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Abstract N°: 117

A Rare Case of Multiple Cutaneous Leiomyomas Exhibiting Features of Reed Syndrome

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Introduction & Objectives:

Cutaneous leiomyomas are rare benign tumors originating smooth muscle, most commonly the arrector pili muscles. They appear as firm, skin-colored to reddish-brown papules or nodules, occuring singly or in clusters, often following a dermatomal or linear pattern. These lesions can be associated with symptoms such as pain or tenderness (1,2). They may appear sporadically or be linked to Hereditary Leiomyomatosis and Renal Cell Carcinoma (HLRCC), an autosomal dominant syndrome caused by mutations in the fumarate hydratase (FH) gene(3). HLRCC is significant due to its association with aggressive malignancies, including type 2 papillary renal cell carcinoma, uterine leiomyosarcoma, and other smooth muscle neoplasms(4,5). The diagnostic and management challenges posed by cutaneous leiomyomas highlight the need for careful clinical evaluation, histopathological confirmation, and genetic testing where appropriate. This case report underscores the unique presentation of this rare dermatologic condition.

Materials & Methods:

A 46-year-old woman presented with a cluster of papules and nodules on her back. The lesions first appeared 7 years ago, doubling in number over time with slight pain and itching. On examination, multiple hyperpigmented, smooth dermal papules and nodules of varying sizes were observed. The lesions were mostly asymptomatic but occasionally caused slight pain. The patient had a history of uterine fibroids in her 30s for which she underwent myomectomy. Family history revealed that her 2 sisters had uterine leiomyomatosis. A skin biopsy confirmed the diagnosis of cutaneous leiomyoma.

Results:

Histopathological examination showed a nodular lesion with smooth muscle cells arranged in interlacing fascicles in the dermis. The cells show cigar-shaped nuclei with indistinct nucleoli and eosinophilic cytoplasm. Lymphoid aggregates are noted with no mitotic activity, atypia or necrosis. The overlying epidermis shows mild papillomatosis and pigment incontinence (figure 1).

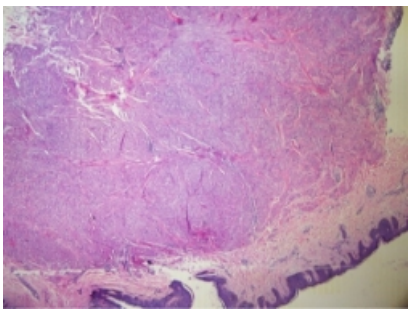

figure 1: haematoxylin and eosin (H&E) stain, x4 magnification, nodular lesion with smooth muscle bundles in the dermis.

Histopathological examination showed positive for smooth muscle active (SMA) and negative for S100. (figure 2)

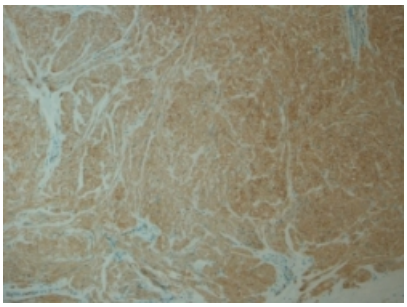

Figure 2: immunohistochemistry (IHC) stain, x10 magnification, SMA positive muscle bundles.

Given the patient’s clinical presentation, surgical and family history, and histopathological findings, a diagnosis of multiple cutaneous and uterine leiomyomatosis, typical of Reed’s syndrome, was established. The patient was referred for genetic testing to confirm the diagnosis of Reed’s syndrome, focusing on mutations in the fumarate hydratase (FH) gene.

Conclusion:

This case underscores the significance of recognizing cutaneous leiomyomas as a rare but clinically relevant entity. Their potential link to HLRCC requires a multidisciplinary approach to diagnosis and management, including genetic counseling and regular surveillance for systemic malignancies. Maintaining a high suspicion and relying on histopathological confirmation are essential in distinguishing leiomyomas from other dermatological conditions. Sharing individual cases adds to the collective knowledge, enhances diagnostic accuracy and management strategies. This case also reminds clinicians that dermatologic findings can reveal underlying systemic disease, emphasizing the value of a holistic approach to patient care.

Abstract N°: 224

Sustained Remission in Hailey-Hailey Disease: A Case Report on the Efficacy of Tetracyclines after Years of Treatment Resistance

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Introduction & Objectives: Hailey-Hailey disease (HHD) is a rare autosomal dominant genodermatosis, characterized by recurrent, ruptured vesicles and eroded plaques predominantly affecting intertriginous areas such as the axillae, groins, and inframammary regions. The condition typically manifests in adulthood and is often associated with chronicity and treatment resistance. We report a case of a 40-year-old female with a five-year history of refractory HHD who achieved sustained remission following the administration of systemic tetracyclines.

Materials & Methods: This case study details the clinical progression of a 40-year-old female diagnosed with Hailey-Hailey disease. Comprehensive clinical history was gathered, encompassing familial predispositions, presenting symptoms, and prior medical evaluations. Physical examinations concentrated on the cutaneous manifestations, which initially presented as erosive lesions in the inframammary region before extending to the neck, axillary and groin areas. Diagnostic evaluations included punch biopsy with corresponding histopathological analysis.

Results: Histological examination via punch biopsy (3.5 mm) revealed acantholysis, intraepidermal vesicles, and perivascular lymphocytic infiltrates, consistent with the diagnosis of HHD. No fungal elements were identified in periodic acid-Schiff (PAS) staining. Clinically, the patient reported severe pruritus, burning sensations, and significant discomfort. Notably, there was no documented family history of dermatological disorders. The patient had undergone treatment with mid-potent topical corticosteroids for over five years, yielding no therapeutic benefit. Preventive measures were implemented to avoid disease exacerbation. The patient was initiated on Doxycycline (200 mg/day) for one week, followed by a maintenance dose of 100 mg/day for one month, in conjunction with topical benzoyl peroxide wash and mupirocin 2% cream. After this regimen, a maintenance dose of 50 mg/day was administered for an additional month. To address the gastrointestinal side effects commonly associated with tetracycline therapy, the patient was concurrently administered probiotics and advised to incorporate a diet rich in fiber. Significant clinical improvement was observed within two weeks, with a marked reduction in inflammation, and absence of active borders. Over time, the patient reported a decrease in pruritus and burning sensations, leading to enhanced quality of life. Notably, no disease recurrence was observed throughout a 24-month follow-up period.

Conclusion: This case underscores the effectiveness of systemic tetracyclines, administered in a tapering regimen, in achieving sustained remission in Hailey-Hailey disease, without precipitating a relapse of symptoms. The synergistic use of oral Doxycycline alongside topical therapies facilitated long-term symptom control in a patient previously resistant to topical corticosteroid treatment.

Abstract N°: 319

A Case of Striate Palmoplantar Keratoderma Responding to Systemic Isotretinoin

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Introduction & Objectives:

Palmoplantar keratoderma (PPK) refers to a group of cornification disorders that exhibit extensive phenotypic heterogeneity. In particular, striate palmoplantar keratoderma (SPPK) has been associated with mutations in more than 20 different genes, notably those involved in keratinocyte function and skin barrier integrity. Among these genes are desmoglein 1 (*DSG1*), desmoplakin (*DSP*), and keratin 1 (*KRT1*). SPPK can severely affect quality of life by causing discomfort and functional limitations. Clinical evaluation, often supported by genetic testing to identify pathogenic variants, is key to confirming a diagnosis of SPPK. Here, we present a case of a 15-year-old female patient with genetically confirmed SPPK who exhibited a positive clinical response to isotretinoin therapy.

Materials & Methods:

The patient's outpatient record, history, and laboratory data were evaluated.

Results:

The patient first presented with eczema on her legs at 2–3 years old. Over time, her eczema regressed; however, she subsequently developed hyperkeratotic lesions on her hands and feet. There was no family history of similar conditions, and she had no known comorbidities. The patient was diagnosed with SPPK, characterized by bilateral linear, hyperkeratotic plaques on her soles. Genetic testing revealed a heterozygous variant in the *DSG1* gene, specifically c.655C>T p.(Arg219*), which was classified as likely pathogenic. This result is consistent with a diagnosis of SPPK. Before the current treatment, the patient used topical moisturizers and salicylic acid creams, which showed no significant improvement. The patient was started on isotretinoin treatment at 0.3 mg/kg, and by the fourth month, after the patient successfully tolerated the treatment, the dose was increased to 1 mg/kg. From 1 mg/kg onwards, the patient showed a successful response to the treatment, and it was observed that the thickness of the hyperkeratotic plaques significantly decreased.

Conclusion:

This case demonstrates that isotretinoin therapy may be effective in a rare *DSG1*-related SPPK. Retinoids are recognized as an important treatment option for PPK, although not all subtypes respond. Our case report suggests that isotretinoin could be considered for SPPK, which can pose significant diagnostic and therapeutic challenges. Nevertheless, further studies are required to confirm these findings.

Abstract N°: 338

Значение микробиома кожи при ихтиозе

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Introduction & Objectives: Ichthyosis (ichthyosis; Greek Ichthyos fish + osis; synonym: diffuse keratosis, sauriasis) is a hereditary dermatosis, characterized by diffuse keratinization disorder of the hyperkeratosis type, formation of fish scales on the skin. The development of ichthyosis is based on genetic heterogeneity, localized on different chromosomes.

