad of symptoms: intellectual disability, spasticity, and ichthyosis. Patients with Sjogren-Larsson syndrome may also have retinopathy and seizures. The suspected mechanism of disease is accumulation of toxic long-chain fatty aldehydes and fatty alcohols. Increased fatty aldehydes impair the epidermal lamellar bodies that are responsible for maintaining the lipid barrier.[1,2] The epidermis' loss of water retentive properties clinically results in ichthyosis. There is no definitive therapy for SLS, but cutaneous symptoms can be improved by maintaining the epidermal water barrier either through adding moisture or removing excess scale. Urea cream has been shown to improve cutaneous symptoms in a cohort study.[3] A topical cholesterol/lovastatin mixture has been reported to improve symptoms by replacing the skin's abnormal lipid composition [1]. Muscle relaxants may also be considered for spasticity including benzodiazepines, anticholinergics, and oral baclofen. Developing treatments are aimed towards fatty aldehyde scavengers, PPAR agonists, and gene therapy.

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Case 35: Ovoidal Palatal Patch in TIF1γ-positive Dermatomyositis

Authors: Aaron D. Smith BS, Peter Jowdy MD, R Hal Flowers MD

HISTORY

An 82-year-old woman with a history of hypertension and intraductal papillary mucinous neoplasm presented with a 20-year history of photosensitive eruptions with pathology showing vacuolar interface dermatitis. She had a history of myositis and now has persistent mild weakness.

PHYSICAL EXAMINATION

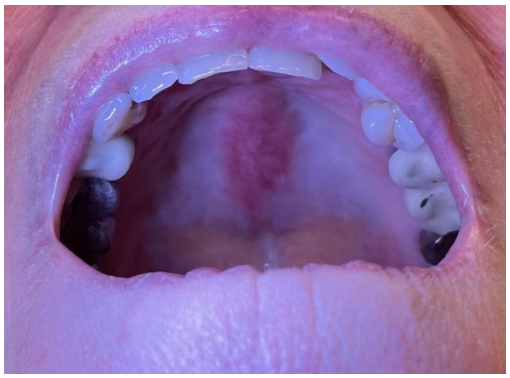
The physical exam revealed erythematous plaques with fine scaling over the forehead, bilateral infraorbital regions, and cheeks, as well as extensive scaling on the posterior scalp. Poikilodermatous changes were noted on the upper back, chest, and extensor surfaces of the bilateral arms, with the right side worse than the left. A heliotrope rash was present around the right orbit. Additionally, there was cuticular hemorrhage with dilated capillary loops of the nailfolds, but no Gottron's papules. An erythematous, well-circumscribed, ovoid patch was observed on the hard palate.

LABORATORY DATA/PATHOLOGY

Autoimmune serologies showed high titer ANA (1:640) with a speckled pattern. Extractable nuclear antigens were negative, but myositis antibody panel showed positive TIF1γ antibody. Muscle biopsies (at age 69) showed perivascular and perifascicular lymphocytic infiltrates and perifascicular muscle atrophy, consistent with dermatomyositis. Recent muscle markers unremarkable.

TREATMENT

Malignancy screens have remained negative since her diagnosis. After experiencing side effects from several conventional agents (methotrexate, mycophenolate mofetil, apremilast) and deferring azathioprine due to similar concerns, she has been receiving IV and SQ immunoglobulin for about 9 years. Improvement is sub-optimal given inability to tolerate higher doses; currently, the patient receives 1g/kg of monthly IVIG. She defers other treatment options.



DISCUSSION

TIF1γ dermatomyositis carries the highest risk of internal malignancy amongst dermatomyositis subtypes. This type of dermatomyositis tends to be amyopathic and present with extensive cutaneous involvement ranging from classic dermatomyositis findings to distinct eruptions such as keratotic palmar papules, psoriasiform plaques, or hypopigmented lesions with telangiectatic patches. The ovoid palatal patch is present in about 40% of patients and is nearly pathognomonic for TIF1γ dermatomyositis. This appears as a symmetric and well-circumscribed violaceous patch across the midline of the hard palate.[1] In the absence of genetic confirmation, this finding may predict a higher rate and severity of internal malignancy, as well as an increased concurrence of amyopathic dermatomyositis.[2,3] Malignancy risk is especially increased within the first five years of diagnosis. Current treatment primarily includes photoprotection and systemic therapy with glucocorticoids, methotrexate, mycophenolate, and IVIG.[4]

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Case 36: Generalized Lichen Sclerosis

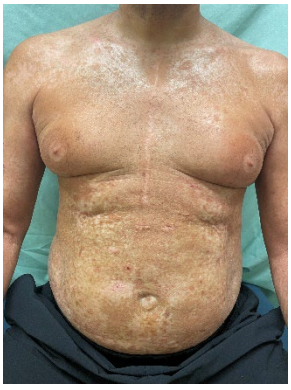
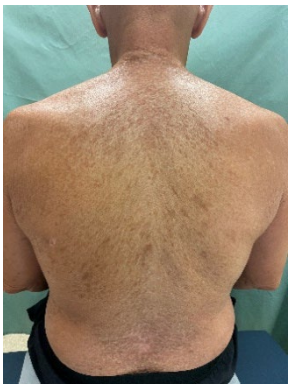
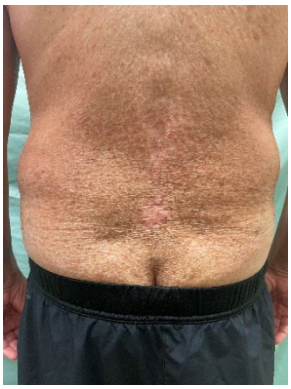
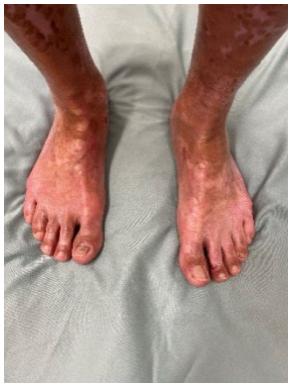
Authors: Aaron D. Smith BS; Peter Jowdy MD; Kenneth Greer MD

HISTORY

A 45-year-old male with a history of diabetes mellitus and hypertension presented with a several-year history of a florid skin eruption that had spread profusely over the trunk, arms, legs, groin, and buttocks. The lesions were described as extremely pruritic, and symptoms worsened upon exposure to water and heat. He did not report any associated dysphagia or Raynaud's phenomenon.

PHYSICAL EXAMINATION

Sclerotic flat-topped patches and plaques with linear lesions, most prominent on the trunk and legs. The lesions were bilateral and somewhat symmetrical. There is mild genital involvement. The face was spared.

TREATMENT AND CLINICAL COURSE

A diagnosis of lichen sclerosis et atrophicus (LS) was made based on history and physical exam findings. Narrow band UVB therapy was prescribed due to the generalized involvement. The patient was lost to follow-up but presented to our dermatology clinic 12 years after his initial presentation for a routine skin check in the setting of immunosuppressive treatments for an imminent kidney transplant due to end-stage renal disease from diabetes mellitus. The patient disclosed that he never received phototherapy. The physical exam showed minimal changes from the initial visit. Topical triamcinolone was provided for symptomatic relief of pruritus.

DISCUSSION

Lichen Sclerosis et Atrophicus is a chronic inflammatory skin disease with an unknown pathogenesis, though it may be linked to autoimmune, genetic, traumatic, and environmental factors. While LS affects both sexes, it has a bimodal distribution, with the highest prevalence in postmenopausal women and prepubescent girls.[1,2] Approximately 15% of LS cases are extragenital, presenting as asymptomatic white opalescent papules that cluster into plaques and slowly progress over time. This progression results in parchment-like skin, typically involving the upper trunk, neck, and shoulders. Lesions are often accompanied by purpura. Histologically, LS is characterized by an atrophic epidermis with the loss of rete ridges, lymphocytes in the basal layer, a subepidermal band of sclerosis, and a deep lichenoid infiltration of lymphocytes.[3] First-line treatment for extragenital LS includes super-potent topical steroids and topical calcineurin inhibitors. Treatments for refractory and severe cases include oral corticosteroids, methotrexate, phototherapy, topical calcipotriol, and acitretin. Novel therapies include JAK-inhibitor baricitinib and IL-6 inhibitor, tocilizumab.

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Case 37: Bannayan-Riley-Ruvalcaba Syndrome

Authors: Aaron D. Smith BS, Catherine E. Lyons BS BA, Peter Jowdy MD, Barrett Zlotoff MD

HISTORY

A 19-month-old boy with macrocephaly presented with an asymptomatic soft mass in the left axilla and bluish discoloration of the upper back that had been present for one year. His family history is significant for papillary thyroid cancer in his mother when she was 6 years old, and large lipomas in his sister and maternal grandmother.

PHYSICAL EXAMINATION

Three rubbery masses in the paraumbilical region and both left and right axilla. Each were of variable sizes ranging from the size of a ping-pong ball to the size of a softball. Hypertrophy of the left index finger. No penile lentigines.

