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Novel variants, phenotype, and treatment in the management of syndromic congenital ichthyosis

Rahul Mahajan, Postgraduate Institute of Medical Education and Research; Garima Dabas, Dipankar De, PGIMER, Chandigarh; Sanjeev Handa, Rakesh Kumar, Devi Dayal, Postgraduate Institute of Medical Education and Research; Renu Suther, Inusha Panigrah

Background: Syndromic congenital ichthyoses (CIs) are genetically determined disorders of cornification that are characterized by generalized scaling along with systemic symptoms. Data on congenital syndromic ichthyosis from developing countries are scarce.

Methods: Retrospective study of congenital syndromic ichthyosis patients attending dermatology clinic in a tertiary care center from 2105-2018. We reviewed epidemiologic and comorbidities data, genotype, clinical presentations, and treatments of syndromic congenital ichthyosis patients. The genetic diagnosis was performed with the help of targeted next-generation sequencing (NGS) and results were confirmed on Sanger sequencing.

Results: Six patients with syndromic CI were diagnosed among 86 patients with CI (8.1%). Among these, three patients of Sjogren-Larrson syndrome (SLS), two patients with Netherton syndrome (NS) and one with Chanarin-Dorfman disease (CDD) were reported. Novel variants reported in 1 patient each of CDD (heterozygous missense mutation in exon 3 and second heterozygous single base-pair insertion in exon 5 of ABHD5 gene), NS (homozygous four base pair deletion in exon 26 of the SPINK5 gene), and SLS (in exon 4 of the ALDH3A2 gene). An atypical phenotype was observed in a patient with NS with associated growth hormone and adrenocorticotropic hormone deficiency but with favorable clinical response to intravenous immunoglobulin.

Conclusions: Our reports point towards the unreported pool of genetic mutations in CI from India. Novel mutations were associated with variable cutaneous and systemic involvement.

Commercial disclosure: None identified.

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Crowdfunding for the treatment of cutaneous malignancies: Trends, correlates, and money raised

Justin L. Jia, BS, Dawson Mills, Department of Dermatology, Stanford University School of Medicine, Stanford, California; Kavita Y. Sarin, MD, PhD

Patients increasingly use online crowdfunding to offset costs associated with disease treatment. Here, we investigate the use of crowdfunding for the treatment of cutaneous malignancies and analyze how characteristics of online crowdfunding campaigns correlate with funds raised. GoFundMe campaigns created before July 2019 were selected based on their inclusion of disease keywords. Several binary variables were recorded from descriptions of melanoma, basal cell carcinoma (BCC), squamous cell carcinoma (SCC), and cutaneous T-cell lymphoma (CTCL) campaigns which met these criteria. Cutaneous malignancy campaigns increased 14.4-fold in quantity over the last 6 years. Of the 676 campaigns included, 420 were for melanoma treatment (62%), 112 for BCC (17%), 75 for SCC (11%), and 69 for CTCL (10%) treatments; melanoma, BCC, SCC, and CTCL campaigns raised $5787, $4815, $4842, and $9096 on average, respectively. We identified specific features associated with higher funds raised, including reports of physicians by name, participation in clinical trials, reports of treatment location, and inclusion of family pictures. Interestingly, funds for scientifically unsupported therapies correlated with significantly higher average funds raised for melanoma ($8829 > $5771, $P < .05$). In addition, inclusion of physician name correlated with significantly increased average funds raised for CTCL ($29,374 > $7242, $P < .05$) and SCC campaigns ($10,701 > $4466, $P < .05$). The authors hope that these findings elucidate why some dermatologic crowdfunding campaigns may raise more money than others. In addition, our results raise concerns about dermatologic crowdfunding's unauthorized use of physician names and possible facilitation of access to scientifically unsupported treatments.