Recently, in dermatological practice, special attention has been paid to the state of the skin microbiome in the clinical course of many dermatoses.

The aim of the study: to assess the state of the skin microbiome in patients with ichthyosis.

Materials & Methods: We examined 34 patients with ichthyosis aged from 2 to 18 years. Among them, 21 were boys and 9 were girls. All patients underwent clinical (dermatoscopy), microbiological and statistical research methods.

Results: In the group of patients with ichthyosis, lamillary ichthyosis was noted in all 34 patients by clinical form. Microbiological studies of the skin of lesions in 34 children with AD revealed the growth of opportunistic microorganisms of staphylococcal flora in 33, which amounted to 97.1%. According to species identification, st. epidermidis was most often cultured - 55.8% (19 of 34), st. aureus - 11.7% (4), (10.8%), st. saprophyticus - 8.8% (3) and s. Hemoliticus - 5.8% (2), respectively. The degree of colonization of st.epidermidis averaged - 37.8+ 1.8 CFU/cm2 and st.aureus - 82.5+ 5.5 CFU/cm2, which was 7.4 times higher than the indicators of healthy individuals. (P <0.05).

Conclusion: thus, the analysis of microbiological studies showed that patients with ichthyosis have a syndrome of increased colonization of staphylococcal flora, which contributes to the aggravation of the clinical course of dermatosis.

Abstract N°: 342

A Family with Cowden Syndrome: Unusual Coexistence with Epilepsy and Autism

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Introduction & Objectives:

Multiple hamartoma syndrome, or PTEN Hamartoma Tumor Syndrome (PHTS), is a rare autosomal dominantly inherited condition caused by mutations in the PTEN tumor suppressor gene, characterized by abnormal cell growth in normal tissues. PHTS includes phenotypes such as Cowden syndrome, Bannayan-Riley-Ruvalcaba syndrome, Lhermitte-Duclos disease, and Proteus (-like) syndromes. Among these, Cowden syndrome uniquely carries a significantly increased malignancy risk, emphasizing the need for early diagnosis. This report presents two cases of Cowden syndrome, a mother and her daughter, with typical clinical and genetic features. Notably, the coexistence of epilepsy and autism in the pediatric patient is an exceptionally rare occurrence, with only one similar case reported in the literature.

Materials & Methods:

Comprehensive clinical data were collected, including dermatological examinations, genetic analyses, and medical histories. Imaging and laboratory tests were conducted to assess systemic involvement. Genetic testing, performed via Whole Exome Sequencing (WES), confirmed PTEN mutations in both cases.

Results:

Case 1:

A 41-year-old woman presented with puffy lips, noted since childhood. Findings included thyroid nodules, intestinal hamartomatous polyps, and breast cysts. Dermatologic examination revealed oral papillomatosis with 1-3 mm smooth, white-pink papules forming a cobblestone pattern on the lips, gingiva, palate, and tongue. Skin-colored papules were also observed on the face and ears.

Case 2:

An 8-year-old girl with epilepsy and autism presented with diffuse skin-colored papules on the trunk and extremities. Macrocephaly, pectus excavatum, scoliosis and infantile hypotonia were noted. Dermatologic examination revealed widespread smooth papules.

Conclusion:

Cowden syndrome is characterized by hamartomas and neoplasms affecting the skin, mucosa, thyroid, breasts, and gastrointestinal/genitourinary tracts. Dermatologic findings are often the earliest and most diagnostic features, with malignancy risks of ~50% in women and ~25% in men.

The epilepsy observed in the girl presented here is infrequently associated with Cowden syndrome. The association of autism and epilepsy accompanying Cowden syndrome has been reported only in one case in the literature. This is likely because the reported Cowden syndrome cases were quite young (16 months to 18 years), and characteristic features like skin lesions and hamartomas often develop later.

In contrast to the previously documented case, a male patient presenting with a de novo PTEN mutation and no family

history of Cowden syndrome, this case involves a familial presentation affecting a mother and daughter. Additionally, PTEN mutations and autism are more frequently reported in males, making our case noteworthy as a rare female presentation.

In conclusion, it is important to report all new cases affected by PTEN gene mutation or Cowden syndrome and to detail the dermatological findings. Early recognition of dermatological findings not only aids in the timely diagnosis of Cowden syndrome but also plays a critical role in reducing the risk of life-threatening malignancies through proactive surveillance and intervention.

Abstract N°: 375

Gorlin Syndrome - A therapeutic challange when targeted therapy is not available

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Introduction & Objectives:

Gorlin-Goltz Syndrome or Nevoid basal cell nevus syndrome is a rare AD genodermatosis caused by the mutation of tumor suppressor genes SUFU or PTCH1, leading to the development of multiple basal cell carcinomas and other benign and malign tumors and more than 100 clinical abnormalities. The basal cell carcinomas might have a more aggressive course but are histologically identical to the sporadic ones. The other features include cardiac and ovarian fibromas, skeletal abnormalities, palmo-plantar pits, medulloblastomas, lymphomesenteric cysts.

Materials & Methods:

Here, we present the case of a 48 years-old male patient, phototype III Fitzpatrick, who presented at our clinic for multiple skin tumors (>30) of various sizes, located on the face, scalp, auricular and periauricular area. The tumors consisted of papules and nodules, varied in color from black-brown to pearly shiny-white, having smooth surface but also crusts.

Anamnestically, the patient confirmed having been diagnosed with odontogenic keratocyst and falx cerebri calcification. Clinical examination revealed macrocephaly, frontal and temporo-parietal bossing, moderate hypertelorism, low-set ears, surgical scar due to cleft lip and palate. Dermoscopy helped us identify typical structures of basal cell carcinoma in the above-mentioned lesions: arborising vessels, translucent margins, central ulceration, hematic crusts, ovoid nests, leaf-like areas, spoke-wheel areas, peppering, blue-gray dots and clods, absence of pigmented network.

Results:

For the nodular tumors, we decided that surgery was the optimal treatment for the patient. We mention that Mohs surgery is not reimbursed in our country. Therefore, excisional biopsies of 5 lesions were performed. The pathology results revealed typical basal cell carcinoma proliferation. None of them presented perineural invasion and all had clear microscopical margins.

Conclusion:

Integrating the clinical and paraclinical findings, the diagnosis of Nevoid basal cell carcinoma syndrome (Gorlin Syndrome) was made. Our patient presented 3 major criteria and at least 3 minor criteria. Confirmation of the diagnosis through genetic testing for PTCH1 and SUFU mutations was not accessible for the patient.

Although recommended, patients with Gorlin syndrome are currently not eligible for reimbursement of systemic hedgehog pathway inhibitors, such as Vismodegib or Sonidegib, in Romania.

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Abstract N°: 392

Segmental Neurofibromatosis Type V: A Rare Genodermatosis

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Introduction & Objectives:

Neurofibromatosis is a heterogeneous group of diseases primarily affecting the nervous system and skin. It is a relatively common genodermatosis characterized by the presence of neurofibromas, café-au-lait spots, and certain neurological and ophthalmological symptoms. Segmental neurofibromatosis type V, also known as localized or mosaic neurofibromatosis, is a rare variant of the disease characterized by skin lesions limited to a circumscribed segment of the body. We report a case of localized neurofibromatosis in the thoracolumbar region.

Materials & Methods:

A 14-year-old female patient, with no family history of neurofibromatosis, presented with multiple brownish macular lesions of varying sizes resembling café-au-lait spots, located on the right thoraco-abdominal area in a unilateral dermatomal distribution, evolving for 2 years. Dermatological examination revealed two firm, painless, flesh-colored nodular lesions measuring 2 cm in diameter on the abdominal area.

The rest of the clinical examination was unremarkable, particularly the neurological and osteoarticular exams.

Ophthalmological examination did not reveal Lisch nodules.

A biopsy of one of the nodules showed a non-encapsulated, circumscribed neurofibroma. The cerebral magnetic resonance imaging and thoraco-abdomino-pelvic computed tomography scans showed no significant findings.

Results:

Segmental neurofibromatosis type V is a very rare genodermatosis, with an incidence 10 to 20 times lower than that of neurofibromatosis. The first case was described by Gammel in 1932. It is associated with a post-zygotic mutation of the NF1 gene, leading to somatic mosaicism. This mutation occurs late in embryonic development, resulting in a localized disease. The disease is twice as frequent in women, with a bimodal peak of occurrence between 10-30 years and 50-70 years. Classic features of segmental neurofibromatosis include café-au-lait spots, freckling, and/or neurofibromas in a dermatomal distribution or following the lines of Blaschko, without family history or systemic involvement. The disease can be associated with systemic involvement and malignant tumors. Malignant tumors associated with segmental neurofibromatosis include tumors of peripheral nerve sheaths, malignant melanoma, breast cancer, colon cancer, gastric cancer, lung cancer, and Hodgkin's lymphoma. The most common tumors are derived from neural crest cells: malignant peripheral nerve sheath tumors and malignant melanoma. Although there are no specific guidelines for managing these patients, an accurate diagnosis is crucial to detect potential systemic complications.

