COSMODERM TIMES

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Dear Doctor,

Greetings from Micro Labs. We are happy to bring you "COSMODERM TIMES" which contains latest and interesting news in the field of Dermatology.

This issue contains:

1. Oral-Facial-Digital Syndrome Type 1
2. A Pilot Study of the Diagnostic Efficacy of Electrical Impedance Spectroscopy vs Dermoscopy for Pigmented Skin Lesions
3. A Comparison of Cutaneous Fungi in Atopic Dermatitis Patients and Healthy People
4. Polypoid Melanoma: Diagnostic Challenges in a Rare Melanoma Variant

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Looking forward to your valuable feedback.

Best Regards,

Medical Team, Micro Labs Limited

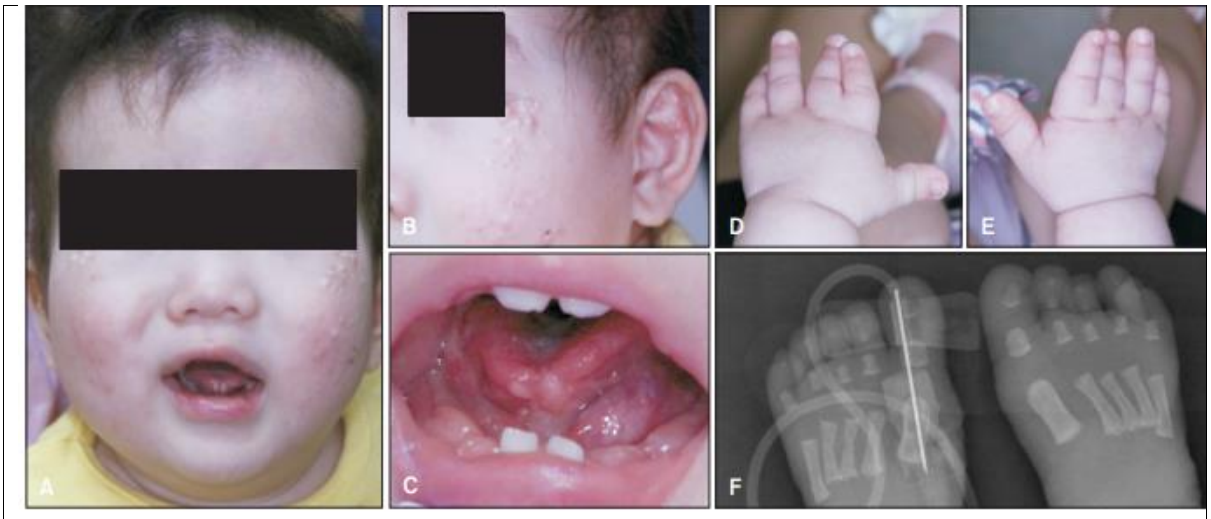
Oral-Facial-Digital Syndrome Type 1

Introduction

Oral-facial-digital syndrome (OFDS) is a combination of disorders affecting the oral cavity, face, and digits. [1] At least 16 subgroups have been identified so far, based on manner of inheritance and various abnormalities of the kidneys, limbs, brain, and other organs. [2] The most common variant, oral-facial-digital syndrome type 1 (OFD1), is inherited in an X-linked dominant manner, with mortality in men. However, more than 70% of OFD1 occurrences are sporadic. It is distinguished by congenital milia and hypotrichosis, which are absent in other variants.

Case Report:

A female 11-month-old appeared with several milia on her face and auricle that had been present from birth (Fig. 1A, B). There was also some hypotrichosis of the temporal region. A deep seated cyst comprising lamellated keratin bordered by squam epithelium with a granular layer, similar with milium, was found by histopathologic investigation of a face skin lesion.She also had congenital defects such as an incomplete cleft palate, a bifid tongue, a short frenulum, anomalous toe deformities, and clino-brachy-syndactyly of the hands. Her karyotype was typical (46, XX).