RADIOLOGY/PATHOLOGY

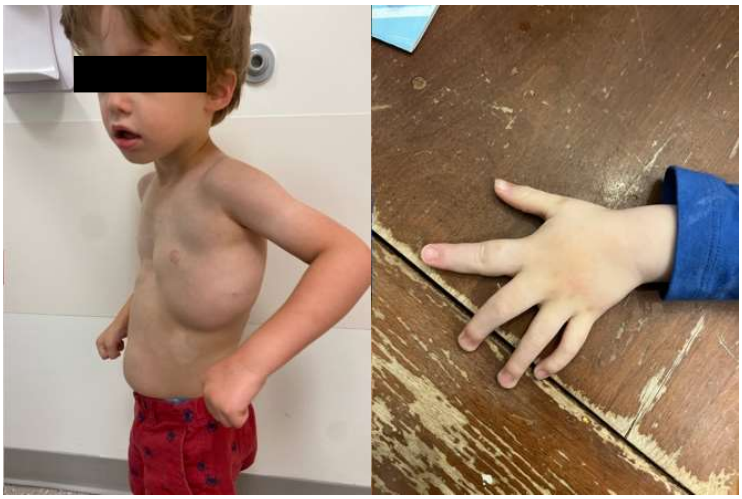
MRI of the abdomen and chest demonstrated large non-enhancing subcutaneous masses with loss of signal on fat saturation. Histopathology was unremarkable. Immunostains for D2-40, CD34, and ERG highlighted vessels in a normal number and distribution, indicating no vascular proliferation.

TREATMENT

Given the combination of familial lipomas, macrocephaly, and early-onset thyroid cancer, an overgrowth syndrome was suspected. Whole exome sequencing revealed a PTEN variant (c.1042A>C, p.Thr348Pro). The presentation aligns with the clinical findings of PTEN Hamartoma syndrome (Bannayan-Riley-Ruvalcaba type). The left axillary lesion was surgically debulked and medical management focused on shrinking lesions to reduce the need for further intervention and prevent recurrence. Sirolimus was started but discontinued due to pneumonia. Further genetic testing at Cleveland Clinic is being done to confirm if his mutation in PIK3CA, currently classified as VUS, is truly pathogenic. This may help guide therapy.

DISCUSSION

PTEN Hamartoma Tumor Syndrome (PHTS) encompasses a sequalae of hamartomas,



developmental anomalies, increased risk of malignancy, and germline PTEN pathogenic variants. [1,2] Bannayan-Riley-Ruvalcaba syndrome (BRRS), a variant of PHTS, includes phenotypic features such as pigmented penile macules, café-au-lait spots, vascular malformations, lipomas, hamartomatous polyps, and Hashimoto thyroiditis. Treatment focuses on managing symptoms with PTEN pathway inhibitors and cancer surveillance, including regular checks for breast, thyroid, uterine, kidney, and skin cancers. Timely diagnosis and understanding BRRS's natural history are crucial for gene-informed management, high-risk cancer surveillance, and addressing neurodevelopmental symptoms. Currently, surgical resection is the most common treatment option but there if often recurrence requiring re-excisions. Sirolimus, and more recently apelisib, have both shown effectiveness in reducing lipomatosis. Patients with PI3K pathway mutations may benefit from the use of apelisib over sirolimus. [3] As seen in this case, dermatologists play a key role in early identification and treatment.

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Case 38: Birt-Hogg-Dubé Syndrome

Authors: Katherine Byrnes BS, Peter Jowdy MD, Mary Noland MD

HISTORY

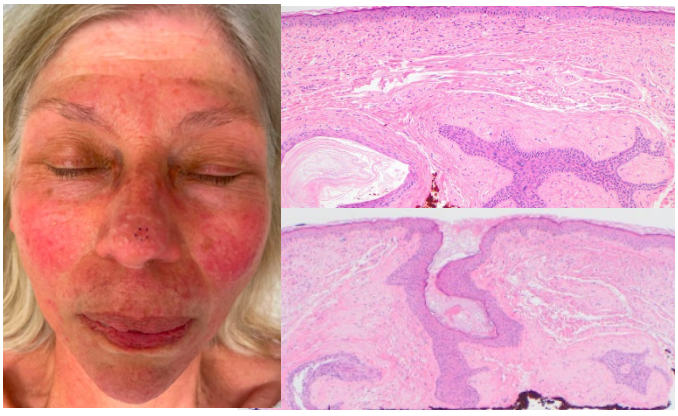
The patient is a 73-year-old female with a past medical history of nonmelanoma skin cancer, breast cancer, colon polyps, osteopenia, and benign paroxysmal positional vertigo, who originally presented to the clinic at age 69 with numerous “fibrous papules” on her face. She reported continued development of new papules and noted that her sister had similar lesions. She denied any personal or family history of known lung or kidney problems.

PHYSICAL EXAMINATION

Hundreds of 1-2mm white, small, fibrous papules on the face and nose. Multiple, small, skin-colored papules were also observed on the left lateral breast/flank area.

PATHOLOGY

Biopsies of papules on the right nasal sidewall and left cheek were performed (age 70). The nasal sidewall papule anastomosing epithelium with adjacent fibrotic stroma. These features were consistent with trichodiscoma (top image). The left cheek papule demonstrated a benign biphasic lesion with follicular and mesenchymal differentiation characterized by the proliferation of thin, anastomosing epithelial cores attached to a dilated follicular structure, with CD34-positive stroma, indicating fibrofolliculoma (bottom).



TREATMENT

After biopsy, the patient underwent genetic testing and was found to have a pathogenic variant in the FLCN gene, confirming the diagnosis of BHD. She was counseled about her genetic syndrome and need for annual MRI to screen for renal cancer. Several of her family members were also found to have BHD, but none have developed renal disease. Her fibrofolliculomas are monitored without cosmetic intervention and she remains without history of renal cell carcinoma or spontaneous pneumothorax. Her comprehensive dermatologic care has also included treatment of actinic keratoses, squamous cell carcinoma in situ, and basal cell carcinomas with cryotherapy and topical 5-Fluorouracil.

DISCUSSION

BHD syndrome is a rare autosomal dominant genodermatosis caused by mutations in the FLCN gene, which encodes folliculin. FLCN interacts with multiple molecular pathways including mTOR signaling. Kidney tumorigenesis is suspected to be related to a somatic “second hit” FLCN mutation. Clinically, BHD is characterized by the triad of fibrofolliculomas, trichodiscomas, and acrochordons. Fibrofolliculomas are the most common finding and are present in greater than 85% of individuals with BHD. The dermatologic manifestations typically appear in the third to fourth decade of life. Although dermatologically benign, patients with BHD syndrome are at increased risk for renal cell carcinoma, spontaneous pneumothorax, and pulmonary cysts. Compared to unaffected siblings, BHD patients have a 7-fold greater risk of kidney neoplasia and 50-fold greater risk of spontaneous pneumothorax. Early recognition of BHD can optimize management, including routine skin examinations, renal imaging, and genetic counseling to mitigate associated risks.

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CASE 39: BASCULE syndrome in an 11-year-old girl

Authors: Dema Abul-Enin BS, BA; Scott Berg MS, MD, Barrett Zlotoff MD

HISTORY

An 11-year-old female with a history of ADHD complained of purple discoloration followed by sore, aching pain with mild swelling in both legs that appeared several minutes after standing or sitting with legs in a dependent position. Episodes occurred 5 – 10 times daily with associated lightheadedness and nausea. Symptoms resolved within minutes of elevating her legs. The patient had begun puberty, as she had started her menses within 6 months of her initial appointment.

PHYSICAL EXAMINATION

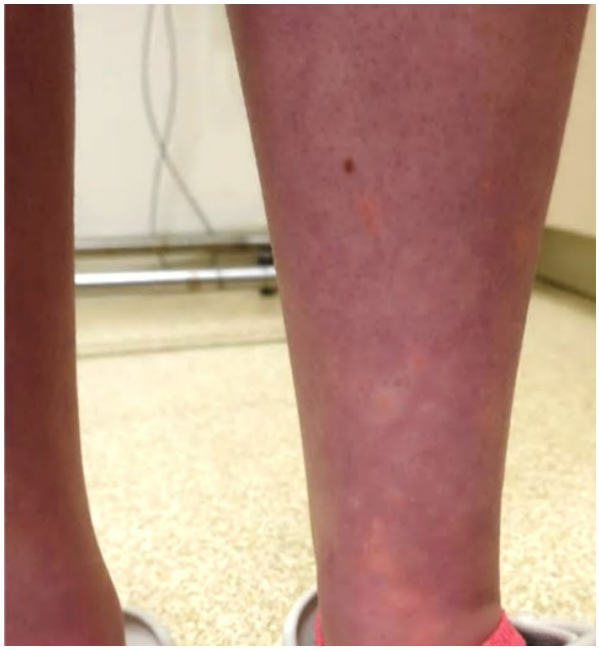
Development of cyanosis of bilateral legs, Bier spots, and pink spots within minutes of standing. Symptoms resolve minutes after elevating legs.