Conclusion:

Segmental neurofibromatosis appears to be more widespread, as most patients are asymptomatic and seek consultation primarily for cosmetic reasons. Long-term follow-up for complications and malignant tumors, as in patients with generalized neurofibromatosis, is always recommended.

Abstract N°: 404

multiple familial trichoepithelioma associated with lentigo maligna melanoma

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Introduction:

Trichoepitheliomas (TE) are benign skin tumors of the pilosebaceous apocrine unit with follicular differentiation. Multiple familial trichoepithelioma (MFT) is a rare autosomal dominant dermatosis (1) linked to mutations in the CYLD gene (2). It is characterized by numerous trichoepitheliomas of the face which can transform into carcinomas.

We report a case of MFT associated with lentigo maligna melanoma, which to our knowledge, is the first case described.

Observation:

A 52 years old female patient, with no particular medical history, was referred to us for a pigmented lesion on the left cheek. The excisional biopsy came back in favor of lentigo maligna melanoma. She then benefited from a revision with exeresis margins according to Breslow index. On skin examination we find firm flesh-colored papular lesions with a central facial topography: base of the nose, nasolabial folds, upper lips and chin that have been evolving since the age of 15. The rest of clinical examination was normal. Her 28-year-old daughter had similar lesions evolving since childhood. No other member of the family had similar lesions. An excisional biopsy of a facial papule was performed on the mother. Histopathology report concluded to trichoepithelioma. Given the clinical, histological appearance and the family context, the diagnosis of multiple familial trichoepithelioma (MFT) was confirmed. Our patient benefited from CO2 laser destruction sessions, however, with recurrence of the lesions.

Discussion:

MFT constitute a rare autosomal dominant genodermatosis, due to mutations in the CYLD gene, located on chromosome 16q12-13. (2) This gene promotes the proliferation and differentiation of germ cells of pilosebaceous units, allowing the development of trichoepitheliomas. (3) MFT affects more women due to its low genetic expressivity and penetrance in men. (4,5) It manifests in childhood or adolescence, with the appearance of papulonodular, flesh-colored, shiny lesions, symmetrically affecting the central region of the face (as reported in our observation), however, TE can also affect the scalp, cervical region and upper chest. The evolution of MFT is marked by the multiplication of lesions (2), and although rare, by malignant transformation of TE into basal cell carcinoma and trichoblastic carcinoma which would be due to the mutation of the CYLD gene (1) (6) justifying regular monitoring of patients.

The association of MFT with lentigo maligna melanoma has not been reported to our knowledge. Our observation would be the first case to be described.

MFT presents a major aesthetic problem and the management of TE remains a challenge, particularly in patients with multiple facial lesions as observed in our patient. Various treatment modalities have been proposed: surgery, electrodessication, dermoabrasion, CO2 laser, Erbium-YAG laser and Imiquimod. (7)

Conclusion:

Transformation of trichoepitheliomas into carcinomas is rare but known. Our observation is distinguished by the association of multiple familial trichoepithelioma with lentigo maligna melanoma, which could probably be the first case to be described.

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Abstract N°: 503

Pachyonychia Congenita Type 3: A Familial Case with KRT6A Gene Mutation Responding to Acitretin

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Introduction & Objectives:

Pachyonychia congenita (PC) is a rare, autosomal dominant genodermatosis most commonly caused by pathogenic variants in keratin genes. Characteristic clinical findings include hypertrophic nail dystrophy, painful palmoplantar keratoderma, oral leukokeratosis, and steatocystoma multiplex. Type 3 PC often presents with these features and displays a familial inheritance pattern. In this report, we present a 17-year-old girl with lifelong nail anomalies, palmoplantar hyperkeratosis, dental defects and steatocystoma multiplex who was diagnosed as pachonychia congenita type 3 and showed clinical improvement with acitretin treatment.

Materials & Methods:

A 17-year-old female was referred for congenital nail dystrophy and painful thickening of the palms and soles. Examination revealed dental irregularities, oral leukokeratosis, palmoplantar keratoderma, numerous trunk cysts consistent with steatocystoma multiplex, and subungual hyperkeratosis of the fingernails and toenails. Family history disclosed similar findings in her older sister, with parental consanguinity. Neither the patient nor her sister had been previously diagnosed. Based on clinical suspicion of pachyonychia congenita, genetic testing exon analysis revealed a heterozygous mutation (c.516_518del) in the KRT6A gene, confirming the diagnosis of pachyonychia congenital type 3. Therapy was initiated with oral acitretin at 25 mg/day and topical 40% urea for nail and plantar lesions. Regular safety labs were conducted, and follow-up appointments were scheduled every three months.

Results:

Significant improvement in plantar keratoderma was observed over several months, with reduced pain and hyperkeratosis. Oral leukokeratosis showed partial regression, and nail dystrophy gradually improved. The cysts in the trunk also showed significant improvement and reduction in size. No major adverse effects or lab abnormalities emerged during treatment. The patient continues under three-month follow-up, maintaining clinical benefit and stable disease features.

Conclusion:

This familial presentation of pachyonychia congenita Type 3 highlights the importance of clinical recognition, early genetic testing, and a comprehensive therapeutic strategy. Acitretin proved both effective and safe in mitigating key aspects of the disease, including nail dystrophy and plantar keratoderma, reinforcing the value of systemic retinoid therapy for symptom control in pachyonychia congenita.

EADV SYMPOSIUM PRAGUE 22-24 MAY 2025

Abstract N°: 578

H Syndrome Presenting with Bilateral Cheek Enlargement and a SLC29A3 Gene Variant

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H Syndrome Presenting with Bilateral Cheek Enlargement and a *SLC29A3* Gene Variant

Introduction: H syndrome is a rare autosomal recessive genodermatosis caused by *SLC29A3* gene mutations. It was first coined in 2008 since most of the clinical features observed in affected patients start with letter 'H'. Here, we present a case of H syndrome with bilateral cheek enlargement, and a novel variant of *SLC29A3* gene mutation in an adolescent female.

Case Report: An adolescent female, born to a non-consanguineous marriage, presented with diffuse, gradual, painless, bilateral cheek hypertrophy with overlying telangiectasias. There were neither palpable intraoral masses nor lymphadenopathies. She was severely stunted, wasted, and had amenorrhea with delayed development of secondary sexual characteristics. She had symmetric, diffuse, fairly defined, indurated brown plaques and patches with overlying black coarse terminal hairs, associated with warmth and tenderness on the hypogastric and sacral area, bilateral anterior, medial, and lateral thighs and legs, with sparing of the knees. The upper extremities, chest, palms and soles were unaffected. Her laboratories showed microcytic hypochromic anemia, normal erythrocyte sedimentation rate and fasting blood sugar. Endocrine work-up showed follicle-stimulating hormone and luteinizing hormone on the low-normal levels while insulin-like growth factor 1 was low. Pure tone audiometry revealed mild sensorineural hearing loss on the left ear while thoracic CT scan with IV contrast showed bilateral pleural and pericardial thickening with mild hepatosplenomegaly. Histopathology of the hyperpigmented plaques on the lower extremities and from a core needle biopsy on the right cheek revealed fibrohistiocytic proliferation with panniculitis, staining positive for CD68 and S100 but negative for CD1a. Genetic testing showed a novel homozygous frameshift mutation variant in *SLC29A3*, supportive of the diagnosis of H syndrome. The patient is being managed with nonsteroidal antiinflammatory drugs, estradiol, and Neodymium-doped Yttrium Aluminum Garnet laser for hair removal.

Conclusion: H syndrome is a rare disorder, and as of this writing, this may be the first reported case in Filipino ethnicity. It is important to consider this condition in patients presenting with hyperpigmentation and hypertrichosis in the setting of multisystemic involvement. Genetic testing is the gold standard of diagnosis but mutations involving unique exons and codons in the *SLC29A3* gene may result in a different phenotype. A multidisciplinary care and regular follow-up must be instituted to better manage symptoms and complications.

EADV SYMPOSIUM PRAGUE 22-24 MAY 2025

Abstract N°: 806

Birt-Hogg-Dubé Syndrome: Exploring a Rare and Complex Familial Genodermatosis

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Introduction & Objectives:

Birt-Hogg-Dubé syndrome (BHDS) is a rare autosomal dominant genodermatosis, with approximately 600 families worldwide reported to be affected. It is characterized by the development of hundreds of small, benign fibrofolliculomas, primarily on the face and upper torso. BHDS predisposes individuals to pneumothorax, pulmonary cysts, and renal tumors. The objective of this study is to analyze the clinical presentation and genetic findings in a patient diagnosed with BHDS.

Materials & Methods:

We performed an analysis of the medical history of a 44-year-old woman diagnosed with Birt-Hogg-Dubé syndrome.

Results: The 44-year-old patient was referred from the Department of Pulmonology to the Dermatology Outpatient Clinic. She had a history of emphysema and multiple episodes of spontaneous pneumothorax, with the first occurrence at age 29. Dermatological examination revealed multiple small, yellowish-white papules on the face, cleavage, and upper back. The lesions had appeared during adolescence and persisted for many years. Genetic testing identified a pathogenic mutation, C1285dupC, in the FLCN gene, confirming the diagnosis of BHDS. The patient's mother, who had a history of emphysema of unknown origin and a renal tumor, also underwent genetic testing, which revealed the same mutation as in her daughter. Both the patient and her mother continue to receive regular care from the Dermatology and Pulmonology Clinics.

