1:00 PM - 1:10 PM

5126 - The Birthmark-Cancer Connection: Unraveling the complex genetic spectrum of vascular birthmarks through cancer genomics/Beth Drolet, MD

Objective 1: Utilize a highly sensitive targeted panel for somatic variant detection performed on DNA extracted from affected tissue from 57 patients

Objective 2: Perform comprehensive phenotyping of patients with a multidisciplinary expert panel utilizing central expert review

Objective 3: Correlate somatic and constitutional variants with critical disease outcomes to prognosticate, direct screening, and guide planning of future clinical trials

ABSTRACT

Children present to dermatologists for evaluation of “vascular birthmarks.” These disorders are remarkably heterogeneous ranging from simple staining of the skin to debilitating skeletal overgrowth and abnormalities of the brain and eye. Despite these severe complications clinicians lack guidelines for screening and there are no FDA-approved drugs to treat the complications. Methods: We assembled a 15-institution network to phenotype patients with vascular birthmarks. Next-generation sequencing (NGS) using a panel enriched for cancer-related pathways was performed on 58 affected tissue samples. Results: Among analyzable samples 74% harbored pathogenic or likely pathogenic variants. The variants were detected at allele frequency ranging from 1–34.9%. We identified 8 somatic variants described in the setting of cancer, but are novel in vascular birthmarks. Conclusion: By leveraging somatic variant detection technology typically applied to diagnostic cancer genomics, we have demonstrated that vascular birthmarks are caused by postzygotic, somatic activating mutations in cell signaling pathways. The application of NGS has accelerated the discovery of novel variants in genes that are known to be associated with vascular birthmarks, somatic variants in additional genes that had not previously been associated with vascular birthmarks, and candidate constitutional and acquired variants, suggesting an increased complexity to these disorders. Improved understanding of the genetic underpinnings will guide diagnostic algorithms and ultimately move the field beyond antiquated diagnostic classifications to a pragmatic genotype-based therapeutic focus.

1:10 PM - 1:20 PM

5293 - Racial/ethnic disparities in healthcare utilization and school attendance among children with atopic dermatitis: a cross-sectional analysis of the Pediatric Eczema Elective Registry/Junko Takeshita, MD, PhD

Objective 1: To identify differences in healthcare utilization for atopic dermatitis by race/ethnicity.

Objective 2: To identify differences school missed due to atopic dermatitis by race/ethnicity.

Objective 3: None

ABSTRACT

Healthcare disparities and school attendance among racial/ethnic minority children with atopic dermatitis (AD) are not well-understood. In a cross-sectional study of children with AD enrolled in the Pediatric Eczema Elective Registry (N=7,901), we aimed to examine the association between race/ethnicity and the following outcomes: 1) healthcare utilization by visit type, and 2) any school missed due to AD within 6 months of enrollment. We used multivariable logistic regression to quantify the relationship between each outcome and race/ethnicity (reference group=whites), adjusting for age, gender, household income, atopic comorbidities, and AD severity (outcome 2 only). Mediator analysis to determine whether AD severity mediated the association between race/ethnicity and healthcare utilization was also performed. Blacks (odds ratio=2.27; 95% CI=1.62-3.17) and Hispanics (3.07; 2.02-4.66) were more likely than whites to have visited the emergency room for their AD. Blacks were also less likely to have seen a dermatologist (0.63; 0.55-0.72). AD severity only partially mediated these associations. Blacks (1.34; 1.15-1.56), Hispanics (1.84; 1.48-2.28), and Asians (3.60; 2.60-4.98) were more likely to have missed school due to AD. Our results suggest disparities in specialty and urgent care for AD and greater negative impact on school attendance among racial/ethnic minorities with AD, independent of disease severity.
F072 – Late-breaking Research Forum
Clinical Studies/Pediatric
Saturday, March 4 - 1:00 PM — 3:00 PM
Room W415D
5145 - TNF inhibitors are associated with reduced risk of all-cause mortality in psoriasis/Jashin Wu, MD

Objective 1: To determine whether use of TNF inhibitors is associated with a change in all-cause mortality in patients with psoriasis

Objective 2: To determine whether use of oral agents or phototherapy is associated with a change in all-cause mortality in patients with psoriasis