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Complementary and alternative medicine use in adults and children with atopic dermatitis

Eran C. Gwillim, MD, Department of Dermatology, Feinberg School of Medicine, Northwestern University; Harrison H. Lee, BA, Jonathan I. Silverberg, MD, PhD, MPH, George Washington University School of Medicine and Health Sciences

Background: Complementary and alternative medicines (CAM) are commonly used by patients with dermatologic disorders. Few studies investigated CAM use among adults and children with AD. We sought to determine the prevalence and types of CAM use in adults and children with AD.

Methods: We performed a cross-sectional online survey study to determine use of CAM in AD. Adults and caregivers of children with AD were invited to complete the survey (completion rate = 95.1%).

Results: Overall, 302 adults (n = 232 [76.8%] female) and 144 caregivers of children (n = 53 [36.8%] female) with ever history of AD completed the survey; 259 (n = 201 [77.6%] female) and 130 (n = 45 [34.6%] female) had AD within the last year. Among adults and children with AD, the three most commonly used CAM were: vitamins (n = 169 [56.0%]; 70 [48.6%]), herbal therapy (n = 106 [35.1%]; 33 [22.9%]) and homeopathy (n = 76 [25.2%]; 29 [20.1%]). Acupuncture (n = 50 [16.6%]; 8 [5.6%], naturopathy (n = 29 [9.6%]; 13 [9.0%]), and Ayurveda (n = 11 [3.6%]; 3 [2.1%]) were less commonly used. Adults with self-reported moderate (n = 36 [34.0%]) or severe (n = 49 [42.6%]) AD use more CAM in the past year than those with mild AD (n = 3 [15.0%]) ($P = .04$). Whereas, there was no significant difference of CAM use in the past year among children based on caregiver-reported AD severity (mild: n = 3 [60.0%]; moderate: n = 15 [30.0%]; severe: n = 27 [39.7%]; $P = .30$).

Conclusions: CAM are commonly used by AD patients, with differing patterns of use in adults and children. Future research is needed to elucidate the therapeutic benefit and/or adverse effects of CAM in AD patients.

Commercial disclosure: None identified.

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Pediatric dermatology e-consultations: Reduced wait times and face-to-face dermatology visits

Kira Seiger, BA, Harvard Medical School, Boston, Massachusetts; Elena B. Hawryluk, MD, PhD, Massachusetts General Hospital, Harvard Medical School; Daniela Kroshinsky, MD, MPH, Joseph C. Kvedar, MD, Shinjita Das, MD

Background: Store-and-forward teledermatology provides pediatricians with specialist guidance for managing skin disease. This study evaluates wait times and face-to-face (FTF) dermatology visit avoidance associated with a pediatric dermatology eConsult service at an urban academic medical center.

Methods: In this retrospective cohort study, electronic medical records were reviewed for patients under age 18 for whom a dermatology eConsult was completed from November 1, 2014, to December 31, 2017. Wait times for eConsult completion and initial FTF dermatology appointments were calculated and compared with usual wait times for new patient office appointments during 2016-2017. Recommendations for FTF dermatology visits and FTF visit attendance were assessed.

Results: One-hundred eighty pediatric patients with 188 unrelated skin conditions ("cases") were referred to the service. The three most common diagnoses were atopic dermatitis, benign melanocytic nevi, and acne vulgaris, comparable to other pediatric teledermatology programs [1-6]. Of 188 cases, FTF dermatology evaluation was recommended for 60 (31.9%). Actual FTF dermatology visit avoid rate was 53.7% of total cases (n = 101 for whom FTF visit was not recommended and dermatology visit did not occur within 90-days after eConsult completion). Average turnaround for eConsult completion was 1.8 calendar days (median 1). For patients with a completed eConsult, average wait time to initial FTF evaluation was 37.3 calendar days (compared with 54.1 calendar days for pediatric patients referred directly to dermatology clinic during 2016-2017).

Conclusions: Pediatric dermatology eConsults reduce wait times for initial specialist input, serve as a triage mechanism to facilitate in-office evaluation, and reduce need for FTF dermatology evaluation.

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