Conclusion:

Birt-Hogg-Dubé syndrome is a rare genetic disorder characterized by skin lesions (fibrofolliculomas), pulmonary symptoms (pneumothorax, emphysema), and an increased risk of renal cancer. As an autosomal dominant condition, genetic testing for other family members is recommended. Interdisciplinary care is essential for the proper diagnosis and management of BHDS.

Abstract N°: 821

Cracking the Genetic Code of Epidermolysis Bullosa with Bioinformatics

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Introduction & Objectives:

Epidermolysis Bullosa (EB) is a group of rare, inherited skin disorders characterized by skin fragility and blistering due to mutations in genes encoding structural proteins essential for skin integrity. Despite significant advances in understanding the clinical manifestations of EB, identifying disease-causing mutations and developing targeted therapies remain challenging due to the complexity of genetic variations. This study aims to explore the role of bioinformatics in analyzing genetic data from EB patients, with a focus on whole-genome sequencing (WGS) and whole-exome sequencing (WES), variant calling and annotation, transcriptomic analysis through RNA sequencing, and protein-structure modeling. The objective is to assess how bioinformatics can improve diagnostic accuracy, facilitate personalized treatment strategies, and advance therapeutic development for EB.

Materials & Methods: A comprehensive review of bioinformatics tools and methodologies was conducted to evaluate their application in EB research. Whole-genome sequencing (WGS) and whole-exome sequencing (WES) were analyzed for their effectiveness in identifying pathogenic mutations. Variant calling and annotation techniques were examined to determine their role in prioritizing disease-associated genetic variants. RNA sequencing data were reviewed to assess transcriptomic changes linked to EB, while protein-structure modeling approaches were explored to understand the functional impact of mutations on skin integrity. Additionally, clinical applications such as the integration of bioinformatics into personalized treatment strategies and gene-editing approaches were analyzed based on recent advancements in the field.

Results: Bioinformatics approaches have significantly enhanced the ability to identify novel EB-associated mutations and predict their pathogenicity. WGS and WES have been instrumental in detecting genetic variations that contribute to disease heterogeneity. Variant annotation tools have improved mutation classification, aiding in more precise genetic counseling. Transcriptomic analyses have revealed gene expression alterations that provide insight into disease mechanisms. Protein-structure modeling has helped elucidate the functional consequences of specific mutations, offering a deeper understanding of their impact on skin integrity. Furthermore, bioinformatics-driven strategies have contributed to early diagnosis, personalized therapeutic planning, and the exploration of gene-editing technologies as potential future treatments.

Conclusion: Bioinformatics plays a crucial role in advancing the understanding of EB by facilitating genetic analysis, improving diagnostic accuracy, and guiding therapeutic interventions. While challenges such as data complexity and limited genetic databases remain, the integration of artificial intelligence, expanded genomic resources, and clinical decision support tools holds great promise for the future. Continued advancements in bioinformatics will further enhance EB research and clinical management, ultimately improving outcomes for patients with this debilitating condition.

Abstract N°: 823

The ABCA12 Gene in Harlequin Ichthyosis: Insights from Bioinformatics and Clinical Research

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Introduction & Objectives:

Harlequin Ichthyosis (HI) is a rare, severe congenital skin disorder that is often life-threatening, characterized by thick, rigid skin and large, diamond-shaped plates. The condition arises due to mutations in the ABCA12 gene, which encodes a crucial lipid transporter protein essential for skin barrier function. Despite significant progress in understanding the genetic basis of HI, challenges remain in diagnosing, managing, and treating affected individuals. This study aims to explore the genetic underpinnings of HI, focusing on ABCA12 mutations and their impact on epidermal differentiation and the formation of the skin's stratum corneum. Additionally, the study evaluates the role of bioinformatics in improving diagnostic precision and therapeutic advancements, including gene therapy and skin-engineering approaches.

Materials & Methods:

A comprehensive review of genetic studies and bioinformatics tools was conducted to analyze their role in understanding HI. Whole-exome sequencing (WES) was examined for its effectiveness in identifying ABCA12 mutations and establishing genotype-phenotype correlations. Transcriptomic analyses were reviewed to assess the impact of these mutations on epidermal differentiation, while protein-structure modeling approaches were used to predict the functional consequences of lipid transporter defects. Furthermore, an evaluation of current therapeutic strategies, including topical treatments, systemic retinoids, and emerging gene-editing techniques, was performed to assess their potential for improving clinical outcomes.

Results:

Bioinformatics tools have greatly enhanced the ability to identify ABCA12 mutations with high precision, facilitating early diagnosis and personalized management strategies for HI patients. WES has been instrumental in detecting pathogenic mutations, while transcriptomic and protein-structure modeling analyses have provided insights into disease mechanisms and lipid transporter dysfunction. Clinically, current treatments remain supportive, but advances in gene therapy and skin-engineering hold promise for future interventions. The integration of bioinformatics in HI research has also improved our understanding of genotype-phenotype correlations, paving the way for targeted therapeutic strategies.

Conclusion:

The application of bioinformatics has significantly contributed to understanding the molecular mechanisms of HI, enabling earlier and more precise diagnoses and informing potential therapeutic approaches. While current treatments remain limited, emerging strategies such as gene therapy and skin-engineering offer hope for improved outcomes. Continued advancements in genetic research and bioinformatics will be essential in developing innovative treatments and enhancing the quality of life for individuals affected by this debilitating condition.

Abstract N°: 857

Case Presentation of Two siblings with Hailey-Hailey Disease

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Introduction & Objectives:

Benign Familial Pemphigus or Hailey-Hailey Disease is an autosomal dominant genodermatosis, caused by the mutation in ATP2C1 gene encoding the Ca2+/Mn2+ ATPase isoform 1 (hSPCA1) pump from the Golgi apparatus, with a role in the desmosomal protein complex, leading to acantholysis. It is typically characterized by vesicles that progress to bullae which then become eroded. They tend to appear in intertriginous areas, including the sides of the back and the neck and may be triggered by heat, perspiration, trauma, friction and UV light. There is no ethnic specificity and the F:M ratio is 1.

Materials & Methods:

We present the case of 2 patients, brother and sister, both affected by the Hailey-Hailey disease, with a positive history of the disease also having affected their father. Both the man and the woman had previous history of gastrointestinal tract cancer, in remission at the moment of the examination.

First, the female patient, 59 years old, presented to our clinic in June 2021. The eruption consisted of erythematous plaques with erosions and painful fissures in the inguinal area, accompanied by burning sensations, in the last 2 months. She mentioned having had several similar episodes in the last 3 decades.

The male patient, 66 years old, a former smoker, presented at our clinic with symmetrical erosive lesions, with central ulceration and hematic superficial crust, erythematous base, circinate well-defined margins, located on the arms, axillary and inguinal folds, scalp, back of the neck and on the back. He reported intense pruritus, pain and burning sensation for about 1 month, with previous similar episodes in the last 25 years. This time, the lesions were triggered by extreme heat and excessive sweat, as the patient presented in July. We mention that he also presented digital clubbing, which we interpreted as a clinical sign of the gastric lymphoma (in remission).

Results:

Considering the fact that both patients had had similar episodes in the past with no concrete diagnosis, skin biopsies were performed. The results showed suprabasilar and intraepidermal clefting, intraepidermal acantholysis of keratinocytes (“dilapidated brick wall”), polymorphic inflammatory infiltrate in the dermis and dyskeratotic keratinocytes with eosinophilic cytoplasm, in both cases. Histology was consistent with Hailey–Hailey disease.

Patients were advised to avoid local humidity and synthetic materials. They were recommended loose clothing and underwear and the use of non-soap surfactants for personal hygiene. The topical treatment consisted of boric acid 2% solution compresses, zinc oxide paste and erythromycin and nystatin lotions, together with systemic tetracyclines for up to 3 months, considering their anti-inflammatory effect. The evolution was good, with remission of the symptoms.

Conclusion:

The positive family history is a very important clue in the diagnosis of the Hailey-Hailey disease, though sporadic cases are also possible. Although not life-threatening, the disease presents a significant impact on the quality of life of the patients, as the symptoms can very often be debilitating. Follow up is necessary, the disease having a chronic, relapsing-remitting clinical course, the treatment at stopping the recurrent disease flares.

Abstract N°: 1047

Evaluation of dermoscopic photoaging in patients of Xeroderma pigmentosum

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Evaluation of dermoscopic photoaging in patients with Xeroderma pigmentosum: A prospective cohort analysis

Introduction & Objectives:

Xeroderma pigmentosum (XP) is an autosomal recessive disorder caused by defective nucleotide excision repair (NER), leading to impaired removal of UVB-induced DNA lesions. While XP serves as a model for premature photoaging, its dermoscopic characteristics remain inadequately characterized. This study aimed to evaluate dermoscopic features of photoaging in XP using the Dermoscopic Photoaging Assessment Scale (DPAS) and its correlation with clinical parameters.

