

**ABSTRACT BOOK RDS 2017 ANNUAL MEETING - University of California San Diego School of Medicine**

**DEFINITION WORKSHOP**

**DEVELOPMENT OF CLASSIFICATION CRITERIA FOR DERMATOMYOSITIS UTILIZING THE DELPHI TECHNIQUE**

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The new ACR/EULAR myositis criteria is able to identify patients with dermatomyositis (DM), however, a significant proportion with amyopathic disease do not meet 2 out of the 3 the hallmark skin signs and fail the classification. To allow a more inclusive definition of cutaneous DM, we pursued to develop a skin-focused classification criteria based on the consensus of international experts in the Rheum-Derm field. An extensive literature review was done. The steering committee then invited experts from North America, Europe and Asia to participate in on-line and face-to-face group discussions to generate items for the pre-Delphi questionnaire. The results of the pre-Delphi were presented at the RDS meeting in November 2016. Subsequent nominal group discussions were held to create items for inclusion in the actual Delphi. Using the REDCap application, participants of the current Delphi are able to rate the appropriateness of certain DM findings broken into the categories of distribution, morphology, symptoms, antibodies, histology, and contextual factors. The relevance score ranges from 1 to 100, with a grade of 100 recommending the item as a distinguishing factor from mimickers. A pre-specified cutoff of items will be decided later by the steering committee. Distribution statistics (median and interquartile range of the proposed items) will be calculated to create a rank-ordered list. The results of this first round of Delphi will be presented in the upcoming RDS meeting. It is our

ultimate goal that the DM classification criteria will create well-defined cohorts for clinical research on novel treatments for this disease.

## **ORAL SESSION I**

(10:00)

### **LASER DOPPLER IMAGING: AN OBJECTIVE OUTCOME MEASURE FOR ASSESSMENT OF CUTANEOUS LUPUS ERYTHEMATOSUS**

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**Objectives:** To assess inter-rater reliability of Laser Doppler Imaging (LDI) and evaluate its validity against gold standard; histology from skin biopsy and other clinical tools including modified Localised Cutaneous LE Disease Area and Severity Index (LCLASI) and Visual Analogue Scale (VAS) scoring of photographs.

**Methods:** A prospective observational study was conducted in patients with CLE flare. Disease activity was assessed using LCLASI and measured using a high resolution LDI system (Moor Instruments) by two raters. Skin biopsy was scored as 0=non-active, 1=mild activity and 2=active. Photographs were assessed by two clinicians using 100mm VAS. Inter-rater reliability and VAS agreement were analysed using Bland-Altman limits of agreement (LOA) and correlation between histology and LDI, LCLASI and VAS using Kendall's Tau-a.

**Results:** 20 patients were studied (19 female, median age 47.2 years and 6 smokers). CLE type were: acute CLE=7, subacute CLE=6 and chronic CLE=7. The inter-rater agreement for LDI was fair-to-good; mean difference 8.3 (95% CI LOA -101 to 85) perfusion unit vs average. While the agreement for VAS scores was fair; mean difference 7.8 (95% CI LOA -26 to 42) mm versus average. In 10 patients with skin biopsy, correlation with histology was better for LDI (tau-a=0.56) than LCLASI [-0.04; difference 0.60 (90% CI 0.21, 0.99)] or VAS [-0.16; 0.71 (0.13, 1.29)].

**Conclusion:** LDI provides a valid quantitative measure of inflammation in CLE. It has a better correlation with histology compared to currently used clinical tools. Further validation and longitudinal analysis will provide evidence on its usefulness in clinical practice.

(10:12)

### **ALTERNATIVE ANTIMALARIAL THERAPY IN PATIENTS WHO ARE HYDROXYCHLOROQUINE ALLERGIC OR INTOLERANT.**

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Antimalarial medications are effective and widely used in the treatment of autoimmune connective tissue diseases, including cutaneous lupus erythematosus (CLE). However, adverse cutaneous reactions to antimalarials are relatively common. The incidence of hypersensitivity

recurrence is unknown in patients who develop a cutaneous hypersensitivity reaction to an antimalarial and are subsequently re-challenged with an alternative antimalarial. This retrospective review includes 11 patients with CLE who experienced cutaneous hypersensitivity following treatment with hydroxychloroquine (HCQ), and who were subsequently treated with alternative antimalarial therapy. Data was also collected on 2 patients who were intolerant of HCQ and subsequently challenged with an alternative antimalarial. No patient experienced a severe reaction to initial antimalarial therapy. Of 11 patients with documented HCQ cutaneous hypersensitivity, all subsequently tolerated either chloroquine (CQ) (N=8) or quinacrine (N=3, one of the three had hypersensitivity previously to both HCQ and CQ). Of 2 patients who were HCQ intolerant due to side effects other than hypersensitivity, both tolerated CQ and achieved at least moderate disease control. Overall, of the HCQ hypersensitive or intolerant patients in this study, 100% tolerated an alternative antimalarial, with 10 tolerating CQ and 3 tolerating quinacrine. All patients with known disease outcomes (N=8) improved to some degree with an alternative antimalarial regimen, with 25% requiring additional systemic medications. This study demonstrates the safety and efficacy of alternative antimalarial therapy in patients unable to continue treatment with HCQ

(10:24 AM)

### **LENALIDOMIDE TREATMENT OF CUTANEOUS LUPUS ERYTHEMATOSUS: THE MAYO CLINIC EXPERIENCE**

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**Background:** Published case series describe lenalidomide as an effective treatment of refractory cutaneous lupus erythematosus (CLE).