(A) Presense of multiple milia on both cheek, predominantly on the left side. (B) Presence of multiple milia in the cheek as well as auricle. Partial hypotrichosis was also noted. (C) Shows bifid tongue and short frenulum. (D, E) Clino-brachy-syndactyly of hand were presented. (F) Anomalous deformities of both toes were seen on X-ray.

OFD1 gene mutation was not discovered after chromosomal microarray and diagnostic exome sequencing. She has no family history of any abnormalities indicating a genetic illness. Furthermore, no abnormalities were discovered during eye examination or stomach and brain ultrasonography. Based on these data, an OFD1 clinical diagnosis was obtained. Her face lesions were treated with a CO2 laser and hand extraction, and have

not returned for two years. She has surgery scheduled in the plastic surgery section for intraoral and digital lesions.

Discussion:

OFDS, which was first characterised by Papillon-Léage in 1954, has been recorded in a number of instances. The condition is predicted to affect between 1/50,000 and 1/250,000 live births. However, there is no clear agreement on how to classify OFDS. Because clinical symptoms of subtypes frequently overlap, classifying a single patient can be challenging. However, OFD1 is recognised from other varieties by its distinctive milia. OFDS milia, which is seen in roughly 30% of people with OFD14, has a more widespread pattern than main congenital milia. Milia frequently persists, despite the fact that it might disappear during the third year of life and leave scars.

Oculodentodigital dysplasia (ODDD) is another disease with clinical characteristics comparable to OFD1. ODDD, on the other hand, differs from OFD1 in that it is a defined ophthalmic alteration caused by a gap junction protein alpha 1 (GJA1) gene mutation. Furthermore, various diseases linked with chromosomal microdeletion and duplication may have clinical characteristics with OFD1. [3]

As a result, substantial skin findings are critical because they aid in the early detection of OFD1. Based on the typical clinical symptoms, the patient in this case was suspected of having OFD1. In individuals with OFDS, continuous monitoring, including renal and brain assessment, should be considered.

Conclusion:

To conclude, OFD1 is important to dermatologists because its unique skin lesions might aid in differential diagnosis from other subtypes, and it necessitates intensive dermatological treatment.

OFD1 is significant to dermatologists because its unique skin lesions can aid in differential diagnosis from other subtypes, and it requires intensive dermatological treatment.

Reference

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A Pilot Study of the Diagnostic Efficacy of Electrical Impedance Spectroscopy vs Dermoscopy for Pigmented Skin Lesions

Introduction

It is critical for dermatologists to appropriately identify concerning pigmented skin lesions (PSLs) for biopsy while avoiding biopsies for benign PSLs. Early detection and removal of melanocytic neoplasms remained the most important prognostic factor for good melanoma outcomes, according to evidence. [1] Dermatologists now depend mostly on visual inspection and dermoscopic examination to guide clinical decision-making for PSLs.This approach, however, is not without flaws: false positive diagnoses result in wasteful excisions, complications, and increases in health-care expenses; of more clinical significance, false negative diagnoses might leave malignant lesions undetected, increasing patient death. [2]

Electrical impedance spectroscopy (EIS) is a non-invasive diagnostic tool used to identify melanoma by measuring the electrical impedance of skin lesions. While this technique has been demonstrated to have a high sensitivity for melanoma detection, data on its influence on clinical decision-making for pigmented skin lesions (PSLs) in comparison to other diagnostic methods is limited. The gadget generates a number ranging from 0 to 10, which corresponds to the possibility that the evaluated lesion is melanoma. EIS scores ranging from 0 to 3 have a negative predictive value of 99 percent, whereas those ranging from 4 to 10 have progressively rising positive predictive values ranging from 7% to 61%. [3] A pilot research was done to investigate how this technique compared to standard dermoscopy in terms of clinical choices for PSLs, in order to acquire a better understanding of its clinical relevance.

Methods

Dermatologists, dermatology residents, and medical students responded to an online survey that asked them about their biopsy decisions for 24 PSLs with various histological findings. Half of the lesions in each diagnostic group were displayed as a clinical picture with an accompanying dermoscopic image, whereas the other half were presented as a clinical image with the matching EIS score.