Materials & Methods:

Twenty patients with XP, diagnosed based on clinical criteria and confirmed via complementation group analysis using DNA sequencing, underwent dermoscopic photoaging assessment with DPAS (0-44) and the Glogau scale. Sun protection behavior was evaluated using the Sun Protection Behavior Scale (SPBS: 0-45) at baseline and reassessed after six months following reinforced photoprotection strategies.

Results:

Eighteen patients had XP-C, while two had XP-A. The mean DPAS score was 13.15 ± 4.22 (range: 6-21), significantly correlating with the Glogau scale ($r=0.63$, $p=0.021$). Dominant dermoscopic findings included hypo- and hyperpigmented macules, lentigines, and telangiectasias, with minimal dermal photoaging manifestations. Actinic keratosis was observed in 77% of patients. No significant correlation was noted between DPAS and baseline SPBS ($r=0.31$, $p=0.31$) or tumor burden ($r=0.47$, $p=0.10$). However, SPBS scores improved significantly at six months ($p=0.01$) following sun-protection reinforcement.

Conclusion:

UVB-induced DNA damage, primarily via cyclobutane pyrimidine dimer formation, is the major driver of epidermal photoaging in XP due to its limited penetration depth till epidermis and superficial dermis. Although UVA can reach the reticular dermis, its oxidative DNA damage is predominantly repaired by the intact base excision repair pathway in XP patients. Consequently, XP accelerates epidermal rather than dermal photoaging due to defective NER mechanisms. Reinforced photoprotection effectively enhances sun-protective behaviors, underscoring the necessity for rigorous UV avoidance and early intervention in XP management.

Abstract N°: 1071

Diseases associated with Epidermodysplasia Verruciformis: 10 cases.

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Introduction & Objectives:

Epidermodysplasia verruciformis (EV) is a rare dermatological condition, typically inherited in an autosomal recessive mode, and is associated with both cutaneous and systemic diseases. Our objective was to determine the epidemiological, clinical, and evolutionary characteristics, as well as the pathological associations of this condition through a hospital-based series.

Materials & Methods:

This is a descriptive retrospective study including cases of EV with a confirmed diagnosis based on a combination of clinical and histological criteria.

Results:

We included 10 patients. The mean age was 24 years (ranging from 20 to 39 years) with a sex ratio of 1.5. The average age of onset was 6 years (ranging from 2 to 15 years). Family history of similar cases was found in 70% of the cases. All patients presented with depigmented, scaly macules, giving a pityriasis versicolor-like appearance, predominantly on the trunk, associated with hyperpigmented or achromic verrucous lesions on the limbs (90%), face (50%), trunk (40%), and pubic region (20%). The diagnosis of EV was clinically suspected and confirmed by histopathological examination. Associated pathologies included: congenital bone dysplasia with kyphoscoliosis (3 cases), restrictive respiratory failure (1 case), digestive tuberculosis (1 case), scabies (1 case), extensive dermatophytosis (1 case), and fibrotic hypersensitivity pneumonitis (1 case). Three patients developed tumor lesions localized on the face and scalp, with histopathological examination confirming the diagnosis of basal cell carcinoma (BCC) and squamous cell carcinoma (SCC).

Conclusion:

EV is a disease that typically manifests in childhood with polymorphic pityriasis versicolor-like lesions associated with verrucous or even pseudo-tumoral lesions, which are disseminated and persistent. It is currently classified into genetic and acquired forms. In our series, the early onset, presence of consanguinity, and similar cases within the family suggest a classic genetic form in all cases. It can be associated with genetic disorders such as chondrodysplasia, neurofibromatosis, and congenital bone dysplasia. It often complicates with skin tumors in photo-exposed areas, including BCC, SCC, melanomas, lymphomas, and eccrine porocarcinomas. Associations with digestive and Hodgkin's lymphomas have been reported. It can also be associated with pulmonary diseases (such as familial idiopathic pulmonary fibrosis) and endocrine disorders (such as autoimmune polyendocrinopathies). One patient in our series had fibrotic hypersensitivity pneumonitis. Associations with viral and fungal skin infections have also been reported. Monitoring of patients with EV is essential to detect complications, particularly malignant transformations.

Abstract N°: 1242

'Undercut sign'- A pattern of scarring alopecia in Junctional epidermolysis bullosa

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Title: 'Undercut sign'- A pattern of scarring alopecia in Junctional epidermolysis bullosa

Introduction & Objectives:

Junctional epidermolysis bullosa (JEB) is a hereditary disorder caused by defective dermal-epidermal adhesion due to protein deficiencies such as laminin-332, type XVII collagen, integrin α6β4, or integrin α3. It is characterized by blistering at trauma sites, resulting in chronic wounds, atrophic scarring, and granulation tissue formation. Complications include anemia, dental abnormalities, laryngeal stenosis, and genitourinary and gastrointestinal issues. Scalp blistering often leads to atrophic scarring alopecia involving the occipital scalp, as demonstrated in our two patients, underscoring the clinical importance of this finding.

Materials & Methods:

A 7-year-old male and a 15-year-old male, both born to non-consanguineous parents, presented with a history of recurrent tense fluid-filled blisters and erosions since birth, healing with atrophic dyspigmented scarring. Neither patient reported mucosal, gastrointestinal, or genitourinary complaints.

Physical examination revealed dyspigmented atrophic scars on acral sites, trunk, and back, along with dystrophic changes in all fingernails and toenails. Milia and granulation tissue were absent. Scalp examination showed well-demarcated scarring alopecia over the occipital and temporal regions with smooth, atrophic, dyspigmented skin and loss of follicular openings. Both patients also had scarring alopecia on the lateral halves of their eyebrows.

Results:

EB matrix analysis confirmed JEB-generalized intermediate (scores of 18 and 17, respectively). Genetic testing in Case 1 identified a heterozygous mutation in integrin β4

Conclusion:

Scarring alopecia in JEB arises from chronic inflammation, blistering, and secondary infections. The occipital region is particularly susceptible due to friction and pressure. We propose the term 'the undercut sign' to represent this localized hair loss pattern involving occipital and temporal scalp, potentially unique to JEB. While diffuse or androgenetic alopecia patterns have been reported in JEB-GI patients, localized scarring alopecia involving the occipital scalp, even in the absence of hallmark findings such as active blistering, granulation tissue, enamel defects, or extracutaneous signs, can serve as a diagnostic clue for JEB-GI.

Abstract N°: 1307

Familial capillary disorder indicative of Menkes Syndrome : Three siblings

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Introduction & Objectives:

Menkes disease is a rare X-linked recessive disorder caused by mutations in the ATP7A gene, which affects copper transport in the body. This leads to copper deficiency, particularly in the brain and other tissues, while copper accumulates in other organs . The dermatological features of Menkes disease are often among the earliest signs, particularly the characteristic hair abnormalities .

We report a family case: three siblings admitted for rarefaction of hair and eyelashes with hypopigmentation and epilepsy. Biological assay and genetic study revealed the diagnosis of Menkes Syndrome.

Cases' report :

We report the case of 3 siblings (two brothers and a sister) aged respectively five-years-old, three-years-old and two-months-old. These children came from non-consanguineous marriage, poorly monitored pregnancies. Admitted in Dermatology department for rarefaction of hair and eyelashes with pili torti at dermoscopy evaluation. The skin is hypopigmented and hyperlaxed . They presented growth delay and progressive neurological deterioration.The genetic study confirmed the diagnosis by mutations in the ATP7A gene. Our patients were put on anticonvulsant treatment, nutritional support and referred to genetic counseling.

Discussion:

Menkes disease MD (Or Kinky hair disease) is a rare genetic disorder that affects copper metabolism, leading to severe neurological and systemic symptoms. It is caused by mutations in the ATP7A gene, which is responsible for encoding a copper-transporting ATPase. This enzyme is crucial for the regulation of copper levels in the body, particularly in the brain and other organs.

MD typically presents in infancy, with symptoms appearing within the first 3 months of life. Key features include: Neurological symptoms : developmental delay, hypotonia , and progressive neurodegeneration. Dermatological features: hypopigmentation , pili torti Hair , cutis laxa, easy bruising. Skeletal abnormalities and systemic manifestations.The diagnosis of Menkes disease is typically made by genetic analysis to identify mutations in the ATP7A gene. In some cases, serum copper and ceruloplasmin levels may be measured.

Currently, there is no cure for Menkes disease, but early intervention with copper histidinate therapy can improve survival and some symptoms. However, the therapy is most effective when given before the onset of significant neurological damage. Other supportive treatments focus on managing specific symptoms, including anticonvulsants for seizures, physical therapy for motor dysfunction, and nutritional support. Despite treatment, the long-term prognosis remains poor, with most affected individuals experiencing severe developmental delays and early mortality. Familial cases of Menkes syndrome have been rarely documented in the literature such as Haddad et al. (2021) , Yi et al. (2020) , Kaler (2011) , Tümer et al. (2010). Our study emphasizes the importance of early diagnosis, genetic counseling, and molecular analysis to identify specific mutations in the ATP7A gene.