**Objectives:** The present study aimed to further characterize lenalidomide use in the treatment of CLE.

**Methods:** A retrospective review of patients treated with lenalidomide for CLE from January 1, 2000, to December 17, 2014, was conducted.

**Results:** Eight of the nine patients (89%) were women. Their median age at initiation of lenalidomide was 62 years (range: 41–86 years). Subtypes of CLE included discoid lupus erythematosus (DLE) (n = 6), lupus panniculitis (n = 2), and subacute CLE (n = 1). Before the initiation of lenalidomide, all patients had been previously treated unsuccessfully or were intolerant to at least one antimalarial and one immunosuppressive agent. With lenalidomide, five patients achieved a complete response (CR), two a partial response, and two had no response (lupus panniculitis). Time to initial response (dose range: 2.5–10.0 mg/d) varied from 2 weeks to 3 months; the median time to CR in five patients was 3 months (range: 3–6 months). The median duration of lenalidomide therapy was 12 months (range: 2–67 months). The median duration of follow-up was 48 months (range: 20–103 months). Adverse effects included mild leukopenia; one patient had deep vein thrombosis of unclear etiology during a hospitalization. No patients developed or showed progression of systemic LE while receiving lenalidomide.

**Conclusions:** Lenalidomide was effective for the treatment of CLE (particularly DLE) but not for the treatment of lupus panniculitis in this series.

Note: This abstract is from the following publication: *Int J Dermatol.* 2016 Aug;55(8):e431-9.

(10:36 AM)

## **MYELOID-DERIVED SUPPRESSOR CELLS (MDSCs) ARE ELEVATED IN THE BLOOD OF DISCOID LUPUS ERYTHEMATOSUS PATIENTS**

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MDSCs are the most potent T cell suppressors, and are markedly proliferated in inflammatory skin diseases such as psoriasis and alopecia areata. Because MDSCs have yet to be studied in patients with discoid lupus erythematosus (DLE), we characterized the MDSC population in DLE peripheral blood mononuclear cells (PBMCs) and evaluated their ability to suppress autologous T cells. PBMCs from 32 DLE patients and 15 age- and gender-matched controls were analyzed using flow cytometry. Monocytic MDSCs were identified by the phenotype of CD14<sup>+</sup> HLA-DR<sup>neg/low</sup>. Furthermore, MDSCs and T cells were purified from DLE PBMCs and co-cultured at different ratios of these cells. T cell function was measured by secretion of IFN- $\gamma$  by ELISA. To examine whether the DC-HIL immune checkpoint critically mediates the T cell suppressor function of MDSCs, 50  $\mu$ g/mL of anti-DC-HIL mAb, which can inhibit MDSC activity, was added to the co-cultures. Monocytic MDSCs in DLE PBMCs (median: 2.05%, IQR: 0.51%-5.07%) were significantly higher compared to healthy control PBMCs (median: 0.5%, IQR: 0.1%-1.39%, p=0.02). Subset analyses of DLE patients showed no significance between those with and without systemic lupus, and treatments. DLE MDSCs (N=2) were found to suppress autologous activated T-cells in a dose-dependent manner, and further addition of anti-DC-HIL mAb partially blocked T cell suppression. The MDSC population is expanded in DLE peripheral blood, and demonstrates dose-dependent inhibition of T cell activation in *in vitro* assays. Their up-regulation in DLE blood may represent the body's response to limiting disease severity, since most patients had mild disease activity.

(10:48 AM)

## **NATURAL HISTORY OF DISEASE ACTIVITY AND DAMAGE IN PATIENTS WITH CUTANEOUS LUPUS ERYTHEMATOSUS ON STANDARD OF CARE TREATMENTS USING LONGITUDINAL REGISTRIES FROM TWO ACADEMIC DERMATOLOGY CENTERS**

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We sought to characterize the disease course of patients with cutaneous lupus erythematosus (CLE) on standard-of-care treatments and identify clinical features associated with different disease activity and damage trends. We performed a prospective cohort study of patients with CLE with  $\geq 3$  study visits within a 2-year time period at two academic dermatology clinics. Improving, stable, and worsening disease activity and damage trends were determined by area under the curve (AUC) and change (slope) in Cutaneous Lupus Erythematosus Disease Activity and Severity Index (CLASI) scores/time, and rates were compared between patient subgroups. 81.8% of patients (N=83) with initial CLASI activity (CLASI-A) scores  $\geq 10$  (N=33) had improving disease activity, compared with 16.0% of those with initial CLASI-A  $\leq 9$  (N=50).