TREATMENT AND CLINICAL COURSE

The patient was diagnosed with Bier anemic spots, cyanosis with urticaria-like eruption (BASCULE) syndrome. Vital signs were negative for orthostatic hypotension, but symptoms were consistent with orthostatic intolerance. Initial recommended therapy was exercise, increased water and salt intake, and compression stockings. On follow-up the patient had increased water intake but did not tolerate compression stockings due to sweating. Clonidine prescribed for anxiety was noted to improve the color change in the legs. Her psychiatrist was concerned that certain medications could worsen her symptoms of BASCULE, but the patient was encouraged to prioritize treatment of her anxiety. The patient was lost to follow up since their last visit in winter 2023.

DISCUSSION

BASCULE syndrome is a benign vasomotor dermatosis characterized by bier anemic spots, cyanosis, and urticaria-like eruption and was first defined in a 2016 case series of four patients (1). Age of onset varies from infancy to middle adulthood, with median onset between ages 12-14 and can be associated with autonomic dysfunction including postural orthostatic tachycardia syndrome (2,3). The lesions can be



associated with tenderness, pruritus or pain (4). The bier anemic spots and cyanosis are attributed to venous pooling from orthostatism, while the cause of the urticaria-like eruption is less understood but suspected to be related to mast cell degranulation (5). There is no well-established treatment, although success has been reported with antihistamines and propranolol (2).

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CASE 40: Dermatomyositis panniculitis

Authors: Scott Berg MD, R Hal Flowers MD

HISTORY

A 79-year-old woman with a 3-year history of treatment refractory, skin-limited, anti–TIF1γ dermatomyositis (DM) and lung nodules suspicious for adenocarcinoma was seen for follow-up with new progressive painful skin nodules of the axillae and upper arms for the past two months.

PHYSICAL EXAMINATION

Subcutaneous nodules on bilateral axillae and upper inner arms. Florid erythema consistent with DM of the anterior chest, dorsal forearms, upper and lower back.

PATHOLOGY

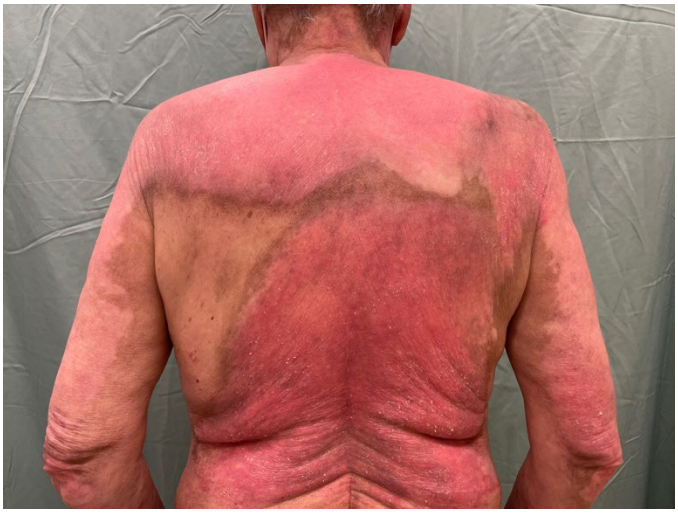
Biopsy of a nodule revealed prominent septal fibrosis with fat necrosis of intervening lobules and abundant mucin deposition in the fibrous septa of the subcutis and dermis. No interface dermatitis was identified.

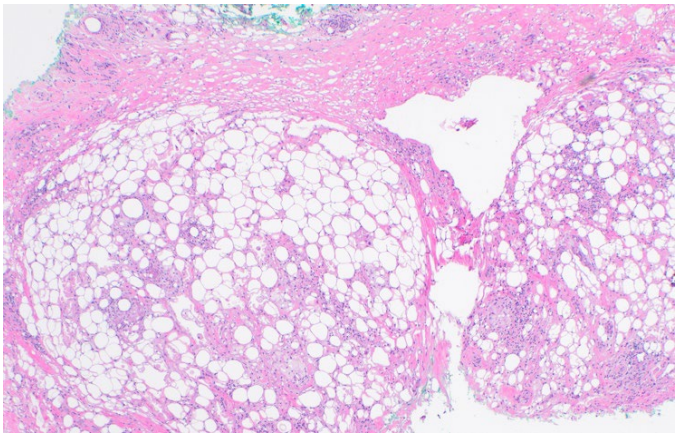
TREATMENT AND CLINICAL COURSE

The patient developed additional nodules of the axillae, chest, and buttocks which on repeat biopsy was most consistent with panniculitis associated with DM. She previously failed numerous therapies for her DM, including hydroxychloroquine, IVIG, rituximab, methotrexate, azathioprine, and apremilast. She improved with oral sirolimus but discontinued due to lower extremity edema. The patient continued to experience intermittent flares treated with prednisone, but long-term treatment options remain limited by her likely lung malignancy.

DISCUSSION

Panniculitis associated with dermatomyositis presents as persistent, indurated, painful nodules of the proximal extremities and abdomen. Histologically, the panniculitis of DM and lupus erythematosus are similar, with diffuse lobular infiltrates of lymphocytes and multifocal hyalinization of the subcuticular fat (1). The average patient age is 40 years old with a female predominance (2). Timing of panniculitis relative to other manifestations of dermatomyositis is variable, including delayed onset by years (2,3). Compared to the cutaneous lesions of DM, the panniculitis can respond more readily (4), simultaneously (5), or remain resistant to treatment (6). Sirolimus is an uncommon treatment for dermatomyositis (7) and was trialed as a safer option in the setting of suspected lung adenocarcinoma.





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CASE 41: Cutaneous Findings in a Germline POLE Mutation

Authors: Nicole Russell BS, Scott Berg MD, Barrett Zlotoff MD

HISTORY

A 10-year-old boy with history of high-grade glioma status post radiation and systematic therapy, CVA, carotid artery dissection, pilomatrixomas and urticaria pigmentosa was referred for dark spots on the neck and trunk.

PHYSICAL EXAMINATION

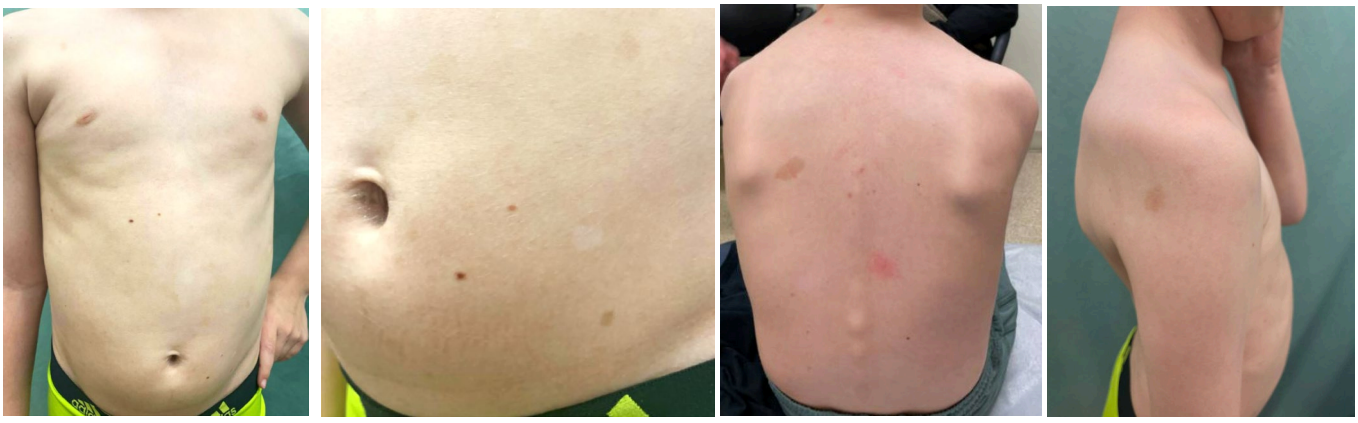
Café-au-lait macules (CALM), hypopigmented patches, lesions consistent with pilomatrixomas, and Darier sign-positive orange-brown macules and papules on the trunk and neck.

DISCUSSION

The *POLE* gene encodes the central catalytic subunit of DNA polymerase epsilon, which synthesizes the leading strand in DNA replication and has important proofreading functions. Prior workup after this patient underwent treatment of a glioma led to a diagnosis of a germline mutation in the *POLE* gene. Pilomatrixomas and café-au-lait macules, seen in this patient, has been reported in another case of a germline *POLE* mutation.[1]

Heterozygous germline mutations in *POLE* and the related *POLD1* were first associated with colorectal adenomas and carcinomas and in 2013 the disease was named polymerase proofreading-associated polyposis (PPAP).[2] Several other malignancies are now associated with PAPP, including upper gastrointestinal, endometrial and ovarian, brain, and breast cancer. This patient's specific variant (S459F) has been associated with colorectal and endometrial carcinoma, and screening colonoscopies and upper endoscopies are recommended in patients with *POLE* mutations.[3]

The cutaneous features reported with *POLE* mutation bears striking similarity to another cancer predisposition syndrome, constitutional mismatch repair deficiency (CMMRD), in which CALMs, pilomatrixomas, and hypopigmented patches (as also seen in our patient) are common.[4] The overlap in cutaneous findings between PAPP and CMMRD is hypothesized to be related to a higher degree of certain somatic mutations made possible by impaired mismatch repair mechanisms. Our patient also has a history of urticaria pigmentosa, which is commonly driven by KIT mutations and has not been reported with *POLE* mutations.