Conclusion:

Menkes disease is a damaging condition that affects copper metabolism and leads to severe neurological and systemic

abnormalities. While current treatments can improve outcomes if initiated early, the prognosis remains poor due to the progressive nature of the disease.

EA DV SYMPOSIUM PRAGUE 22-24 MAY 2025

Abstract N°: 1377

the gene' story

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Introduction & Objectives:

EBP X-linked dominant chondrodysplasia punctata 2 (CDPX2) is a rare genetic disorder with a prevalence of fewer than 1 in 400,000 newborns, with multi-system involvement and over 95% of the patients being female. This syndrome presents with dermatological, skeletal, neurological, and ophthalmological complications. Among these presentations, the dermatological management of ichthyosis is particularly challenging and crucial, as skin manifestations are significant and often provide critical diagnostic clues. This report highlights the complexities of diagnosing and treating such a multifaceted disorder, underscoring the essential role of dermatology within a multidisciplinary care framework.

Materials & Methods:

A one-day-old full term female neonate was referred with complaints of red, scaly and peeling skin at birth. Her APGAR score at birth was unremarkable. The pregnancy was uneventful and no significant family history from paternal side. Her mother was adopted and did have an episode of early miscarriage of a male foetus, a few years prior. Physical examination revealed an asymmetrically distributed erythematous blanching rash with fine feathery scaling along the Blaschko lines involving the baby's trunk and upper thighs, with the left side being more prominent. The rest of the examination was unremarkable. Clinical differentials at this point included congenital ichthyosis syndromes. As such, genetic testing emphasising on the ichthyosis panel was promptly arranged. In the meantime, an interim management plan was put in place for regular bath emollients and regular four hourly application of Fifty: 50 liquid paraffin ointment as an emollient.

Results:

This child had whole exome sequencing (WES) analysis for sequence variants in a virtual pattern for ichthyosis and erytherokeratoderma genes and palmoplantar keratoderma on the genomic DNA extracted from blood. Sequence variants were confirmed by fluorescent bidirectional Sanger sequencing using a new DNA dilution which revealed a diagnosis of EBP X-linked dominant ichthyosis (chondrodysplasia punctata- 2). This child had shown heterozygosity for EBP c.439C>T p.(Arg147Cys) pathogenic variant.

She is having regular follow-ups in the Dermatology department. Over time, the erythema and scaling have improved, revealing an underlying mottling appearance of the skin. This once again has improved over the last few months. She has been referred onwards to the paediatricians for regular monitoring and review of her syndromic diagnosis. She is currently 2 years old, with a height of 89 cm (50th percentile) and weight of 14.9 kg, placing her weight above the 91st percentile, which is currently managed under dietary supervision. She exhibits global developmental delays, being assessed for possible autistic spectrum disorder, and she is unsteady on her feet with delayed walking. Ophthalmological examination detected a squint, with no presence of cataracts. She is currently being managed through a collaborative effort across a broad range of specialties addressing her multifaceted needs.

Conclusion:

This case illustrates the fundamental role of mutational genetic testing to confirm the diagnosis of CDPX2. This case also highlights the essential role of dermatology in the multidisciplinary management of CDPX2, depicting how comprehensive

care can significantly enhance patient outcomes and quality of life.

EA DV SYMPOSIUM
PRAGUE
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Abstract N°: 1408

bullous congenital ichthyosiform erythroderma: from clinical suspicion to diagnostic confirmation

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Introduction & Objectives:

Bullous Congenital Ichthyosiform Erythroderma** (BCIE) is a rare genodermatosis, first described by Brocq in 1902, with an incidence of 1 in 300,000 live births. It is characterized by erythroderma, epidermal fragility, and skin barrier dysfunction from birth. Diagnosing BCIE is complex due to its phenotypic variability and the overlap with other congenital blistering diseases. It is associated with mutations in KRT1, KRT2, and KRT10, which affect epidermal keratinization.

Materials & Methods:

A clinical, histopathological, and genetic study was conducted on a pediatric patient initially diagnosed with dystrophic epidermolysis bullosa (DEB). A diagnostic reassessment by a multidisciplinary medical board was prompted by the persistence of atypical signs.

Results:

Case Presentation:

A six-year-old patient presented with generalized blisters and erosions from the first hours of life, initially diagnosed with dystrophic epidermolysis bullosa (DEB) (COL7A1 mutation: c.3550+4A>C). The presence of hyperkeratosis, palmoplantar keratoderma, and malodorous acral fissures led to a diagnostic review. A biopsy revealed epidermolytic hyperkeratosis (EHK), confirming BCIE.

Therapeutic Interventions:

Treatment with low-dose oral isotretinoin was initiated in the absence of acitretin, accompanied by emollients, 20% urea, topical antibiotics, and chlorhexidine baths. The patient showed significant clinical improvement.

Conclusion:

Discussion:

BCIE is an autosomal dominant disease with severe cutaneous manifestations from birth. Distinguishing it from epidermolysis bullosa and other congenital ichthyoses is challenging. Histopathology is crucial, revealing massive hyperkeratosis with vacuolar degeneration of keratinocytes. Treatment with systemic retinoids has proven effective in reducing hyperkeratosis and enhancing skin barrier function.

Conclusion:

This case underscores the importance of clinical, histopathological, and molecular correlations in diagnosing BCIE. Despite genetic advances, the molecular heterogeneity of the disease poses a challenge. A multidisciplinary evaluation is vital to optimize management and enhance the quality of life for patients. Regular reviews by medical boards are recommended to refine diagnostic and therapeutic strategies for complex pathologies like this one.

Abstract N°: 1612

Pruritus and neuropsychiatric symptoms among patients with Darier disease – an overlooked and interconnected challenge

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Title: Pruritus and neuropsychiatric symptoms among patients with Darier disease – an overlooked and interconnected challenge

Introduction & Objectives: Darier disease (DD) is a rare genetic autosomal dominant disorder caused by ATP2A2 gene mutations affecting calcium homeostasis in keratinocytes. Pruritus, a frequently overlooked symptom in DD, can lead to physical and emotional complications, especially in patients with DD who are genetically predisposed to psychiatric comorbidities. The combined emotional impact arising from genetic predispositions influencing psychological phenotypes and reactive phenomenon (emotional stress triggered by pruritus) presents a distinct clinical scenario that has yet to be evaluated. Thus, this study aimed to analyze pruritus and other related dysesthetic symptoms in patients with DD, and explore their correlation with disease severity and body surface area (BSA) involvement.**

Materials & Methods: The information analyzed in this study was extracted from the database of a previously published clinical study on DD. The data related to pruritus, which has yet to be published, was collected using two methods: (1) Dermatology Quality of Life Index, and (2) a physician-led interview using a clinical questionnaire. Descriptive statistics, Chi-square tests and Wilcoxon two-sample tests were used for analysis.

Results: A total of 76 patients, with equal distribution of gender and a mean age 44 years, were analyzed. The prevalence of pruritus was 90.8%, far surpassing other symptoms including pain (34.3%), tingling (38.2%), discomfort (57.9%), burning (32.9%), and malodor (43.4%). Various types and locations of DD lesions were pruritic, including papules and plaques on the ears, temples, neck, chest, and buttocks. Twenty-one patients had anxiety, mood, or psychotic disorders, and 58 reported neurological disorders. More patients with severe (92.3%) and moderate disease (97.3%) reported itch, compared to mild (84.0%). While pruritus was also associated with %BSA involvement, this was not statistically significant. Patients reporting pruritus had significantly higher DLQI itch scores (2.4 ± 1.0) than those who did not (1.5 ± 0.6), indicating accurate symptom reporting (p=0.03).

Conclusion: A striking majority of DD patients experience pruritus, with higher prevalence among those with severe disease and greater %BSA involvement. Clinicians should recognize pruritus as a key therapeutic target and adopt comprehensive treatment approaches that both address the neuropsychiatric comorbidities and additional psychological burden of pruritus in patients with DD. Moreover, the potential role of pruritus in the exacerbation of genetically predisposed neuropsychiatric conditions among patients with DD requires further evaluation.

Abstract N°: 1625

Pachydermoperiostosis, a rare hereditary disorder and ulcerative colitis and Ankylosing spondylitis, case report and literature review

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Introduction & Objectives:

Pachydermoperiostosis (PDP) or primary hypertrophic osteoarthropathy (HOA) is a rare osteo-cutaneous disorder which inherited in AR manner and characterized by skin thickening (pachydermia), finger and toes clubbing (Acropathy), hyperhidrosis, bone formation and joint pain and swelling.

This case report highlights the clinical cutaneous manifestations of PDP including skin thickening of face, fingers clubbing and internal diseases presentations and joint pain in a 27-year-old male.

Materials & Methods:

This gentleman referred to our clinic from Internal ward where he had been admitted to manage his underlying diseases including Ulcerative colitis (UC) and Ankylosing spondylitis (AS)

His characteristic clinical features during physical examination were compatible with PDP.

All presented data were collected by Direct physical exam, Paraclincal investigations and etc…

Confirmatory laboratory tests including prior GI endoscopy and imaging showed UC and AS.

Results:

Pachydermoperiostosis, is an Oseteocutaneous disorder with specific skin changes including increasing skin stiffness, forrowing and aged face, also digital and toes enlargement called clubbing are characteristic which both were present in our case.