Patients with baseline CLASI-A  $\leq 9$  had higher percentages of stable (56.0% vs. 9.1%) and worsening disease activity (28.0% vs. 9.1%) than those with initial CLASI-A  $\geq 10$  ( $p < 0.0001$ ). Linear regression analyses showed significant improvement in CLASI-A scores over time in patients with baseline CLASI-A  $\geq 10$  ( $p < 0.0001$ ), baseline CLASI damage (CLASI-D)  $\geq 10$  ( $p = 0.0001$ ), African Americans ( $p = 0.049$ ), and disease duration  $\leq 1$  year ( $p = 0.01$ ). 46.4% and 5.4% of patients with baseline CLASI-D  $\geq 10$  ( $N = 28$ ) and  $\leq 9$  ( $N = 55$ ), respectively, had improving disease damage trends. Patients with baseline CLASI-D  $\leq 9$  had higher percentages of stable (67.3% vs. 35.7%) and worsening disease activity (27.3% vs. 17.9%) than those with initial CLASI-A  $\geq 10$  ( $p = 0.0003$ ). Thus, most patients with high and low baseline CLASI-A and -D showed improving and stable trends, respectively. This data may serve as historical controls for future clinical trials.

(11:00 AM)

### **CHILBLAIN LUPUS ERYTHEMATOSUS - CLINICAL AND HISTOLOGIC CHARACTERIZATION OF AN INSTITUTIONAL COHORT**

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Chilblain lupus erythematosus (CHLE) is a rare form of chronic cutaneous lupus characterized by lesions in an acral distribution exacerbated by exposure to cold. There is a paucity of literature concerning patients with CHLE, with the last cohort reported over 15 years ago. Additionally, diagnostic criteria for CHLE are not well-defined.

**Methods:** We conducted a retrospective chart review of CHLE patients seen at the Cleveland Clinic from 1980-2015. Patients were included based if they satisfied one “required” criteria (acral skin lesions induced by exposure to cold) and at least 1 of 2 “major” criteria: (1) documented histologic evidence of CLE, (2) met ACR criteria for SLE.

**Results:** We identified 18 patients who met our criteria for a diagnosis of CHLE (16 women; 2 men). CHLE lesions were most commonly found on fingers ( $n = 16$  pts), hands (11), toes (10), feet (7), and ears (4). Most patients (11/18) had a diagnosis of SLE prior to CHLE onset and suffered from Raynaud’s phenomenon (13/18). All patients were ANA positive; other common autoantibodies included dsDNA(10/15), SSA(7/16) and RNP(7/15). Twelve patients had biopsies of a CHLE lesion, and these typically displayed superficial and deep inflammation with variable interface change. Treatment strategies varied, but included both immunomodulatory medications and vasoactive medications. The majority of patients had improvement of their CHLE lesions with treatment.

**Conclusions:** CHLE is a rare subtype of CLE seen at our institution, and mainly occurred in women with SLE. Treatment with immunomodulatory and/or vasoactive medications tends to be beneficial in affected patients.

## **ORAL SESSION II**

(11:30 AM)

### **TOFACITINIB CITRATE FOR REFRACTORY CUTANEOUS DERMATOMYOSITIS: AN EXPANDED SERIES DEMONSTRATING IMPROVEMENT IN SEVEN**

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Janus kinase (JAK) inhibitors are currently being investigated in various inflammatory skin diseases given their ability to modulate signaling of multiple cytokines including interferon. We have previously published a series of tofacitinib demonstrating improvement in 3 patients with recalcitrant cutaneous dermatomyositis (DM). We aim to present an expanded series of seven patients with refractory cutaneous DM treated with tofacitinib. All 7 patients are female with an age range of 30 to 59 years. Four patients were treated with tofacitinib 10 mg twice daily; the remaining 3 were treated with 5 mg twice daily. Classic DM was present in 5 patients, whereas 2 had amyopathic DM. Treatment duration ranged from 4 to 20 months. Disease activity was assessed using the validated Cutaneous Dermatomyositis Disease Area and Severity Index (CDASI) score. All 7 patients experienced a clinically significant improvement in cutaneous DM, as defined by the most recent validation study to be a 4- to 5-point change in CDASI activity score. The mean improvement in CDASI activity score in our 7 patients was 13 points. All patients also noted improvement in pruritus. Tofacitinib was well tolerated, and no adverse effects were noted in our small cohort. Patients on higher doses had greater improvement, likely indicating a dose-response relationship. To our knowledge, this is the largest series of patients treated with tofacitinib for recalcitrant cutaneous DM to date. Given the limited number of agents available for management of this condition, this series highlights an alternative treatment option for patients with refractory cutaneous DM. Category: Lichenoid inflammatory skin disease (dermatomyositis)

(11:42 AM)

### **APPLICABILITY OF OF EULAR/ACR CLASSIFICATION CRITERIA FOR DERMATOMYOSITIS TO AMYOPATHIC DISEASE**

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**Background:** Existing classification systems for idiopathic inflammatory myopathies (IIM) fail to classify and/or diagnose patients with amyopathic dermatomyositis (ADM).

**Objective:** In light of the new European League Against Rheumatism (EULAR)/American College of Rheumatology (ACR) classification criteria for IIM, we sought to evaluate the

likelihood of the skin variables included in the EULAR/ACR criteria, Gottron's sign, Gottron's papules, and heliotrope rash, to give a high probability of classifying patients with ADM.

**Methods:** This retrospective study evaluated 211 adult patients with dermatomyositis at the University of Pennsylvania. The EULAR/ACR criteria were used to determine the probability of classification for patients with ADM.