Results

Decisions made using EIS had a mean sensitivity of 75% for melanomas/severely dysplastic nevi compared to 66 percent for dermoscopy decisions. While dermatologists used EIS or dermoscopy with equal sensitivity (81 percent vs. 81 percent), residents and medical students used EIS with much better sensitivity. Respondents who reported using dermoscopy only infrequently exhibited the biggest improvement in sensitivity and specificity when using EIS over dermoscopy. The area under the curve (AUC) obtained

by EIS was 0.78, which was much bigger than the AUC produced by dermoscopy (Figure 1)

Discussion

Dermatologists frequently have difficulties in detecting melanoma, especially in patients with multiple nevi. The clinical assessment of PSLs takes into account both lesion-specific information and patient-derived melanoma risk factors. [4]

In this study, which compared the individual impacts of EIS and dermoscopy on clinical decision-making, EIS-assisted judgments were more accurate than dermoscopy-assisted decisions. The degree of training has a significant influence on the observed results. All but dermatologists reported an increase in the sensitivity of their biopsy judgments with EIS compared to dermoscopy, and all three saw an improvement in the specificity of their biopsy decisions with EIS compared to dermoscopy. While dermatologists' biopsy judgments were sensitive with both EIS and dermoscopy, this group also demonstrated the highest gain in specificity with EIS when compared to dermoscopy.

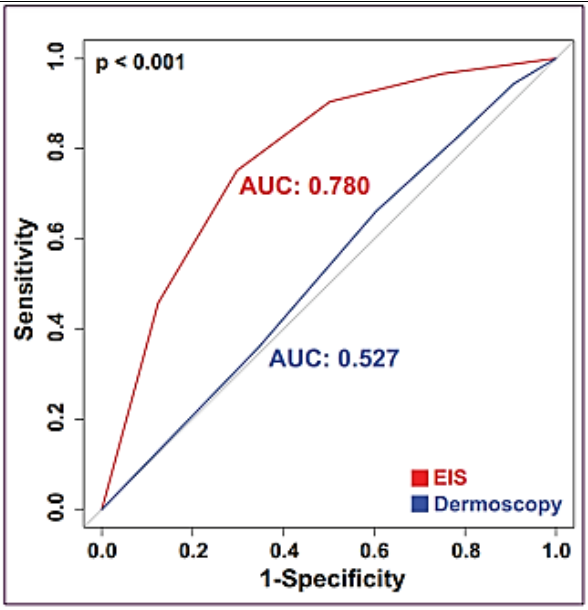

Figure 1

This suggests that, while dermatologists in this study still biopsied with superior sensitivity, they were more prone than residents and medical students to biopsy benign lesions when utilising dermoscopy, resulting in a decreased specificity of their dermoscopy-assisted biopsy judgments.

Lesions rated 1-3 were regarded unbiopsied for this analysis, whereas those scored 4- 5 were considered biopsied. In comparison to the pivotal trial's binary biopsy option, this strategy may have resulted in a higher number of lesions considered unbiopsied, reducing sensitivity, and fewer lesions biopsied, increasing specificity. However, because this strategy was used equally to dermoscopy- and EIS-assisted biopsy choices, relative accuracy was not changed. In practical application, technologies that aid in properly selecting equivocal lesions for biopsy would produce the highest yield in detecting melanomas without biopsying benign lesions.

Conclusion:

Implementing diagnostic methods that increase accuracy in the diagnosis of malignant PSL is critical for improving patient outcomes and reducing needless biopsies. In this pilot

study, dermatologists made biopsy judgments with comparable sensitivity when using EIS or dermoscopy. Those with less dermoscopy expertise (i.e., residents, medical students) displayed considerably greater sensitivity when utilising EIS over dermoscopy. Given that not all doctors are trained in dermoscopy and since this study found that EIS assists individuals who use dermoscopy rarely, EIS may complement dermoscopy by assisting a larger variety of doctors in making better PSL diagnostic judgments. This will eventually enhance patient treatment and minimise the morbidity and mortality associated with a melanoma diagnosis.