On the other hand, association of PDP with inflammatory bowel diseases and rheumatologic diseases is quite intersting.

Most of reports, highlighted the association between PDP and crohn disease, but our research showed association between pachydermperiostosis and Ulcerative colitis and ankylosing spondylitis simultaneously.

Conclusion:

Pachydermoperiostosis is a rare disorder and some features of the disorder mimicks physiologic and pathologic conditions.

For exmaple its aged appearance in facial area may be confused with normal aging process

And because of the burden the disease and associations as mentioned earlier, become more familiar with the disorder seem reasonable.

Abstract N°: 1651

Artesunate treatment in a patient with severe junctional epidermolysis bullosa

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Introduction & Objectives:

Severe junctional epidermolysis bullosa (sJEB) is a rare genetic, often postpartum lethal epithelial disease, predominantly caused by biallelic nonsense/premature termination codon (PTC) sequence variants in the LAMB3 gene. LAMB3 encodes Lamb3, the β3 chain of the trimeric, hemidesmosomal skin anchor laminin-332. PTC mutations prevent synthesis of full-length functional proteins, and truncated Lamb3 prevents formation of laminin-332, with devastating clinical manifestations. The condition is currently incurable and would require long-term systemic correction. Artesunate is an FDA-approved, bioavailable small molecule drug for systemic treatment of malaria. Targeted ribosome editing in yeast and human cells identified artesunate as an efficient modulator of Lamb3 protein translation enhancement. Therefore, the parents of a sJEB child agreed to start the off-label treatment with artesunate.

Materials & Methods:

Quantification of immunofluorescence investigation of keratinocytes: HaCat model cells comprise samples PARENTAL CONTROL (original HaCat keratinocytes), HETERO CONTROL and HOMO CONTROL, where homozygote and heterozygote deficiencies in LAMB3 expression were generated using CRISPr/Cas9. HOMO ART, a homozygote-deficient LAMB3 R635X cell culture was treated with artesunate 1uM for 4 days. Samples of E6/E7 immortalized keratinocytes were derived from cells isolated from unaffected proband, HC-1399 CONTROL, from LAMB3 R635X heterozygote carrier, HC-1199 CONTROL and from the homozygote patient, JEB-1422 CONTROL, and from cells isolated from the patient treated with artesunate 1uM for 4 days, JEB-1422 ART.

Results:

Artesunate was administered initially in September 2023 intravenously with a total amount of 3 mg q.d., then switched to an oral application with 10 mg daily (1 ml of 10 mg/ml oral suspension) and increased to 20 mg after 3 months. An immunofluorescence (IF) investigation of patient skin was performed after 5 months of treatment and showed only a marginal increase in expression of Lamb3 protein in the skin. After 6 months, oral dosing was increased to 40 mg daily. There were signs of clinical efficacy: during the first 3 months, no effect on wound healing was observed according to the treating physician's assessment. After 6 months of treatment (1 week of 40 mg oral artesunate), the patient's wounds on the nates began to show gradual healing. However, the patient succumbed to the illness in spring 2024.

Conclusion:

Safety was acceptable, and in areas where artesunate had local access (oral cavity, perianal), positive clinical effects were

reported by treating physicians and visually documented. There are reasons why the clinical outcome in this patient could not be changed: 1) Bioavailability of artesunate in the skin as opposed to the blood system. 2) The duration and dosage of treatment, which probably preclude the accumulation of Lamb3 as expected from the mode of action. 3) Effects of prenatal loss of LAMB3 (immune dysregulation).

EADV SYMPOSIUM PRAGUE 22-24 MAY 2025

Abstract N°: 1672

Segmental Darier Disease: A Case Report

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Introduction & Objectives: Darier disease (DD), or keratosis follicularis, is a rare autosomal dominant genetic acantholytic dermatosis caused by mutations in the ATP2A2 gene in keratinocytes. This mutation results in the loss of intercellular adhesion, leading to acantholysis. The prevalence ranges from 1 in 30,000 to 1 in 100,000, with equal gender distribution. DD typically presents as pruritic, generalized, symmetrical keratotic papules and plaques, mainly in seborrheic and intertriginous areas. Segmental Darier disease is a rare, localized form of this condition. This report aims to highlight the clinical and histopathological features of segmental Darier disease and discuss the management strategies.

Materials & Methods: A 57-year-old male with no significant medical history, except for hypercholesterolemia, presented with pruritic lesions. There was no family history of similar conditions. Physical examination revealed brown papules following Blaschko lines on the right upper extremity and upper back. No mucosal or nail involvement was observed. Dermoscopic examination showed yellow-brown polygonal areas. Laboratory investigations, including CBC, liver and kidney function tests, sedimentation rate, and CRP, were within normal limits, while lipid profile indicated hyperlipidemia. A skin biopsy was performed for histopathological examination.

Results: Histopathological analysis showed papillomatosis and parakeratosis with focal intraepidermal acantholysis and dyskeratotic cells. The upper dermis exhibited increased vascularity and mild perivascular lymphocytic infiltration. Immunohistochemical staining was negative for S-100 and CD1a. Based on clinical, dermoscopic, and histopathological findings, a diagnosis of segmental Darier disease was made. The patient was treated with acitretin at 10 mg/day. Follow-up demonstrated significant improvement in the lesions.

Conclusion: Segmental Darier disease, a rare form of Darier disease, requires careful clinical and histopathological evaluation for accurate diagnosis. This case underscores the efficacy of acitretin therapy in managing segmental Darier disease. Regular monitoring is essential due to the potential neuropsychiatric side effects associated with retinoid therapy. This report contributes to the growing body of knowledge on this uncommon dermatological condition and highlights the importance of considering segmental Darier disease in the differential diagnosis of linear acantholytic dermatoses.

EA DV SYMPOSIUM PRAGUE 22-24 MAY 2025

Abstract N°: 1710

Segmental Darier Disease in a Blaschkoid Distribution: A Rare Case Report and Systematic Review of the Literature

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Introduction & Objectives:

Darier disease (DD), also known as keratosis follicularis, is a rare autosomal dominant disorder characterized by pruritic, warty keratotic papules and plaques in seborrheic and intertriginous areas. In some cases, a segmental form presents with localized lesions along the lines of Blaschko, resembling those of generalized DD. Genomic mosaicism in segmental DD lesions is a recent concept observed in individuals with this disease.

We present a case of segmental DD. Since this is a rare genodermatosis, we made a comprehensive review of the previously reported cases and evaluated the compatibility with our case.

Case Report & Methods:

A 41-year-old woman presented with a 2-year history of odorless, red-brown hyperkeratotic papules localized to the right forehead and right trunk. The lesions worsened in summer, with menstruation and sweating. She had no comorbidities, no family and medication history. Histopathological features are shown in Figure 1.

She had previously attempted diverse topical treatments with minimal results. We opted for low-dose oral isotretinoin (10mg/day) and which led to alleviation of symptoms within one month.

A systematic search was conducted using the terms ‘segmental Darier’, ‘localized Darier’, ‘unilateral Darier’, and ‘zosteriform Darier’ across PubMed, Google Scholar, and Scopus, since 2000. 67 articles and 83 cases were included, after eliminating irrelevant and duplicate studies. We analyzed demographic, clinical, histopathological, and treatment data, and compared our case with literature reports.

Results:

There was a female predominance of 60% (n=49). The mean age at diagnosis was 44.7 years (median=42.5), with a mean onset age of 33 years (median=32). The diagnostic delay had a standard deviation of 11 years (Fig 1). The majority (n=76, 89%) were classified as type 1 segmental DD, with localized blaschkoid lesions. Trunk was the most common site of involvement (Fig 3). Histopathologically, all cases showed classical DD features. Disease progression analysis revealed sun exposure and sweating, especially in summer, as the most common exacerbating factors (43%). Treatment modalities and responses are given in Table 1.

Conclusion:

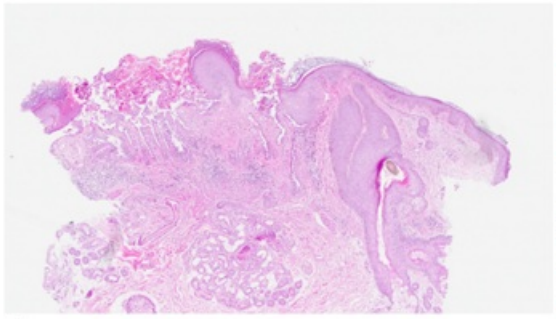
The clinical presentation of most patients closely resembled classical DD, with the exception of the lateralized and blaschkoid distribution. Malodor (2%) and secondary infections (3%) were less frequent than in classical DD. Diagnosis was primarily based on clinical and histopathological findings, with genetic testing conducted in only 12 cases (14%).