**Results:** 73.7% of patients with ADM would be classified as having a reasonable probability of IIM based on the new EULAR/ACR criteria. 26.3% of patients with ADM would not meet the suggested 55% minimum probability cutoff to be classified based on the EULAR/ACR criteria.

**Limitations:** This study was conducted with retrospective design at a dermatology practice at a tertiary academic medical.

**Conclusions:** The three skin variables included in the EULAR/ACR classification criteria for IIM may not be adequate for classifying ADM. It is important to consider additional variables such as skin biopsy to encompass more of these patients and prevent the inclusion of any skin diseases mimicking ADM.

(11:54 AM)

## **MALIGNANCIES MISSED BY AGE- AND GENDER-APPROPRIATE SCREENING IN DERMATOMYOSITIS PATIENTS AND AN UPDATE OF DERMATOMYOSITIS IN A LARGE PATIENT COHORT IN THE UNITED STATES**

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**Background:** Many of the more common types of malignancy associated with DM are not routinely screened for by age and gender appropriate USPSTF (United States Preventative Services Task Force) screening guidelines; thus, best malignancy screening practices are still widely debated.

**Objective:** To determine the percent of malignancies associated with DM that age and gender appropriate USPSTF recommended cancer screening would likely fail to capture.

**Methods:** The MarketScan database was queried for every patient diagnosed with DM. Patients were identified by entry of ICD-9 code 710.3 at least twice separated by 6 months with three years of time prior to and following the index diagnosis. Using ICD-9 codes, the number and types of cancers associated with the DM patients were determined. The list of malignancies in the DM cohort was compared to the list of USPSTF screened organs.

**Results:** 398 patients were identified. The majority of patients were 50 to 60 years-old ( $p < 0.0001$ ) followed by the 40 to 50 years old ( $p < 0.001$ ). 150 cancers were diagnosed. Of the 150 cancers, 75 (50%) were in organs not recommended for screening by age- and gender-appropriate USPSTF guidelines.

**Conclusions:** Here, we have found that age and gender appropriate screening according to the USPSTF guidelines would likely fail to capture half of malignancy in DM patients in our US study set. One of the major causes of death in dermatomyositis patients is malignancy; thus, more studies need to be completed to determine best cancer screening practices for DM patients.

(12:06 PM)

## **REEXAMINING "MECHANIC'S HANDS" AS A CHARACTERISTIC SKIN FINDING IN DERMATOMYOSITIS**

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Mechanic's hands is a poorly defined clinical finding that is found in a variety of rheumatologic diseases. Morphologic descriptions include hyperkeratosis on the sides of the digits that sometimes extend to the distal tips, diffuse fingerpad +/- palmar scale, and, more recently observed, linear scaly papules in a similar lateral distribution. The association of mechanic's hands with dermatomyositis (DM), although recognized, is still debatable. To better define and characterize the clinicopathologic features of mechanic's hands and, assess its true prevalence in DM, a literature search was performed using Pubmed from February 20, 1975 to December 2, 2016 using the terms "mechanic's hands", "mechanics hands", "dermatomyositis", "histopathology", and "antisynthetase syndrome". Following review of these studies, we found that mechanic's hands is a characteristic skin change in DM because (1) it is prevalent among DM patients, many of whom do not possess antisynthetase antibodies; (2) it coexists with other hallmark DM skin changes such as Gottron's papules, Gottron's sign and heliotrope rash; and, (3) biopsied cases of mechanic's hands reported in the literature show features of interface dermatitis, colloid bodies and interstitial mucin consistent with DM. The association of mechanic's hands with antisynthetase syndrome is also present in the literature, however, the relationship between DM and antisynthetase syndrome needs further clarification because both have overlapping clinical criteria. A more specific definition of mechanic's hands would help to determine its usefulness in classifying and clinically identifying patients with dermatomyositis, with implications related to subsequent screening for associated comorbidities, and inclusion to clinical trials.

(12:18 PM)

### **OUT-OF-POCKET PATIENT COSTS, ADDED VALUE OF PULMONARY EVALUATION, AND RADIATION EXPOSURE IN POSITRON EMISSION TOMOGRAPHY COMPARED WITH BROAD MALIGNANCY SURVEILLANCE IN DERMATOMYOSITIS: AN UPDATE.**

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**Background:** We previously presented data on costs of PET compared to broader malignancy screening. Questions were posed regarding out-of-pocket costs, radiation exposure, and other applications of PET in DM.

**Objective:** To compare out-of-pocket cost of PET to conventional malignancy screening panels for DM, review radiation exposure in PET, and the utility of pulmonary applications.

**Methods:** The MarketScan database was queried for every instance of PET and the broader screening panel from 2005 to 2014 and the mean patient deductible and copay costs were recorded and compared, as were the costs of pulmonary function testing (PFTs). A literature review was conducted to evaluate and quantify the radiation exposure of PET scan variants and their utility in pulmonary disease.



**Results:** The cost to patients (co-pay plus deductible) for whole body PET-CT was \$108.20 less for men and \$110.21 less for women. The total cost of PFTs was \$205.02.