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CASE 42: Scleredema on extracorporeal photopheresis

Authors: Leon Zheng BA, Scott Berg MD, R Hal Flowers MD

HISTORY

54-year-old woman with no significant medical history presented in 2016 for 8 months of progressive skin hardening and tightening that began with the neck and upper back and progressed centrifugally. The remainder of her review of systems was negative.

PHYSICAL EXAMINATION

Diffuse skin hardening of the neck, shoulders, upper back, and proximal extremities with milder involvement of peripheral face and distal extremities.

PATHOLOGY AND LABORATORY DATA

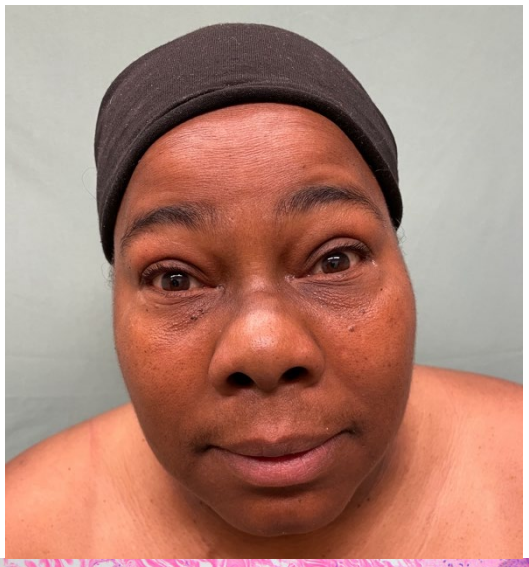
Biopsy from the left arm was consistent with scleredema, demonstrating separation of the collagen fibers in dermal connective tissue and increased dermal mucin. Serum protein electrophoresis demonstrated an IgG kappa monoclonal protein; workup for myeloma including bone marrow biopsy was negative.

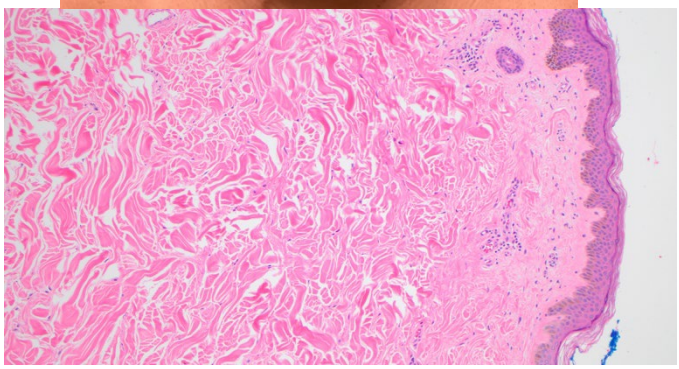
TREATMENT AND CLINICAL COURSE

The patient failed numerous treatments due to lack of response or adverse effects, including prednisone, cyclosporine, azathioprine, methotrexate, IVIG, PUVA, lenalidomide, and bortezomib. She started extracorporeal photopheresis (ECP) two days a week, every other week with hematology-oncology. After two months she had substantial improvement in skin tightening and range of motion, which was maintained at last follow-up eight months into therapy.

DISCUSSION

Scleredema is a rare skin disease characterized by induration primarily of the upper body with excessive collagen and mucin deposition in the dermis. It is associated with monoclonal gammopathies as seen in our patient, as well as infections and diabetes mellitus (1). No specific treatment has been established, but therapies include corticosteroids, hyaluronidase, IVIG, antibiotics, immunosuppressants, phototherapy, and electron beam therapy (2). ECP separates leukocytes from plasma and exposes them to a photosensitizing agent and ultraviolet A light before reinfusion. It first





was approved in 1988 for the treatment of Sézary syndrome and has roles in the treatment of GVHD and scleroderma (3). The first case report of ECP in scleredema was published in 2000 in a patient who had marked improvement with treatment (4). Our patient had considerable early improvement on ECP, although her rate of improvement plateaued, and we have recently added mycophenolic acid.

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CASE 43: Generalized HPV-associated SCCIS in the setting of AIDS

Authors: Nicole Russell BS, Scott Berg MD, R Hal Flowers MD

HISTORY

A 46-year-old man was admitted with a new diagnosis of HIV with a CD4+ count of 4/µL. Dermatology was consulted for generalized pruritic scaly papules, present for the past year.

PHYSICAL EXAMINATION

Numerous scaly, hyperkeratotic, hyperpigmented papules – many in linear groups – scattered on the anterior trunk and markedly on all extremities.

PATHOLOGY

Biopsy of four separate papules all demonstrated squamous cell carcinoma in situ (SCCIS) within a verruca. P16 immunostain demonstrated strong and diffuse block-like staining. In situ hybridization was performed on one lesion and was positive for high-risk HPV genotypes.

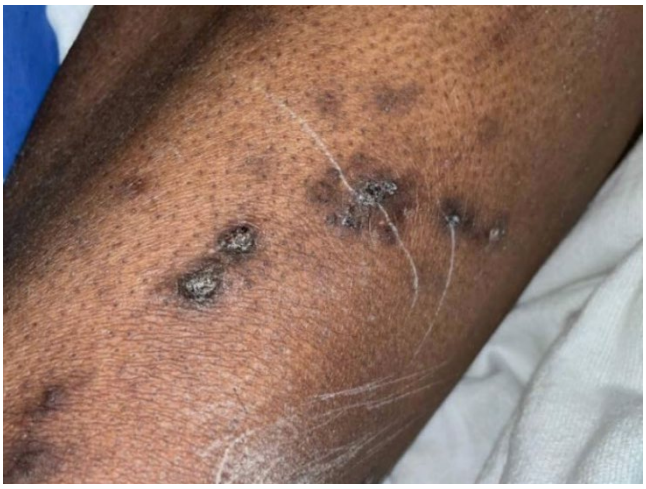
TREATMENT AND CLINICAL COURSE

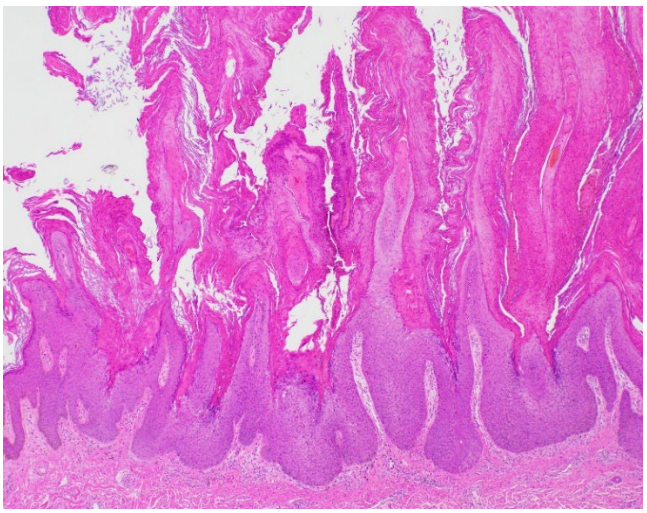
Two biopsy-proven SCCIS underwent curettage and 10 similar lesions were treated with cryotherapy. His CD4+ count improved on HIV therapy and his pruritus resolved. On his most recent follow-up the patient had complete resolution of all skin lesions with a CD4+ count of 171/µL.

DISCUSSION

This patient had a new diagnosis of HIV/AIDS with widespread lesions with biopsies demonstrating SCCIS arising within verruca. High-risk HPV types, especially HPV16/18, are most associated with cervical cancer, anogenital tract cancer, and head and neck cancers. Aside from digital SCC, cutaneous SCC is not linked with high-risk HPV types. There is evidence that beta HPV has a role in initiation of some cutaneous SCC, but high-risk HPV types, as found in this patient, are in the alpha genus.

Generalized SCCIS from high-risk HPV within verruca in the setting of HIV/AIDS does not appear to be previously reported. Patients with HIV have a higher burden of HPV-associated cancers, including anal, penile, and vulvovaginal disease and a higher incidence of common and plantar warts. Treatment of HIV is associated with decreased prevalence of high-risk HPV infection, and a subset of studies have shown reduced HPV-associated cancer risk. Vaccination for HPV in HIV-positive patients is safe and produces an immunologic response, although data on effectiveness is limited.





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CASE 44: Dyskeratosis Congenita

Authors: Taylor Fleshman MD, Lydia Luu MD, Shira Lanyi MD, Bridget Bryer MD

HISTORY

A 16-year-old female with a history of pancytopenia, latent tuberculosis, and a heart murmur with syncope presented for evaluation of skin pigmentation changes and hair loss. She previously noticed spots on the tongue, progressive hyperpigmentation of the hands and wrists, and ridging of the fingernails at ages 2, 7, and 8, respectively. She also noted hair loss and burning of the scalp. Her 18-month-old sister had similar oral lesions, but her other siblings were healthy. She reported three prior bone marrow transplants in her home country, and recent bone marrow biopsy revealed hypocellular marrow with decreased trilineage hematopoiesis.