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Given that not all providers are educated in dermoscopy and that EIS favours those who use it rarely, EIS may complement dermoscopy by assisting a larger variety of clinicians in making better PSL diagnostic judgments.

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A Comparison of Cutaneous Fungi in Atopic Dermatitis Patients and Healthy People

Introduction

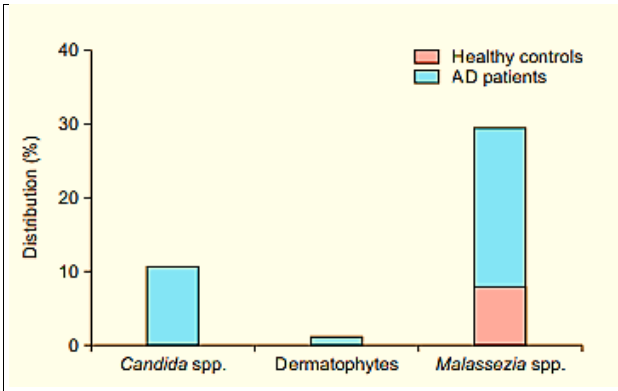
Atopic dermatitis (AD) is a recurrent inflammatory skin condition caused by a variety of circumstances. [1] AD is a difficult illness since it can be caused by a variety of causes, including genetic, environmental, and microbial. [2] Patients with Alzheimer's disease have a weaker skin barrier, making them more prone to microbial invasion. Due to a lack of interest and a limited methodological approach, extensive research on cutaneous fungal microbiota is limited in comparison to that on bacterial microbiota in Alzheimer's disease. Although enough study has not yet been conducted, numerous ideas emphasise the fungal load on disease development. For example, Alzheimer's disease patients who did not react well to standard treatment improved when given antifungal medications. Furthermore, it is known that the cutaneous fungal variety and richness are changed in Alzheimer's disease patients as compared to healthy persons. [3]

Method

Skin samples from AD outpatients and healthy people were taken and examined. Candida, dermatophytes, and Malassezia, three AD-associated fungal taxa, were studied utilising specialised primer and amplification approaches. The fungal distributions of both groups were compared after the amplicons were sequenced.

Results

A total of 211 patients and 23 healthy people were examined. Candida species was found in 10.90% (23/211) of the 211 patients (Fig. 1.), but not in 0% (0/23) of the healthy people. Candida albicans was the most often identified species in patients (5.21 percent), followed by Candida parapsilosis (3.79%). 1.42 percent (3/211) of patients tested positive for dermatophytes, whereas 0% (0/23) of healthy people tested positive. Malassezia species were found in 20.85% (44/211) of patients and 8.70% (2/23) of healthy people, respectively. Malassezia restricta was the most often identified species in the Alzheimer's disease patient group and the only species discovered in the healthy control group.



Detection frequency of Candida spp., dermatophytes, and Malassezia spp. in healthy controls and patients with atopic dermatitis (AD). Candida spp. were detected in 10.90% AD samples and 0% samples from healthy controls. Dermatophytes were detected in 1.42% AD samples and 0% samples from healthy controls. Malassezia spp. were detected most frequently in 20.85% AD samples and 8.70% samples from healthy controls.

Discussion:

While bacteria have been thoroughly investigated, the impacts of fungus have gone unnoticed. Fungi are not a direct cause of Alzheimer's disease because they are present in healthy people's skin, but they appear to contribute to the start and worsening of the disease. Local and global trials have shown a shift in the makeup of the fungal population in lesional or non-lesional skin of Alzheimer's disease patients. [4] This shift might be attributed to the changed physiological skin environment that occurs as Alzheimer's disease advances. Filaggrin mutation, for example, caused epidermal barrier failure in Alzheimer's disease patients. [5]

Malassezia is also linked to a number of human skin illnesses and disorders, with Malassezia restricta being the most common species in individuals with seborrheic dermatitis and Alzheimer's disease. M. restricta and M. globosa are considered to have a significant part in the development of Alzheimer's disease since these two Malassezia species are abundantly found in virtually all Alzheimer's patients, validating the study's conclusions. Because of their lipophilic nature, Malassezia is usually examined in sebum-rich regions such as the scalp, chest, and back. In this investigation, however, all swab samples were acquired from the antecubital region since it is the most prevalent location of AD and an easiest area to collect samples from. Furthermore, the skin microbiome exhibits phylogenic diversity based on body part.