**Conclusions:** Out-of-pocket costs to patients for PET were less expensive than conventional screening costs. PET-CT capabilities in evaluating interstitial lung disease may position it as an alternative to PFT's, adding further value. During a PET-CT, a patient absorbs around 25 milliSieverts (mSv) of radiation. 7-8 mSv derive from the 18-FDG, roughly the equivalent of a barium swallow. The remainder results from the CT. PET-CT is more expensive overall than conventional screening panels, but it is less expensive for patients. The additional radiation exposure from a PET-CT is relatively small. The capabilities of PET-CT in evaluating interstitial lung disease add value to this single-screening modality.

(12:30 PM)

### **FACTORS ASSOCIATED WITH CLINICAL REMISSION OF SKIN DISEASE IN DERMATOMYOSITIS**

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Quantitative estimates of the duration and severity of cutaneous disease are lacking for adult dermatomyositis (DM) patients. The Cutaneous Dermatomyositis Disease Area and Severity Index (CDASI) quantifies cutaneous disease activity (CDASI-a), and allows for objective assessments of disease severity. Our aims were to estimate the percentage of DM patients with clinically significant skin inflammation who achieve clinical remission (CR) during a 3-year follow-up period and to examine the relationship between skin disease course and selected clinical variables. CR was defined as a CDASI-a  $\leq$  5 with no individual erythema score  $>$ 1 and no ulcerations. In total, 74 adult DM patients with baseline CDASI-a scores  $\geq$  12 were included in our study. The median duration of follow-up was 17.5 (11-28) months, with a median of 4 (3-6) months between scores. Multivariable logistic regression analysis was performed using age, race, gender, amyopathic status, DM-associated malignancy, baseline CDASI-a, disease duration at baseline, time until first systemic therapy, autoantibody phenotype, and medication exposure as covariates. Overall, 28 patients (38%) entered CR. Increased age (OR 1.07, 95% CI 1.02-1.12;  $P=0.01$ ), DM-associated malignancy (OR 14.46, 95% CI 2.18-96.07;  $P=0.006$ ) and mycophenolate mofetil (OR 6.00, 95% CI 1.66-21.78;  $P=0.0064$ ) were significantly associated with CR in multivariable regression analysis. CR was relatively uncommon in our population despite the use of aggressive systemic therapy. Our findings suggest that MMF should be considered a first line agent for DM skin disease, while simultaneously highlighting a need for new therapies to more effectively treat skin disease in DM.

## **ORAL SESSION III**

(1:30 PM)

### **PAIN IN MORPHEA PATIENTS CORRELATES WITH A POOR QUALITY OF LIFE AND DOES NOT CORRELATE WITH DISEASE ACTIVITY**

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Although cosmetic and functional impairment are well described sequelae of morphea, pain in morphea is poorly understood. We performed a cross-sectional study of the Morphea in Adults and Children Cohort in order to describe and analyze the patient characteristics, pain severity, quality of life, and disease activity and damage of patients with significant pain symptoms. We queried the Cohort for patients who had consistently reported pain across multiple study visits. We excluded patients with pain attributed to other inflammatory processes. Eighteen patients met inclusion criteria and were analyzed. Patients were 88% female, 61% Caucasian, with a median age of 63 years. The median pain severity was 7/10 and pain frequency in the past 4 weeks was 75% of the time. No correlation was found between morphea subtype and pain severity ( $p=0.12$ ). Patients had largely inactive morphea, with a median localized scleroderma activity index (LoSAI) of 0 (IQR: 0-4) and median damage index (LosDI) of 12 (IQR: 5-18). Pain severity did not correlate with disease activity ( $r=-0.01$ ,  $p=0.9$ ), however, the degree of pain severity showed a positive correlation with the dermatology quality of life index ( $r=0.54$ ,  $p=0.02$ ). Half of patients reported a fibromyalgia diagnosis. In conclusion, lesional and non-lesional pain are not uncommon in morphea, and tend to occur later in the disease when damage predominates over activity. This implies that treatment of pain should not be targeted toward inflammation. The presence of pain produces negative impact on life quality, making its evaluation and treatment an important part of patient care.

(1:42 PM)

### **EVALUATING RESULTS OF AN INTERFERON- $\gamma$ RELEASE ASSAY IN PATIENTS WITH AUTOIMMUNE DISEASE ON HYDROXYCHLOROQUINE**

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QuantiFERON-TB Gold In Tube (QFT-GIT) is an interferon- $\gamma$  release assay used to screen patients for tuberculosis before starting or while on immunosuppressive therapies. Clinical studies on QFT-GIT testing show an indeterminate rate of 1.7% while other studies suggest higher rates among immunosuppressed populations. Hydroxychloroquine is proposed to reduce levels of interferon- $\gamma$  and therefore may affect the results of a QFT-GIT test. The relationship of hydroxychloroquine use to QFT-GIT results has not been studied. The medical records of 119 patients with autoimmune disease enrolled in prospective longitudinal databases with QFT-GIT testing were reviewed. Patients were sorted into groups based on the presence or absence of hydroxychloroquine use within one year of QFT-GIT testing. Additionally, we evaluated the concomitant use of prednisone and disease modifying anti-rheumatic drugs (DMARDs), such as mycophenolate mofetil, methotrexate, azathioprine, and dapsone. In the study population of

119 patients, 46 were in the hydroxychloroquine group, while 73 did not use hydroxychloroquine in the year prior to testing. There were 24 indeterminate, 92 negative, and 3 positive QFT-GIT results total. The hydroxychloroquine group had significantly more indeterminate QFT-GIT results (37.0%) compared to the non-hydroxychloroquine group (9.59%) ( $p < 0.001$ ). There was no significant difference in concomitant use of prednisone or DMARDs ( $p = 0.437$ , and  $p = 0.085$ , respectively) between groups. These results reveal that patients taking hydroxychloroquine at the time of QFT-GIT testing are significantly more likely to have an indeterminate result compared to those not taking the medication. Further studies are needed to evaluate the most appropriate tuberculosis screening in patients taking hydroxychloroquine.