PHYSICAL EXAMINATION

Reticular hyperpigmented patches and macules scattered over anterior neck, inguinal area, dorsal wrists, and hands. Longitudinal ridging of the nails bilaterally. Missing teeth and evidence of hyperpigmented macules and nodules on the tongue and hard palate. Nonscarring alopecia of the vertex scalp with perifollicular scale and erythema.

LABORATORY DATA AND TREATMENT

CBC revealed pancytopenia with WBC 3.76, Hgb 10.7, Plt 81. Telomere length testing revealed abnormally short telomeres. The patient was diagnosed with dyskeratosis congenita. Follow up was arranged with pediatric hematology/oncology for management of associated aplastic anemia. Annual skin checks were recommended for increased risk of cutaneous malignancy.

DISCUSSION

Dyskeratosis congenita is a genetic disorder of telomere maintenance resulting in bone marrow failure and the characteristic triad of reticulated hyperpigmentation, nail dystrophy, and oral leukoplakia. It carries an increased risk for cutaneous and hematolymphoid malignancies, and bone marrow failure is the most common cause of death. The inheritance may be autosomal dominant, autosomal recessive, X-linked, or, rarely, sporadic. Diagnosis requires at least two of the four previously mentioned major features and two or more multisystem features, including epiphora, developmental delay, pulmonary disease, periodontal disease, esophageal stricture, hair loss or graying, hyperhidrosis, and malignant lesions. Telomere length testing is below the first percentile, and genetic testing may confirm the diagnosis but has low sensitivity. No targeted therapies exist, and hematopoietic stem cell transplantation is the only curative treatment.

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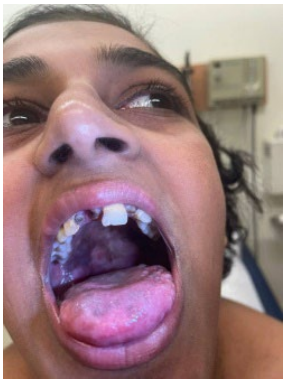
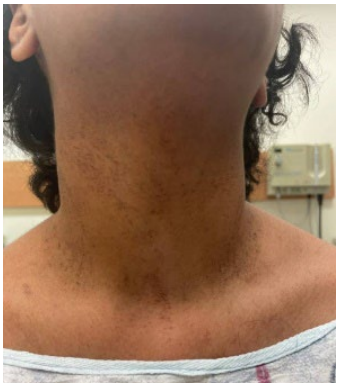
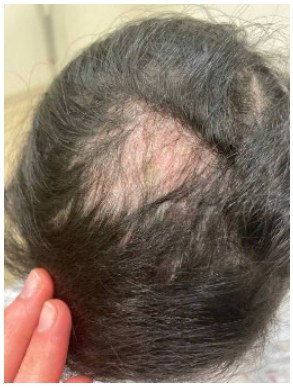
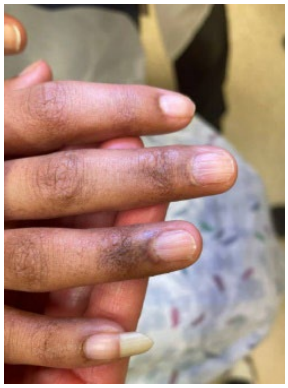





CASE 45: Osteoma Cutis & Albright's Hereditary Osteodystrophy

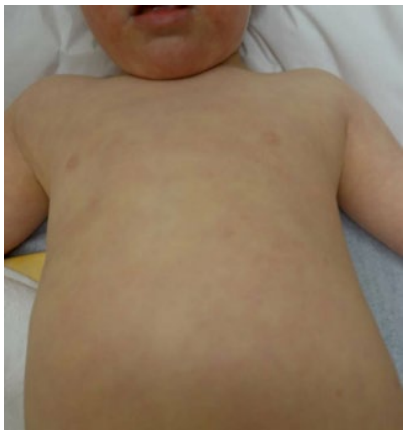
Authors: Margaret Mercante BA, Shira Lanyi MD, Barrett Zlotoff MD

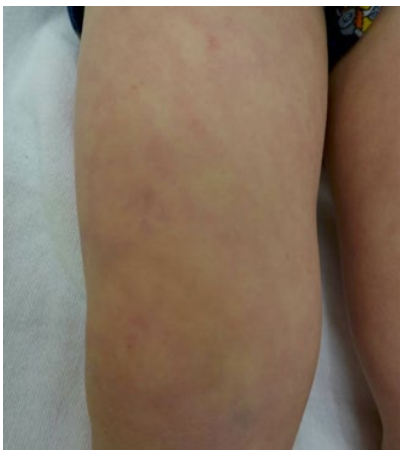
HISTORY

A three-year-old male with Albright hereditary osteodystrophy (AHO) with associated pseudohypoparathyroidism type 1A, hereditary alpha tryptasemia, and eczema presented with spots on the lower back, head, and legs. The patient's mother noticed skin abnormalities at six months, and he was initially diagnosed with spots of anetoderma on his abdomen. One year later, firm nodules on his scalp were diagnosed as osteoma cutis.

PHYSICAL EXAMINATION

There were multiple atrophic macules scattered over the left abdomen, chest, bilateral lower legs, and back. Numerous macules with have a bluish hue and few appear firm. There are linear dermal firm plaques with a calcified feel on palpation at the waistline.





PATHOLOGY

Initial biopsy of a representative lesion of the scalp was significant for osteoma cutis. Biopsy of a representative lesion of anetoderma demonstrated sparse mast cells and VVG demonstrated loss of elastic fibers in the deep dermis. No cutaneous mastocytosis was appreciated using CD2 and CD25 staining.

TREATMENT

The patient was diagnosed with macular atrophy of skin associated with GNAS mutation, and he was started on topical 25% sodium thiosulfate (STS) cream for areas of osteoma cutis ossifications. STS treatment was well-tolerated by the patient.

DISCUSSION

Osteoma cutis, a rare intramembranous ossification condition in which mesenchymal cells differentiate into bone without previous cartilage formation, is a hallmark of several genetic conditions, including AHO.[2] AHO characterized by round face, short stature, brachydactyly, subcutaneous ossification and dental anomalies is due to an inactivating mutation in GNAS1. AHO is commonly associated with pseudohypoparathyroidism type 1A, as was the case with this patient. Cutaneous ossification can cause severe disability, joint stiffness, pain, and inflammatory flares. Anetoderma has been hypothesized to represent the precursor lesion to osteoma cutis within this patient population. Histologically, the dermal lesions that stain SMA+ are thought to represent primitive mesenchymal osteoprogenitor cells. SMA expression is a marker of early osteoprogenitor cells within the periosteum, further supporting this hypothesis. STS increases calcium solubility, causes vasodilation and also has an antioxidant effect on endothelial cells.[3] Review of the literature has found that 91.7% of pediatric patients responded to sodium thiosulfate treatment in the setting of ectopic calcification with an excellent safety profile.[1] For the patient presented in this case, topical STS was utilized to help prevent progression of the precursor anetodermic lesions to osteoma cutis and improve overall quality of life.

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CASE 46: Infectious Chronic Granulomas in TAP2 Deficiency Syndrome

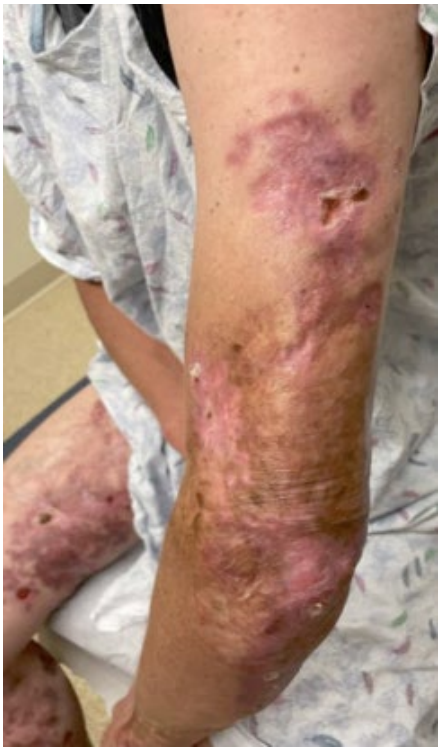
Authors: Margaret Mercante BA, Shira Lanyi MD, Barrett Zlotoff MD

HISTORY

A 38-year-old male from the Mennonite community with MHC class I deficiency presented for ulcerating lesions. Patient has a personal history of skin lesions beginning at 18 months of age which progressed to ulcerations by age 3-4 after completing childhood vaccines. Due to family history of chronic granulomas due to MHC class I deficiency, the patient was tested by genetics and was positive for homozygous TAP2 deletion. Skin biopsy showed necrotizing granulomas. He was treated with systemic steroids for years but developed osteoporosis and was started on adalimumab in the last five years, along with other treatments.

PHYSICAL EXAMINATION

Examination of the skin revealed widespread atrophic and shiny plaques on left upper arm and right leg with violaceous and dark red surrounding rim and overlying telangiectasias, ulceration and areas of prior grafting.



PATHOLOGY

Biopsy revealed palisaded granulomatous dermatitis. Sections show a superficial and deep dermal infiltrate of lymphocytes with plasma cells and mononuclear and multinucleated histiocytes. The histiocytes form epithelioid granulomas, some palisading around a central area of necrobiosis.