Objective 3: None

ABSTRACT

Importance: Whether treatment of psoriasis skin disease reduces the risk of all-cause mortality has not yet been fully elucidated. Methods: This was a retrospective cohort study using the Kaiser Permanente Southern California (KPSC) health plan from 1-1-2004 to 12-31-2014. Patients had at least 3 ICD-9 diagnostic codes for psoriasis. Propensity score-adjusted multivariable cox regression assessed hazard ratios (HR) of all-cause mortality. Results: Of 18,154 patients included, 1,463 received TNF inhibitors for at least 2 months, 3,579 were TNF inhibitor-naive and received oral agents or phototherapy, and 13,112 were not treated with TNF inhibitors, oral therapies, or phototherapy. After adjusting for CV risk factors, the TNF inhibitor cohort had a significantly lower hazard of all-cause mortality compared with the topical cohort (HR, 0.47; 95% CI, 0.36-0.61). The oral/phototherapy cohort also had a lower hazard of all-cause mortality compared with the topical cohort (HR, 0.86; 95% CI, 0.75-0.99). Conclusions: We observed a significantly lower risk of all-cause mortality in patients with psoriasis treated with TNF inhibitors compared to topical therapy. These findings suggest that TNF inhibitors may have benefits beyond treatment of skin disease in mitigating all-cause mortality risk, however, a randomized clinical trial is needed to confirm these findings.

1:30 PM - 1:40 PM

5279 - Pharmacokinetics, Safety and Efficacy of Dupilumab in a Pediatric Population with Moderate-to-Severe Atopic Dermatitis: Results from an Open-Label Phase 2a Trial/Michael J. Cork, MD, PhD

Objective 1: To assess the pharmacokinetics of dupilumab in a pediatric population with moderate-to-severe atopic dermatitis.

Objective 2: To assess the safety of dupilumab in a pediatric population with moderate-to-severe atopic dermatitis.

Objective 3: To assess the efficacy of dupilumab in a pediatric population with moderate-to-severe atopic dermatitis.

ABSTRACT

Objective: To assess safety, efficacy, and pharmacokinetics of dupilumab in pediatric patients with moderate-to-severe atopic dermatitis (AD). Methods: This phase 2a, multicenter, open-label, ascending-dose, sequential-cohort study (NCT02407756) included adolescents (12–17 years) with moderate-to-severe AD and children (6–11 years) with severe AD, uncontrolled by topical medications. Patients received 2mg/kg or 4mg/kg single-dose subcutaneous dupilumab with 8 weeks follow-up, followed by 4 weekly 2mg/kg or 4mg/kg doses. Results: 40 adolescents/38 children (mean Eczema Area and Severity Index [EASI]±SD=31.7±16.00/35.9±17.22) were enrolled; 22.5% adolescents/16.2% children did not respond to ?1 previous systemic treatment. Single-dose AUClast estimates were 132 vs 346 mg.h/L for 2mg/kg vs 4mg/kg, respectively, consistent with target-mediated drug disposition. Dupilumab was well-tolerated; most AEs were mild, transient and un-related. At Week 12, in adolescent 2mg/4mg cohorts, baseline EASI improved by 66.4%/69.7%, peak pruritus Numerical Rating Scale [NRS] by 30.8%/37.6%; 10%/35% achieved an Investigator Global Assessment [IGA] 0–1. At Week 12 in children 2mg/4mg cohorts, baseline EASI improved by 76.2%/63.4%, peak pruritus NRS by 41.6%/39.6%; 16.7%/21.1% achieved IGA0–1. Conclusions: The dupilumab non-linear pharmacokinetic profile was consistent with adults; efficacy and safety from this study complement data from adult trials with the significant clinical benefit confirming Th2-mediated pathophysiology for AD in children.
**Objective 1:** To determine whether socioeconomic status is associated with age at initial presentation for children with infantile hemangioma

**Objective 2:** To determine which socioeconomic factors are associated with delayed age at initial presentation for infantile hemangioma

**Objective 3:** None

**ABSTRACT**

Background: Improved outcome for children with complicated infantile hemangiomas (IH) is associated with earlier subspecialist evaluation; despite this, many children present beyond the rapid proliferative phase of growth. Pediatric health disparity is a well-established phenomenon in primary care and for common dermatologic diseases, but studies exploring access to care for patients with IH and patient outcomes are lacking.

Methods: We conducted a retrospective cohort study of 862 children presenting for IH evaluation during a 3-year period at a tertiary care referral hospital and evaluated whether socioeconomic status (SES) was associated with age at presentation for IH, using generalized ordered logistic regression. Insurance provider and zip code served as a proxy for SES.

Results: Controlling for IH severity, medical assistance insurance (lower SES) was associated with later presentation for dermatologist evaluation (p=0.017). Subgroup analysis showed that having insurance providers that are members of our institution’s payor program mitigates delayed presentation, rendering the association between SES and age of presentation insignificant (p=0.489).

Conclusions: Children with IH of lower SES have delayed presentation for subspecialty care. Institution sponsored insurance mitigates against the adverse effect of lower SES. Further investigation is warranted to determine the effect of health disparities on long term outcomes in IH.