(1:54 PM)

## **AUTOIMMUNE DISEASES IN PATIENTS WITH CUTANEOUS LUPUS ERYTHEMATOSUS**

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Increased rates of co-existing autoimmune diseases in the systemic lupus erythematosus (SLE) population have been reported.<sup>1</sup> Little is known about the prevalence of autoimmune diseases in cutaneous lupus erythematosus (CLE) without SLE (CLE-only) patients. Thus, we conducted a cross-sectional study among patients with CLE-only enrolled in the Cutaneous Lupus Registry at University of Texas Southwestern Medical Center from November 2008 to February 2017. Patients with  $\geq 4$  out of 11 American College of Rheumatology criteria for SLE were excluded. For each patient, we collected demographic and clinical information including concomitant autoimmune disease diagnoses, which were confirmed by other subspecialty records, and listed by Hayter and Cook.<sup>2</sup> The primary and secondary outcomes were presence of  $\geq 1$  autoimmune disease and individual autoimmune diseases, respectively. Of the 129 CLE-only patients, 17.8% (23/129) had a co-existing autoimmune disease. Rheumatoid arthritis and Sjogren's syndrome (3.1% each) were the most frequent autoimmune diseases. Univariable analyses showed that CLE-only patients who were Caucasian ( $p = 0.03$ ), non-smokers ( $p = 0.008$ ), had subacute cutaneous lupus ( $p = 0.04$ ), or positive anti-nuclear antibodies (ANAs) ( $p < 0.0001$ ) were more likely to have a concomitant autoimmune disease. Based on our multivariable analyses, CLE-only patients who were Caucasian (Odds ratio (OR):3.07,  $p = 0.03$ ), had non-smoking histories (OR:3.34,  $p = 0.02$ ), or positive ANAs (OR:5.03,  $p = 0.002$ ) were significantly associated with having autoimmune diseases. Our CLE-only patient cohort showed higher prevalence of co-existing autoimmune diseases than what is reported in the general population (4.5%).<sup>2</sup> Patients with CLE-only, particularly those with Caucasian race, non-smoking history, or positive ANAs, should be monitored closely for concomitant autoimmune conditions.

(2:04 PM)

## **A MOUSE MORPHEA MODEL REVEALS CXCL9 DRIVEN PATHOGENESIS**

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Morphea, also known as localized scleroderma, is an autoimmune disease that clinically presents as single or multiple sclerotic changes in the skin. This is a chronic disorder that leads to cosmetic and functional impairment. The treatment options for morphea are limited, and pathogenesis is poorly understood with theories involving vascular and immune dysfunction that initially present as inflammatory changes. In order to better understand the pathogenesis, we performed a series of experiments to evaluate the roles of different chemokines. A previous study looking for biomarkers for morphea in humans found that CXCL9 is highly expressed in lesional skin. We used the morphea mouse model, which uses subcutaneous bleomycin injections, to induce morphea-like fibrosis in mice. We first assessed expression of the chemokines using flow cytometry on morphea REX3 mice, which report the expression of CXCL9 and CXCL10 with fluorescent proteins. In concert, we determined the extent of skin fibrosis using Sercoll collagen assay and histological evaluation of dermal thickness with trichrome stain. We found upregulation of CXCL9 and CXCL10 in dermis of mice injected with bleomycin as compared to the control. We next tested mice deficient in CXCL9, CXCL10 and CXCR3, and found that CXCL9 and CXCR3 deficient mouse strains were protected from bleomycin induced fibrosis. Overall, these experiments showed that CXCL9 plays an important role in the pathogenesis of morphea, and targeting the production or function of CXCL9 can prove to be an effective treatment strategy.

## **CASE PRESTATIONS**

(3:00 PM)

### **STIMULATOR OF INTERFERON GENES-ASSOCIATED VASCULOPATHY WITH ONSET IN INFANCY: AN EXPANDED SPECTRUM OF DISEASE AND TREATMENT WITH TOFACITINIB**

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Stimulator of interferon genes-associated vasculopathy with onset in infancy (SAVI) is a newly described autoinflammatory disease caused by autosomal dominant gain-of-function mutations in *TMEM173*. The few reported patients presented in infancy with signs and symptoms of systemic inflammation including prominent cutaneous vasculopathic changes as well as interstitial lung disease which may progress to severe pulmonary fibrosis and respiratory compromise. However, the full spectrum of disease is not well described due to the limited number of patients reported to date. Additionally, treatment options reported in the literature are limited with variable outcomes. We present a patient with a family history of early deaths attributed to "sarcoidosis." Our patient presented at age 5 with refractory polyarthritis years before developing interstitial lung disease and cutaneous vasculopathy, at which time he presented to our combined connective tissue skin disease (CTD) clinic at age 14. Whole exome sequencing revealed a *TMEM173* p.V155M mutation confirming a diagnosis of SAVI. His disease was responsive to tofacitinib with improvement of all SAVI disease manifestations. Our clinical, histologic, and genetic findings serve to expand the phenotypic spectrum of *TMEM173* mutations, demonstrate the variable presentation of SAVI in older children, and highlight tofacitinib as a viable treatment option. Given the vasculopathic skin changes in patients with SAVI, these patients may present to CTD experts as an initial point of contact. Given this, it is important for CTD experts to be aware of the full phenotypic spectrum of disease as well as possible treatment options for this rare disorder.