TREATMENT

Findings were consistent with chronic granulomatous skin diseases, possibly rubella virus related, from childhood MMR vaccine. He was started on plaquenil, doxycycline, and nitazoxanide due to suggested efficacy of nitazoxanide and plaquenil combination against rubella and inflammation virus. Doxycycline was added both for its anti-inflammatory effect and antimicrobial activity.

DISCUSSION

Bare lymphocyte syndrome due to TAP2 mutations is characterized by a severe down-regulation of MHC class I expression. TAP2 deficiency is exceedingly rare, and as exemplified by this case, often runs in consanguineous families. Disease typically presents within the first 6 years of life with recurrent bacterial infections of the upper respiratory tract. Patients develop necrotizing granulomatous skin lesions that primarily appear after the age of 15.[1] This patient is the older cousin of our other TAP2 deficiency SEC case and represents severe skin manifestations secondary to rubella virus-associated granulomas after receiving the MMR vaccine in childhood. His diagnosis came in young adulthood after failed treatments for what was suspected ulcerating NLD. Treatment options are limited. Immunosuppressive medications in these patients are contraindicated due to the concern of progression of both the skin and pulmonary disease. TNF-alpha inhibitors, can demonstrate some efficacy in treating skin manifestations, but the risk of disseminating walled off rubella is of concern. Other therapies that have some efficacy against rubella associated granulomas include nitazoxanide in combination with hydroxychloroquine, which does have some anti-viral effects for these patients. The most efficacious therapy at this time is allogeneic stem cell transplant, however, severe GVHD may complicate the course.[1] In addition, MHC class I protein expression is not restricted to hematopoietic stem cells and may therefore not correct all disease manifestations. These patients represent a clinical challenge.

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CASE 47: Obinutuzumab for Pemphigus Vulgaris

Authors: Margaret Mercante BA, Shira Lanyi MD, R Hal Flowers MD

HISTORY

A 43-year-old female presented for follow up pemphigus vulgaris (PV). In 2021, her initial severe PV was treated with high dose steroids and lymphoma dosing of rituximab (RTX). Upon receiving several rounds of maintenance therapy due to mild disease recurrence, she developed a persistent, erythematous pruritic eruption of the arms, legs, and trunk and an itchy throat, headache, and wheezing upon infusion. Treatment was discontinued, yet her pruritus and rash did not recede.

PHYSICAL EXAMINATION

Patient exam was notable for numerous edematous pink papules coalescing into plaques on the trunk, thighs, lower legs, and upper arms. Persistent post-inflammatory hyperpigmentation from prior pemphigus was present on the back.

PATHOLOGY

The biopsy demonstrated a superior perivascular dermatitis with numerous eosinophils and a negative DIF, favored to represent RTX hypersensitivity reaction.

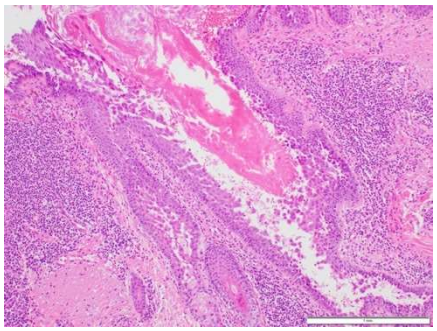
TREATMENT

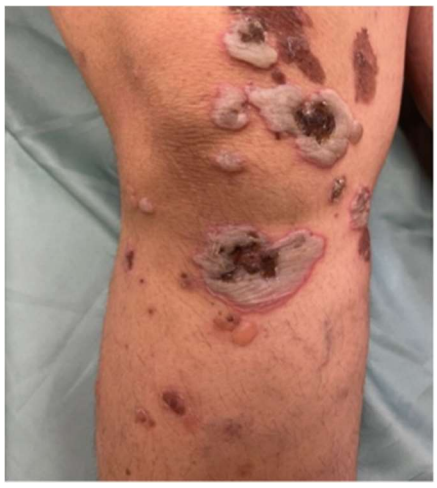
Given recurrent pemphigus with superficial blistering and high titer serum Desmoglein 1 antibodies (Dsg 1), retreatment was necessary. After failure to respond fully to mycophenolate mofetil, azathioprine, and IVIG following a taper of prednisone, B-cell depletion therapy with Obinutuzumab was pursued. She tolerated two of three infusions well, but due to development of a headache, did not complete her third infusion. Follow up to evaluate Dsg response is upcoming.

DISCUSSION

PV is a severe autoimmune blistering skin disease characterized by autoantibodies directed against mucocutaneous desmosomal proteins. B-cell depletion is highly effective in pemphigus, resulting in the widespread use of RTX for this disease. While generally well tolerated, hypersensitivity and infusion reactions are not uncommon. For these patients, obinutuzumab might prove to be a potential treatment choice.

Obinutuzumab and RTX both target CD20+ B-cells and induce antibody-dependent cell death. However, rituximab is a type I chimeric mouse/human monoclonal antibody, and obinutuzumab is a humanized type II monoclonal antibody that replaces the mouse antibody with human sequences, making it less immunogenic. The Fc region of obinutuzumab is more effective than rituximab in recruiting effector immune cells, leading to antibody-dependent cellular cytoxicity, phagocytosis and direct cell death. [1] This drug may be a treatment option for PV patients in the setting of RTX hypersensitivity reactions. [2]





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CASE 48: Cold Agglutinin Disease with Livedo Reticularis

Authors: Clara Smith BS, Shira Lanyi MD, R. Hal Flowers MD

HISTORY

A 72-year-old male with a past medical history of myelofibrosis status post matched unrelated donor stem cell transplant and cold agglutinin disease (CAD), specifically hemolytic anemia, presented for a worsening, diffuse rash on the lower legs and abdomen. Though asymptomatic, the patient noted improvement in coloration with warmth and worsening with cold. Patient was followed closely by his hematology and oncology team who noted recent worsening thrombocytopenia and made a diagnosis of immune thrombocytopenic purpura (ITP) in association with autoimmune hemolytic anemia, also called Evan's syndrome.

PHYSICAL EXAMINATION

On the bilateral lower extremities and abdomen was a generalized, macular, non-blanching rash in a reticular pattern with violaceous discoloration.

LABORATORY DATA

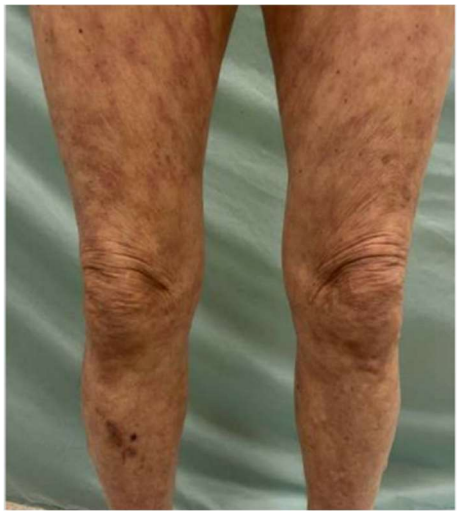
Serologic work up was notable for a cold agglutinin titer of 1:1024. Haptoglobin was less than 1, LDH 346, and reticulocyte count was elevated, diagnostic of hemolysis. Platelets were 71 and hemoglobin was 7.9.

TREATMENT

The patient was diagnosed with livedo reticularis in the setting of CAD. The patient was vaccinated for encapsulated organisms and started on sutimlimab, a C1s inhibitor, prior to stabilizing on rituximab. Given the history, the dermatology team opted against further biopsy and workup of the livedo reticularis and to follow up if the livedo reticularis did not improve following his rituximab and sutilimab infusions. On follow up with oncology several months later, the livedo had resolved.

DISCUSSION

Livedo reticularis (LR) is a cutaneous clinical finding characterized by either a transient or persistent violaceous net-like pattern. LR is a manifestation of disruption of cutaneous blood flow that may occur in a plethora of physiologic or pathologic states. CAD is a rare cause of LR. CAD is a form of acquired autoimmune hemolytic anemia secondary to recent infections, particularly mycoplasma pneumonia, or an underlying lymphoproliferative disorder, as in this patient. In our case, the presence of cold agglutinins and subsequent hemolysis resulted in severe anemia and decreased oxygenated blood flow to the skin likely leading to LR. Primary treatment of LR includes treating the underlying disorder and maintaining the patient in a warm environment. Sutilimab was chosen as treatment as it selectively inhibits the classical complement pathway and has been shown to demonstrate efficacy against ITP. In this patient, this therapy resulted in complete resolution of the clinical findings of LR and is a fascinating correlation between cutaneous manifestations of disease and physiologic conditions.



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CASE 49: Tuberous sclerosis diagnosed in adulthood

Authors: Muhammad Zulfiqar, Wilson Omesiete MD, Barrett Zlotoff MD

HISTORY

A 32-year-old patient with a history of polycystic kidney disease (PCKD) status post renal transplant presented for routine skin cancer screening. During the encounter, a personal history of intellectual disability and a family history of renal pathology were identified. The patient had no history of seizures and had never undergone genetic testing.