Conclusion:

Finally, the study examined the distribution of Candida, dermatophytes, and Malassezia in the skin of Alzheimer's disease patients and healthy people. In Alzheimer's disease patients, the distribution of fungus in all three genera was changed. Malassezia species abundance and diversity were greater in AD patients than in healthy persons in this study. It is thought that the weakened skin barrier in AD allows for more diverse Malassezia colonisation than healthy skin. In turn, the changed mycobiome may activate the skin immune system, resulting in inflammation and eczema. Fungi are not a direct cause of AD based on the makeup of normal skin microflora, but an imbalance in their composition appears to be connected with AD development or aggravation.

The distribution of Candida spp., dermatophytes, and Malassezia spp. changes as AD progresses.

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Polypoid Melanoma: Diagnostic Challenges in a Rare Melanoma Variant

Introduction

Polypoid melanoma is a rare kind of melanoma that manifests as an ulcerated, exophytic nodule that might be amelanotic and affects younger patients. The initial lesion has a sluggish radial development phase but can evolve into a fast rising vertical phase within months. [1] PM may be divided into two subgroups, each of which predicts a greater death rate. Nodular melanoma is the most aggressive, with 5- and 10-year survival rates of 51.67% and 38.75%, respectively. [2] Amelanotic melanomas can develop from any histological subtype and account for 2-20% of all melanomas. A lesion's amelanotic status is determined by either a visual examination of the lesion prior to biopsy or the absence of melanin pigment on Hematoxylineosin stained sections. Pigment deficiency has also been linked to enhanced fast growth, increased Breslow thickness, and mitoses. These characteristics might result in a late biopsy and a bad outcome.

Case Presentation:

A 46-year-old Caucasian female with a history of hypertension, asthma, and hyperlipidemia came with a 5-year-old growth on the right lateral thigh. The patient reported that it had grown gently over time but had recently noticed a significant increase in size. It also got itchy, discoloured, and crusted at the lesion's tip, although it denied any bleeding or discomfort. On physical examination, a 3 cm flesh-colored exophytic and pedunculated plaque with a crusted necrotic tip extending down to half the plaque primary tumour stage of pT4b of a polypoid melanoma was discovered. For further management, the patient was promptly sent to a surgical oncologist. (Figure 1)

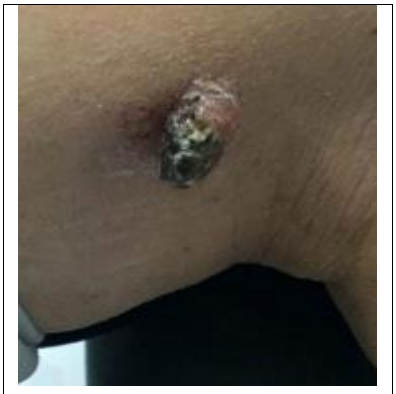

Figure 1. Polypoid melanoma. Clinical picture of an exophytic pedunculated flesh colored plaque with necrosis and ulceration at the tip extending to middle of lesion.

Discussion

Although rare melanoma variations account for fewer than 5% of all melanomas, misdiagnoses can delay treatment. The polypoid variant's surface appearance is typically described as a pedunculated or sessile lesion that can be ulcerated with or without pigmentation. Because of the morphologic variety, the physician's differential is unfocused and prone to inaccuracy. If the skin is melanotic, consider infarcted intradermal nevi, nodular basal cell carcinoma, or Merkel cell cancer. If the lesion is amelanotic, it may resemble an acrochordon, pyogenic granuloma, or vascular malformation. When an ulcer develops, the possibility of keratoacanthoma dermatofibrosarcoma protuberans enters the picture.