#### Teaching points:

- A chronic, symmetric, deforming seropositive arthritis can be the presenting manifestation of SAVI.
- Vasculopathic changes may be the presenting skin findings of SAVI, even in older children.
- Whole exome sequencing can be an important resource in the diagnosis of SAVI.
- JAK inhibition should be considered as a viable treatment option for patients with SAVI.

(3:10 PM)

### **NEW ONSET OF LUPUS NEPHRITIS DURING USTEKINUMAB THERAPY FOR PSORIASIS**

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Ustekinumab is a safe and effective therapy for moderate to severe psoriasis. Psoriasis can on occasion co-exist with systemic lupus erythematosus (SLE). We present two cases where patients treated with ustekinumab for refractory psoriasis and/or psoriatic arthritis developed new onset renal nephritis. In each case, the time lag between initiation of ustekinumab and onset of lupus nephritis was approximately 2 years. In both patients, the psoriasis responded to ustekinumab. Our index case had a prior diagnosis of SLE with minimal disease activity and thrombocytopenia as presenting feature. The second case had no prior history of SLE. Concurrent features of SLE included perniosis, acral lesions, and small vessel vasculitis. Discontinuation of ustekinumab led to improvement in vasculopathy/ vasculitis but recurrence of psoriasis. Ustekinumab therapy has rarely been associated with small vessel vasculitis and the development of autoimmune bullous disease. Ustekinumab has been used to treat cutaneous lupus erythematosus and is currently under investigation as a therapy for SLE. Based on this observation, we suggest that concurrent inhibition of IL12/IL23 may, on occasion, promote auto-antibody mediated immune disease.

(3:20)

### **A CASE OF NXP2 DERMATOMYOSITIS PRESENTING WITH DYSTROPHIC CALCINOSIS CUTIS AND PRIMARY BILIARY CIRRHOSIS**

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A woman in her 40s presented to our clinic for management of biopsy-proven calcinosis cutis. On examination, the patient had multiple firm subcutaneous nodules and plaques on the trunk and the extremities, with no other cutaneous features of dermatomyositis (DM) or systemic sclerosis (SSc). However, muscle weakness was noted on strength exam. Two years prior, the patient noted weakness and experienced an 80-pound weight loss, attributed to new-onset dysphagia and a 2-week hospitalization in an outside hospital ICU for upper gastric bleeding. No formal diagnosis was made. Despite normal muscle enzymes, our clinical suspicion for an underlying inflammatory myositis was high. An MRI of the bilateral thighs and a muscle biopsy were performed, confirming a diagnosis of DM. A myositis panel demonstrated anti-MJ/NXP2 autoantibodies, which was clinically consistent with the patient's phenotype of severe myositis, dysphagia, calcinosis cutis, and minimal skin findings. Notably, anti-mitochondrial autoantibodies (AMA), which are strongly associated with primary biliary cirrhosis (PBC), were also incidentally noted, and a subsequent liver biopsy confirmed the diagnosis of PBC. This is the first reported case of PBC in a patient with NXP2 DM. Although there is a known association between PBC and SSc, no such association exists with DM. From a dermatology standpoint, patients with NXP2 DM tend to have minimal skin findings, but often develop calcinosis cutis over time. As we know that early and aggressive treatment of DM may prevent calcinosis cutis, it is critical to recognize this variant of DM in order to prevent this complication.

### Teaching Points:

- NXP2 dermatomyositis is associated with an increased risk of calcinosis, prominent muscle involvement, dysphagia, and peripheral edema. These patients are more likely to have minimal skin disease, and less likely to be amyopathic.
- It is important to recognize this phenotype in order to treat muscle disease early and aggressively, and ideally prevent calcinosis cutis.
- There is no known association between autoimmune liver disease and inflammatory myositis. Hence, a cholestatic pattern or disproportionate elevation of LDH/AST/ALT compared to CK/aldolase should prompt evaluation for liver disease in patients with myositis as this cannot be attributed to the myositis alone.