PHYSICAL EXAMINATION

Examination revealed numerous fleshy, pedunculated, perinasal papules consistent with adenoma sebaceum (angiofibromas), scattered hypopigmented macules and patches on the trunk and extremities consistent with confetti macules and ash-leaf spots, and a flesh-colored, mamillated plaque on the lower back consistent with a shagreen patch.

PATHOLOGY

Two initial biopsies were obtained from lesions in the perinasal area. Histopathology revealed benign polypoid lesions with multiple dilated blood vessels in a background of fibrous tissue with scattered stellate fibroblasts.Patient was unable to complete genetic testing due to the associated expense.

DISCUSSION

Tuberous Sclerosis (TS) is an autosomal dominant disorder that often presents in early childhood. It is characterized by the development of benign tumors in multiple organs such as the brain, skin, kidneys, and lungs. Common skin manifestations include hypomelanotic macules and patches, facial angiofibromas, and shagreen patches. A patient may also present with seizures, developmental delay, and behavioral issues. Although rare, tuberous sclerosis can present in adults with complications of renal angiomyolipomas, lymphangioleiomyomatosis, and an increased risk of certain cancers. As such, regular monitoring of other organ systems is important. Similar dermatologic manifestations such as confetti-like hypopigmentation, angiofibromas, and collagenomas can also be observed in Multiple Endocrine Neoplasia Type 1. However notably different systemic manifestations are observed in those cases. Notably, the PKD-1 and TSC-2 genes are both localized on chromosome 16, and large deletions may result in both diseases concomitantly. We present this case to highlight the morphology of the cutaneous findings of TS in darker skin and remind the clinician to consider the diagnosis even in adult patients.

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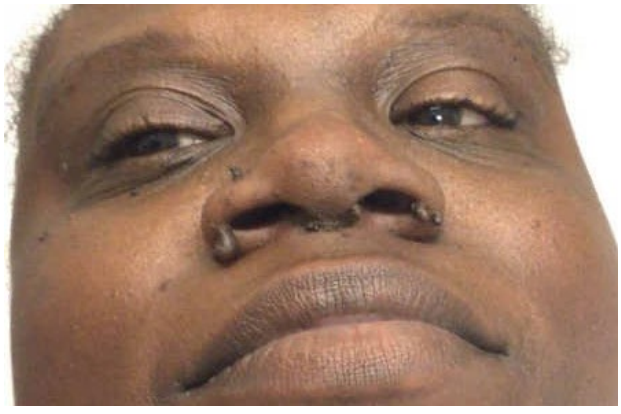

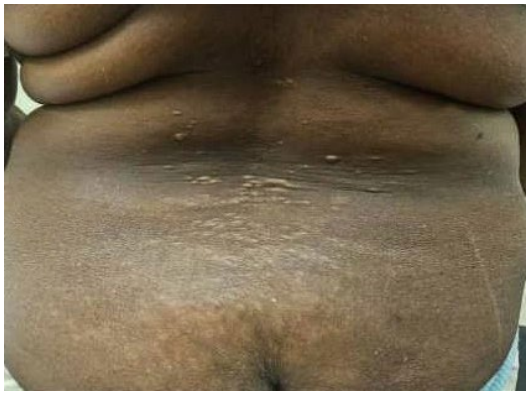

CASE 50: Autosomal Dominant Albopapuloid Epidermolysis Bullosa Pruriginosa

Authors: Muhammad Zulfiqar, Wilson Omesiete MD, R Hal Flowers MD

HISTORY

A 27-year-old female with a history of eczema presented with lower extremity blistering, nail dystrophy, and numerous papular scars on her hands and feet. The blisters were intensely pruritic and frequently bled when manipulated. She described similar symptoms in her father and youngest daughter.

PHYSICAL EXAMINATION

Examination revealed atrophic white-pink scars and slightly hypertrophic pink flat-topped papules admixed with milia on the pretibial legs. She had nail dystrophy bilaterally on the first through third fingers and pink, papular scarring over the bilateral dorsal hands.

RADIOLOGY/LABORATORY DATA/PATHOLOGY

Genetic testing revealed a pathogenic variant in the COL7A1 gene, specifically c.6017G > C (p.Gly2006Ala). This allowed for the confirmation of the diagnosis of autosomal dominant dystrophic epidermolysis bullosa pruriginosa (EBP).

DISCUSSION

Autosomal Dominant Albopapuloid Epidermolysis Bullosa Pruriginosa (EBP) is caused by COL7A1 gene mutations encoding type VII collagen and is characterized by trauma-induced blisters, intense pruritus, lichenified lesions, albopapuloid lesions, milia, and nail dystrophy.[1,2] The presence of albopapuloid lesions, which can appear as slightly depressed, white-pink areas resembling atrophic scars or as small ivory-white papules, is a distinctive feature and is rarely seen in patients with EBP. Notably, dupilumab has been successfully utilized in multiple patients as a recent treatment for this particular subtype of EB.[3]

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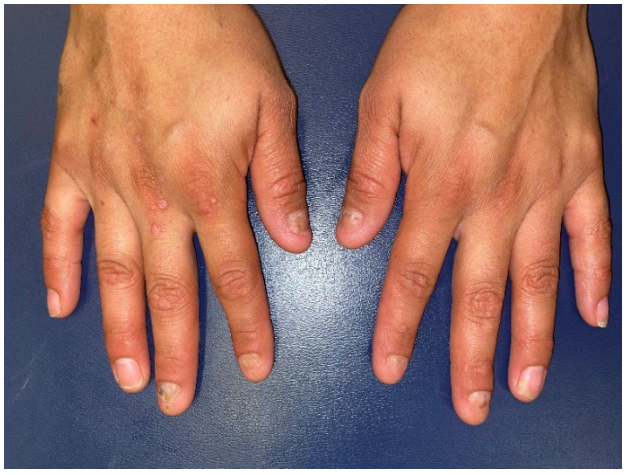

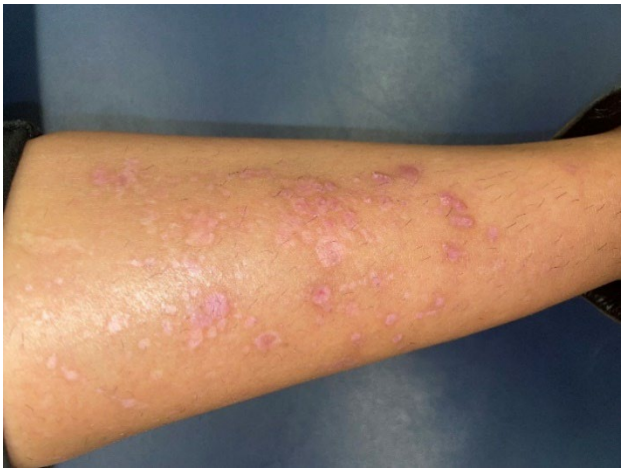

CASE 51: Dermal VZV in an Immunosuppressed Patient

Authors: Olivia Lim BA, Wilson Omesiete MD, R. Hal Flowers MD

HISTORY

A 41-year-old woman with stable SLE, developmental delay, and epilepsy presented with nonhealing left-sided leg ulcers which had been present for 6 months and were treated with multiple courses of antibiotics. Her SLE was inactive on methotrexate 10 mg weekly and prednisone 5 mg daily and had previously manifested with polyarthritis, positive ANA (1:640), anti-dsDNA, pancytopenia, and proteinuria.

PHYSICAL EXAMINATION

The left lower extremity exhibited multiple large malodorous ulcers with surrounding erythema, overlying necrotic crust, and pitting edema. The right had patches of post-inflammatory hyperpigmentation at sites of prior ulceration. Both legs were warm to the touch with palpable dorsalis pedis pulses.

LABORATORY DATA/PATHOLOGY

Incisional biopsy demonstrated mixed dermal and subcutaneous inflammation. Given evidence of viral cytopathic effect, immunohistochemical staining for Varicella zoster virus (VZV) was performed and found to be diffusely positive in the dermis with no evidence of epidermal VZV. PCR swabs of multiple ulcers confirmed the presence of VZV and ruled out Herpes simplex virus (HSV)-1 and HSV-2.

TREATMENT

Treatment was initiated with valacyclovir 1000 mg three times daily and oral cefadroxil 500 mg twice daily for possible bacterial superinfection. Ulcers began showing improvement after two weeks of treatment, and she was continued on the antiviral regimen for 4 months with slow but sustained improvement in her ulceration.

DISCUSSION

Herpes zoster (HZ) is a cutaneous viral disease that typically presents with a dermatomal vesicular eruption. Immunosuppressed patients are more likely to have atypical HZ involving chronic ulceration and disseminated distribution, making diagnosis a challenge. This unique case of HZ in a woman with systemic lupus on immunosuppressive therapy manifested as persistent lower extremity ulceration with diffuse dermal and endothelial infection and secondary panniculitis without epidermal involvement. Recognizing atypical manifestations of HZ in the setting of immune compromise is critical to accurate diagnosis and prompt therapy.