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**Objective 1:**

**Objective 2:**

**Objective 3:**

**ABSTRACT**

**Full abstract unavailable**
2:00 PM - 2:10 PM

5209 - Diacerein Orphan Drug Development for Epidermolysis Bullosa Simplex: A Phase 2/3 Randomised, Placebo-Controlled, Double-Blind Clinical Trial/Johann Bauer, MD, MBA

Objective 1: Evaluate the safety and efficacy of the small molecule diacerein for the rare skin disease EBS

Objective 2: Evaluate the pharmacokinetics of rhein in a small cohort of patients

Objective 3: None

ABSTRACT

Objectives: Clinically meaningful treatment of EBS requires significant and sustainable reduction of blister formation. Currently, there is no treatment for this rare condition. Materials & Methods: We conducted a randomized, controlled, Phase 2/3 trial of topical application of 1% diacerein cream in 17 EBS patients. A 4-week intervention phase (primary end point: reduction in blister numbers >40%) and a 3-month follow-up were conducted in 2 sequential years, with a cross-over of groups after year 1. Reaching initial blister numbers (+/-10%) was a secondary end point. Pharmacokinetic analysis of rhein was performed in 2 patients. Results: 60% of diacerein patients and 15% of placebo patients had a reduction in blister numbers >40% after 4 weeks of intervention. At the 3-month follow-up, 12.5% of diacerein patients and 67% of placebo patients reached baseline blister numbers. The highest level of rhein in urine and serum was 39.9 ng/mL and 20.1 ng/mL, respectively, which is at least 100 times lower than the level after oral administration. No adverse effects were observed. Conclusions: This trial provides evidence that the orphan drug diacerein 1% cream is highly efficacious and may provide a sustainable long-term effect on blister formation in EBS patients.

2:10 PM - 2:20 PM

5324 - Longitudinal Development of the Skin Microbiome During the Neonatal Period/Kimberly Capone, PhD

Objective 1: Describe changes in the skin microbiota throughout the neonatal period (first month)

Objective 2: Investigate longitudinal changes in the neonatal skin microbiota with use of two skin care regimens. The treatment group included a prescribed skincare regimen, while the control group continued use of existing products.

Objective 3: None

ABSTRACT

An infant experiences its first major exposure to microbes at birth. These initial microbial populations evolve over time, adapting to characteristics of each body site. Previously, we demonstrated that after one month, the skin microbiota has evolved from birth, where it’s dominated by vaginal and/or environmentally-acquired microbes, to an infant skin microbial profile. Changes in the skin microbiota throughout the neonatal period (first month) have not been well described and may provide insights into the links between establishment of skin microbiota in early life and disease. This study measured longitudinal changes in the neonatal skin microbiota with use of two skin care regimens. The treatment group included a prescribed skincare regimen, while the control group continued use of existing products. Study results demonstrate, for the first time, the skin microbiota evolved to an infant-like skin microbial profile within the first week of life. It is dominated by 75% Staphylococcus and Streptococcus species, in contrast to infants older than one month, where 50% of the skin microbiota consists of those species. The skin microbiota changes over the neonatal period, largely through increased evenness, and represents an early view of the evolution of the skin microbiome.
2:20 PM - 2:30 PM

5196 - Objective measures of psoriasis severity predict mortality: a prospective population-based cohort study/Megan H. Noe, MD, MPH

Objective 1: Determine the all-cause mortality rate for adults with psoriasis based on objective measures of disease severity

Objective 2: Use objective measures of skin severity to examine severe psoriasis as an independent risk factor for death

Objective 3: None

ABSTRACT

Psoriasis is associated with premature death; however, to our knowledge, there have been no prospective, broadly representative studies evaluating how objective body surface area (BSA) measurements impact mortality. We conducted a prospective cohort study using The Health Improvement Network, an electronic medical records database in the United Kingdom. Patients have measures of psoriasis severity prospectively reported by a general practitioner (survey response rate: 95.7%). Charlson Comorbidity Index was used to adjust for baseline health status. Among 8760 psoriasis patients and 87,600 controls, there were 125 deaths (3.35 deaths per 1000 person-years, 95% CI: 2.81-3.99) and 1188 deaths (3.24 deaths per 1000 person-years, 95% CI: 3.06-3.43), respectively. An increased risk of death was seen in those with >10% BSA (severe disease) (HR: 1.82, 95% CI: 1.26-2.64), but not in those with <2% BSA (mild disease) (HR: 0.87, 95% CI: 0.67-1.14) or in those with 3-10% BSA (moderate disease) (HR: 0.81, 95% CI: 0.59-1.12) after adjusting for age, sex, and Charlson Comorbidity Index. Traditional mortality risk factors do not fully explain the increased risk of death in psoriasis patients with > 10% BSA. Further research is necessary to determine if treatment for psoriasis will decrease the risk in those with severe disease.