(3:30)

### **A RARE CASE OF LOCALIZED SCLEROSIS OF THE SCALP**

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A Hispanic woman in her 80s with a reported 30-year history of systemic sclerosis with interstitial lung disease and pulmonary hypertension, stage 4 chronic kidney disease, hepatic steatosis and hypertension, presented to our connective tissue disease (CTD) clinic for evaluation and management of scarring alopecia of the vertex with a biopsy consistent with morphea. On skin examination, the patient had an extensive, sclerotic plaque with scarring alopecia on the vertex, hyperpigmentation of sun-exposed skin, and subtle malar hypertrichosis. She had no sclerodactyly and normal nailfolds. She denied a history of Raynaud's. Given our clinical suspicion of porphyria cutanea tarda (PCT), urine and plasma testing for porphyrins was performed. Results showed high levels of uroporphyrin and hepta-carboxylated porphyrin, consistent with a diagnosis of PCT. Review of the patient's records demonstrated that she initially presented in her 50s with biopsy-proven inflammatory myositis, whereas her skin changes all developed more recently. Herein, we present a rare case of alopecia porphyrinica, which can be the presenting sign of PCT and is characterized by histopathologic changes compatible with morphea or scleroderma, hence requiring porphyrin testing for diagnosis. In this case, systemic involvement by a CTD caused a delay in diagnosis of PCT. Given the fact that skin manifestations of PCT may be disfiguring and are recalcitrant to treatment, we believe it is important for clinicians to be aware of this rare manifestation of PCT, not only to treat patients early, but also to avoid misdiagnosing these patients with other sclerosing disorders of the skin.

### Teaching Points:

- Sclerodermoid changes and scarring alopecia of the vertex in PCT are uncommon, but well described. They can constitute the first cutaneous findings of the disease.
- PCT must be suspected in the presence of sclerodermoid changes in sun-exposed skin or scarring alopecia, along with abnormal liver tests.

- Skin biopsy may not be helpful in confirming the diagnosis, which is based on porphyrin levels.

(3:40)

### **UNILATERAL VOCAL CORD PARESIS IN CLASSIC DERMATOMYOSITIS**

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Proximal symmetric muscle weakness is a common manifestation of dermatomyositis, though small muscles may also be involved. Despite being well-documented in systemic lupus, vocal cord dysfunction has only been reported in severe systemic dermatomyositis<sup>a</sup>. A 23-year-old female professional vocalist with a two-year history of moderately well-controlled classic dermatomyositis on hydroxychloroquine, methotrexate, and prednisone, with skin and muscle involvement, developed worsening myalgias, arthritis, and typical rash while tapering prednisone. She noted new-onset hoarseness that was initially attributed to gastroesophageal reflux. Endoscopy demonstrated mild gastritis and a 24-hour esophageal pH probe demonstrated reflux episodes, but symptoms did not noticeably respond to ranitidine and esomeprazole therapy. Esophageal manometry did not demonstrate muscular abnormalities. Stroboscopy demonstrated left vocal cord paresis, Reinke edema attributed to laryngopharyngeal reflux, and a left vocal cord cyst. Further workup by CT scan, edrophonium stimulation, laryngeal electromyography, and nerve conduction studies were unremarkable, and the patient started speech therapy. Due to worsening muscle symptoms, prednisone was increased during workup, leading to improvement in hoarseness. Subsequent prednisone tapers and dose escalations inversely correlated with hoarseness symptoms. Dose adjustment of methotrexate, in combination with continued speech therapy, led to gradual improvement of hoarseness and stability in cutaneous and systemic symptoms. Vocal cord paresis has been described in asymptomatic professional vocalists<sup>b</sup>, but this patient's striking correlation between dermatomyositis flares with hoarseness and immunosuppressive titration is suggestive of treatment effect. While not typical, the patient's professional training may have provided exquisite sensitivity to her unilateral vocal cord paresis.

#### Teaching Point:

- Dermatomyositis may affect laryngeal muscles and cause vocal cord paresis with resultant hoarseness. Treatment of underlying dermatomyositis, in combination with speech therapy, may lead to clinical improvement.

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(3:50)

### **NECROBIOTIC XANTHOGRANULOMA MASQUERADING AS SARCOIDOSIS AND COGAN'S SYNDROME**

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We present a case of necrobiotic xanthogranuloma (NXG) masquerading as sarcoidosis and Cogan's syndrome. A 55 year-old woman initially presented with ocular symptoms, prompting a diagnosis of anterior uveitis and episcleritis, followed shortly thereafter by the development of bilateral labyrinthitis. Subsequently, she developed periocular papules and underwent biopsy at an outside institution, which showed chronic granulomatous inflammation with CD68+ histiocytes, occasional giant cells, and focal lipid dropout, thought to be "consistent with sarcoidosis". She had intermittent sensorineural hearing loss, which in the context of chronic recurrent uveitis, raised the possibility of Cogan's syndrome. A workup for systemic sarcoidosis did not show cardiac or pulmonary disease; laboratory studies were notable for a monoclonal gammopathy of undetermined significance (IgG Kappa paraprotein), prompting bone marrow biopsy, which was negative. Over the next two years, the patient was treated with numerous agents, including intralesional triamcinolone, prednisone, hydroxychloroquine, methotrexate, mycophenolate mofetil, azathioprine, and infliximab. Despite a multidrug regimen consisting of prednisone, hydroxychloroquine, azathioprine, and infliximab 10 mg/kg every four weeks, she continued to develop new facial papules with persistent ocular symptoms. On exam, the patient had numerous papules coalescing into plaques on the bilateral malar cheeks, right temple, and right upper eyelid, with a yellow-orange hue and prominent telangiectasias centrally. Given clinical concern for a xanthomatous process, she underwent repeat skin biopsy in our connective tissue disease clinic, which showed changes consistent with NXG. The patient will see hematology in follow up for a discussion of treatment options, including IVIG and alkylating agents.