Melanoma is usually commonly associated with "discoloured moles" in the general population. When aberrant, non-pigmented lesions appear, they may go undiscovered for longer and cause less concern in the carrier than their pigmented counterpart. Such characteristics may lead to avoiding seeing a doctor, let alone a dermatologist. Due to the lack of colour, the ABCDE (asymmetry, border irregularity, colour, diameter, and evolution) acronym used to identify suspicious skin lesions for melanoma is impaired once in the office.

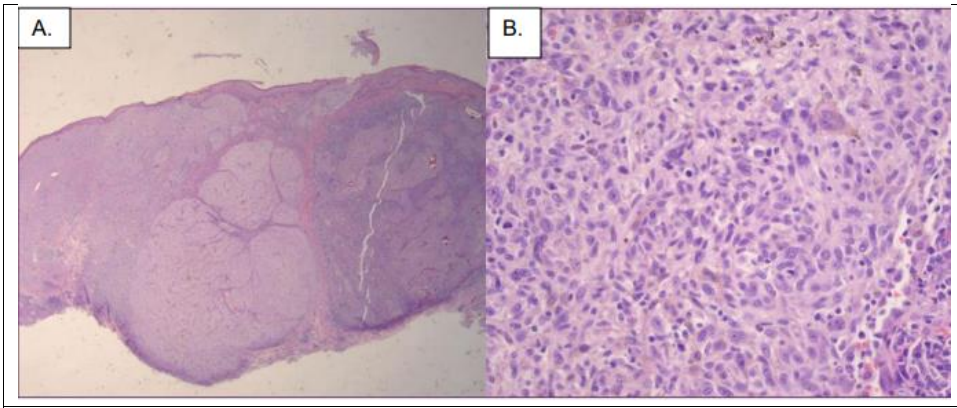

Figure 2 A) H&E 4x. Collection of melanocytes in a both epidermis and dermis with ulceration. B) H&E 40x. Grouping of atypical melanocytes with scattered mitoses present

When one of the five criteria is met, the other features of the malignancy must be at a later stage of development for visual diagnosis. Again, uncertainty exposes the disparity to possible distractors. Previous studies have found clinical misdiagnosis, with one finding that melanoma was only included in the differential 32 percent of the time for amelanotic melanomas and 94% of the time for pigmented variations. [3]

Patients diagnosed with polypoid melanoma were seen months after the initial growth appeared in this case and others found in the literature. The lesions were biopsied only after they changed to a more aggressive growth pattern, either due to the patient's dismissal or due to misdiagnosis, with an average time to diagnosis of 15.6 months. This patient's results classified her as pT4b, with a thickness greater than 4.0 mm and ulceration. With the final diagnosis, our patient can proceed to the next steps of wide local excision, sentinel lymph node biopsy, and adjuvant chemotherapy, which are both standard treatments. [4,5]

Conclusion

Unfortunately, melanomas are often overlooked when they do not appear in their normal form. A lack of patient education and self-advocacy contributed to the 5-year development of this polypoid melanoma. Increased knowledge of uncommon melanoma variations across medical disciplines, particularly in general care, may result in a lower biopsy threshold for ambiguous growths, leading to faster identification and therapy. While the ABCDE algorithm is the gold standard for melanoma diagnosis, alternative modalities, such as the "Ugly Duckling Sign," and especially for nodular melanoma, the "EFGs"

(Elevated, Firm, Growing), may be useful in the early identification of these aggressively malignant subtypes.

This malignant melanoma presents a wide range of clinical manifestations, from an amelanotic, sessile papule to a pedunculated, melanotic nodule. Its growth pattern also adds to the uncertainty of the lesion, with a gradual development stage that might spontaneously switch into fast growth. This may be difficult for all parties involved, including the patient, primary care providers, and dermatologists.

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