2:30 PM - 2:40 PM

5328 - Risk of Developing Pyoderma Gangrenosum after Procedures in Patients with a Known History of Pyoderma Gangrenosum/Fandi Xia, AB

Objective 1: To quantify the risk of developing a new lesion of pyoderma gangrenosum (PG) following procedures in patients with a known history of PG.

Objective 2: To identify patient and procedure related risk factors for the development of PG following procedures in patients with a known history of PG.

Objective 3: To determine the timing of new PG following procedures in patients patients with a known history of PG.

ABSTRACT

The risk of developing pyoderma gangrenosum (PG) after procedures in patients with known histories of PG is unknown. We reviewed the medical records of all patients with a clinically confirmed diagnosis of PG at Brigham & Women’s Hospital and Massachusetts General Hospital from 2000-2015 and evaluated the likelihood of developing a new lesion of PG following a procedure. We identified 166 patients with PG who underwent a total of 592 procedures. 35 (5.9%) procedures led to recurrence of PG, 90% of which occurred within the first 30 days after the procedure. A multivariable mixed logistic regression model showed that, in comparison to skin biopsy, small open surgeries had an adjusted odds ratio (OR) of 17.19 (3.27, 90.34) for PG occurrence; large open surgeries an OR of 6.01 (1.76, 20.53); and Mohs surgery/skin excision an OR of 6.04 (1.71, 21.38). Patients with PG present at the time of procedure had an OR of 5.41 (2.05, 4.28) of developing a new PG lesion. Procedure location, age, gender, and medical comorbidities were not significant risk factors. These data quantify the occurrence of PG following procedures in patients with a prior history and identify risk factors for PG occurrence.
5192 - Prevalence of primary focal hyperhidrosis (PFHh) among teens 12-17 in US Population/Adelaide A. Hebert, MD

Objective 1: To obtain PFHh prevalence data in US teens age 12-17.

Objective 2: To obtain other PFHh data in US teens age 12-17.

Objective 3: None

ABSTRACT

Background: Limited data exists on the prevalence, onset and impact of PFHh among teens. Methods: A US-representative, online consumer panel of teens was surveyed*. Responses were capped at 1,000 and, after validation, 981 complete responses were used in analysis. Classification of PFHh was based on published diagnostic criteria. Results: 17.1% of teens surveyed experience excessive, uncontrollable sweating based on published diagnostic criteria. Among these, 68.6% indicated daily impairment from sweating is major or moderate. Average reported age of onset was 11 years; 27.5% reported onset ≤ 10 years. Most teens (92%) indicated they sweat from 2+ focal areas. Average reported number of focal sweating areas was 5. Conclusion: PFHh appears to be more common among teens than previously published (previous estimates 2.1% [1] and 1.6% [2]). Related daily impairment is considerable. Accurate diagnosis, effective treatment, and more study of PFHh in teen populations is needed.

2:50 PM - 3:00 PM

5124 - Implementation of a Teletriage System Improves Access to Dermatologic Care in an Underserved Clinic: A Retrospective Review/Peter Chansky, BA

Objective 1: Compare the average time to teledermatology response with the average time to next available dermatology clinic

Objective 2: Determine the number of cases that were triaged via teledermatology without the need for in-person evaluation

Objective 3: Calculate the number of appointments saved per clinic and the resulting percentage increase in access to in-person dermatologic care due to teletriage

ABSTRACT

Puentes de Salud is a multi-disciplinary clinic providing care to uninsured Latino immigrants (1). Volunteer dermatologists support one monthly clinic, however, the volume of dermatology referrals led to significant wait times and delayed care. We implemented a teletriage system using the AAD’s teledermatology smartphone application (AccessDerm) to accelerate and expand access to dermatologic care. Clinic PCPs referred patients with dermatologic concerns via teletriage prior to scheduling in-person appointments. Retrospective data was collected regarding timing, diagnosis, and management of teledermatology cases. Sixty consultations were submitted over 2.5 years. The mean time to teledermatology response was 1.4 days (SD 3.07) versus 13.4 days (SD 8.93) to the next dermatology clinic (p<0.0001). Compared to self-reported PCP plans, teledermatology altered the treatment in 57 cases (95%). In total, 42 cases (70%) were triaged via teledermatology without need for in-person evaluation. An average of 1.4 out of 8 appointments were saved per monthly clinic, allowing an 18% increase in access to in-person dermatologic care. In summary, teletriage is a means to expand access to dermatologic care in underserved populations, including uninsured immigrants. In our study, teledermatology was sufficient to triage 70% of cases, significantly reducing the time to dermatologist evaluation and increasing availability of in-person appointments.