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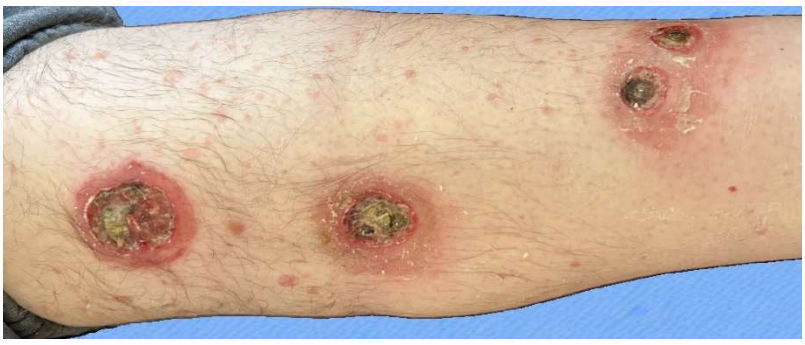
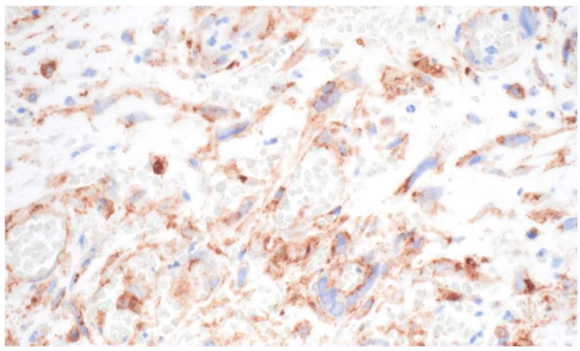


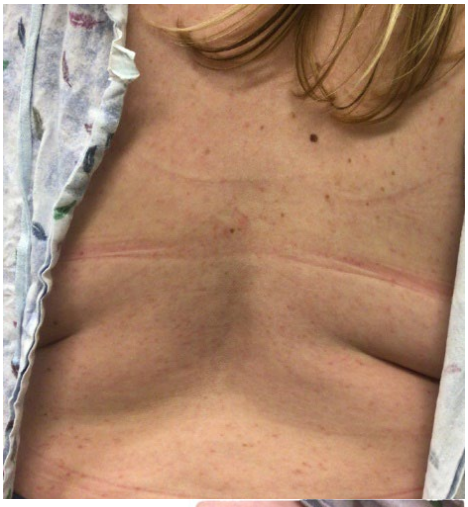


CASE 52: Cutaneous mastocytosis treated with avapritinib

Authors: Nakul Dar, Gabriella Norbert, Wilson Omesiete MD, R. Hal Flowers MD

HISTORY

A 44-year-old woman presented with a several year history of a pruritic rash on her back that spread to the trunk and proximal extremities. The rash worsened with sunlight and hot showers. She had failed treatment with cetirizine, fexofenadine, ranitidine, natural light, prednisone and cromolyn.

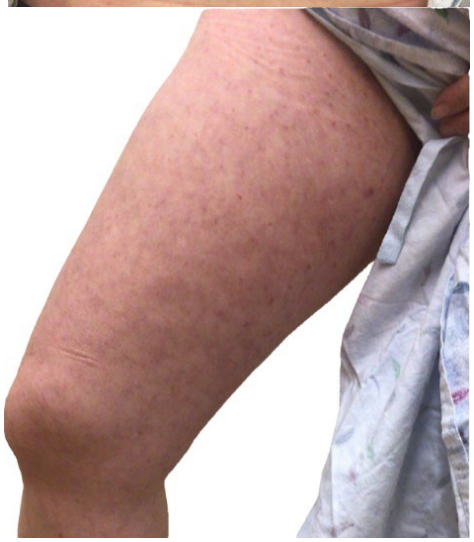


PHYSICAL EXAMINATION

Innumerable, monomorphic, reddish-brown, telangiectatic macules and thin papules were concentrated on the inferior trunk and medial thighs with a positive Darier's sign. There was no palpable lymphadenopathy.

RADIOLOGY/LABORATORY DATA/PATHOLOGY

Skin biopsy revealed notable superficial dermis telangiectasias with ectatic vessels surrounded by a mononuclear cell infiltrate with increased mast cells. The findings were consistent with cutaneous mastocytosis. Bone marrow biopsy showed atypical mast cells with a pathogenic KIT D816V mutation. Complete blood counts and serum tryptase were within normal limits.



TREATMENT

No notable benefit was seen from pimecrolimus 1% cream, tacrolimus 0.1% ointment, phototherapy, ruxolitinib phosphate 1.5% cream, pulse dye laser therapy, or compounded topical imatinib. Oral avapritinib 25 mg daily resulted in dramatic improvement in both pruritus and flare frequency.

DISCUSSION

Cutaneous mastocytosis can be a manifestation of indolent systemic mastocytosis and commonly presents as yellowish to red-brown macules or papules. Diagnosis is supported by characteristic lesions, a positive Darier sign, and biopsy showing increased mast cells or KIT mutations. Indolent systemic mastocytosis, marked by abnormal mast cell proliferation, affects adults and can cause mild symptoms like flushing and diarrhea. Avapritinib, a tyrosine kinase inhibitor targeting the KIT D816V mutation, is now FDA-approved for this condition after successful PIONEER trial results .[1] It is approved for up to 200 mg daily for mastocytosis, but skin efficacy has been seen at doses of 25 mg daily.[2]

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CASE 53: Epidermal Nevus Syndrome with Visceral Involvement

Authors: Josephine Arewa BA, Wilson Omesiete MD, Barrett Zlotoff MD

HISTORY

A 47-year-old female presented with previously diagnosed widespread hairless, painful, pruritic lesions of the entire body with associated eye irritation and dysphagia previously diagnosed as epidermal nevus syndrome. She had the lesions since birth but has reported gradual worsening of symptoms since puberty and associated exertional dyspnea, bloating, constipation, and pelvic pain. Flares are associated with menses. The cutaneous lesions have been treated over the past several decades with serial CO_2 laser ablation. Topical MEK inhibitor, trametinib 1% cream, was trialed but discontinued due to minimal improvement and adverse effects including acne and periorificial dermatitis.

PHYSICAL EXAMINATION

Widespread hairless, yellowish-brown, verrucous and hyperkeratotic plaques with variable thickness and strong midline delineation showing right-sided predominance and a Blaschkonian distribution in areas with involvement of the head, trunk, and extremities.

PATHOLOGY/RADIOLOGY/LABORATORY DATA

Histology revealed epidermal hyperplasia with papillomatosis and associated inflammation. Lesional genetic testing confirmed HRAS Gly12Asp mutation. Endoscopy identified esophageal epidermal nevi and bladder papillomas. Ophthalmologic assessment identified subepithelial corneal scarring and labs show hypophosphatemia. Radiographic imaging demonstrated thoracolumbar scoliosis and bilateral renal calculi but no changes consistent with rickets.

DISCUSSION

Epidermal nevus syndrome is an umbrella term encompassing numerous presentations with cutaneous hamartomas that are often associated with ophthalmologic, neurologic, and skeletal abnormalities. In this case, multilineage somatic activating mutations in HRAS (Gly12Asp) lead to mosaic cutaneous, visceral, and skeletal lesions, elevated FGF23 which leads to hypophosphatemia. Many features observed here are compatible with Schimmelpenning syndrome. In addition to cutaneous and visceral hamartomas- these patients can develop urothelial cancers, rhabdomyosarcoma, and complications associated with congenital cranial arachnoid cysts, requiring monitoring.

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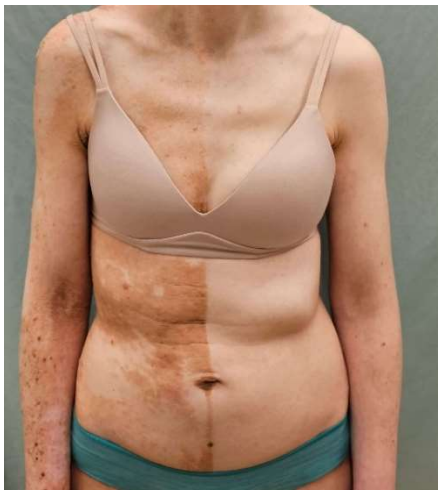

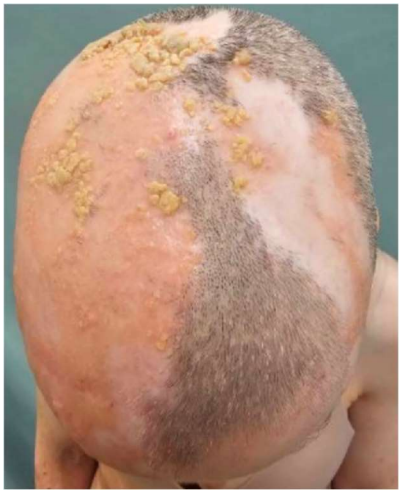

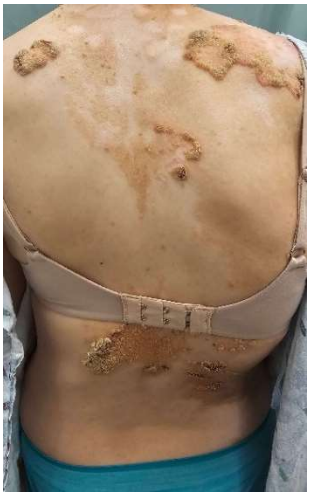

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