In our case, while the patient’s age at diagnosis and gender were consistent with the literature, the age of onset was higher than the literature mean and the time to diagnosis was shorter. The clinical features, including lesion lateralization and trunk involvement, were consistent with published data. Symptoms worsened during summer due to heat and sweating, as reported in the literature. A favorable clinical response within one month was achieved with oral retinoid

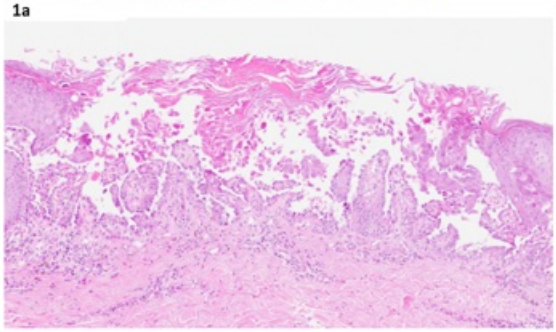
treatment, consistent with previous reports.

Given the rarity of segmental DD, we present this case report and systematic literature review. Our case aligns with the literature in terms of demographic, clinical and histopathological features, and treatment response. As no prior systematic review on this topic exists, we believe this study offers valuable contributions to the literature.

Figure 1



1a



1b

Both biopsies revealed acantholysis causing suprabasal cleft formation in the epidermis. There are villous structures lined with a single layer of basal cells. There is hyperkeratosis on the epidermal surface, and dyskeratotic cells (corp-grain) were observed among the acantholytic cells. There is minimal perivascular inflammation in the dermis.

Figure 2

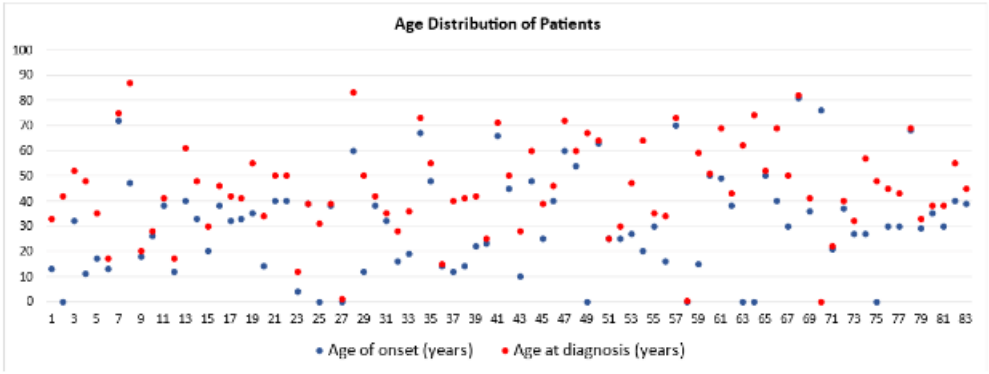
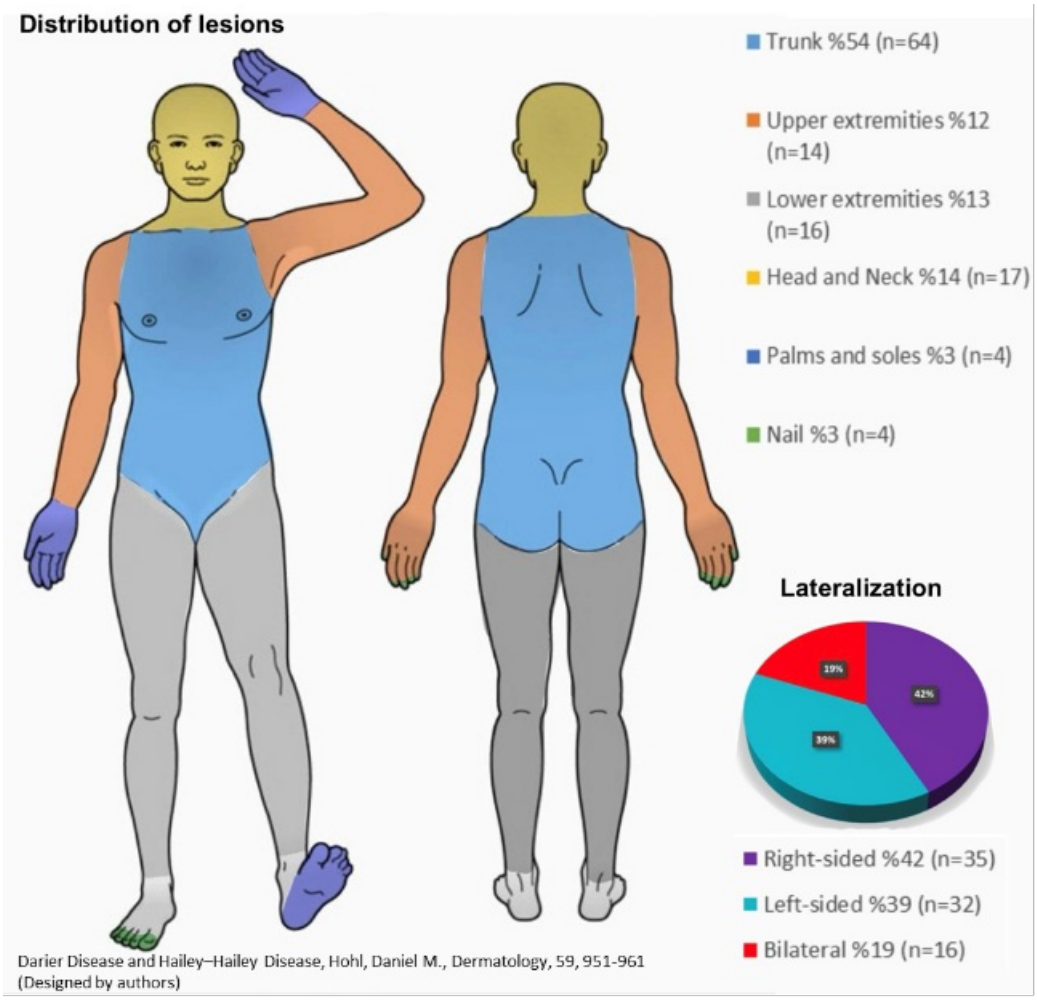


Figure 3



Darier Disease and Hailey–Hailey Disease, Hohl, Daniel M., Dermatology, 59, 951-961
(Designed by authors)

Table 1

Treatment (N = 107)	Well Response	Partial Response	No Response
Keratolytics (11)	9%		
Topical corticosteroids (29)	34%	17%	48%
Topical retinoids (35)	48%	40%	11%
Topical D3 analogues (3)	66%		
Oral retinoids (21)	61%	19%	4%
Oral antibiotics (3)	33%		
Cryotherapy (2)	100%		
CO_2 laser (3)	66%	33%	

Abstract N°: 1723

H syndrome: a diagnosis not to be dismissed

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Introduction & Objectives:

Syndrome H is a rare, newly described genodermatosis characterized by the frequency of skin lesions strongly suggestive of the diagnosis. It deserves to be better investigated, as it can be associated with life-threatening systemic disorders.

Materials & Methods:

A case report

Results: A 14-year-old patient from a 1st degree consanguineous marriage with a past medical history of a type 1 diabetes presented for asymptomatic hyperpigmented patches on the inner thighs evolving for 6 years. Clinical examination revealed symmetrical hyperpigmented plaques that were indurated on palpation, suggesting atypical plaques of morphea due to the presence of hyperpilosity. The rest of the examination revealed a camptodactyly-type toe deformity and hallux valgus. A skin biopsy showed a slightly acanthosic epidermis and a hyperpigmentation of the basal layer. A dense lymphoplasmacytic inflammatory infiltrate occupied the dermis and descended to the fatty lobules, associated with lipophages, significant fibrosis and thickening of the elastic fibers. Immunohistochemistry revealed CD68 and PS 100 positivity and CD1a negativity. H syndrome was retained. Disease assessment revealed conductive hearing loss. Cardiac ultrasound was without abnormalities.

Conclusion:

H syndrome is an autosomal recessive genodermatosis, most common in Arab races, secondary to mutation of the SLC29A3 gene. Syndrome H is classified in the group R of histiocytosis and is considered as an inherited form of Rosai Dorfman disease. Hyperpigmented plaques usually associated with induration or hypertrichosis represent the most common feature of the disease. It affects mostly the inner thighs or calves, and it is usually associated with “camptodactyly”, as in our patient’s case. The clinical presentation may include growth retardation, deafness, hepatomegaly, splenomegaly, hypogonadism, hyperglycemia and life-threatening cardiac abnormalities.

Our case is original in that it illustrates the typical presentation of H syndrome, which although rare, deserves to be better known given the severity of associated systemic damage that can be life-threatening and functionally compromising.

EA DV SYMPOSIUM PRAGUE 22-24 MAY 2025

Abstract N°: 1770

status epilepticus in an unrecognized nf1 : a case report

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Introduction & Objectives:

Neurofibromatosis type 1 (NF1), an autosomal dominant disorder linked to chromosome 17 (1/3,500 births), is characterized by café-au-lait macules, neurofibromas, and skeletal anomalies. While epilepsy in NF1 is rare and often associated with gliomas or cortical malformations, this case report highlights an exceptional etiology: temporal lobe epilepsy secondary to sphenoid dysplasia-induced temporal lobe herniation. We aim to elucidate this rare mechanism of epilepsy in NF1, underscore diagnostic challenges, and emphasize the importance of craniofacial imaging in NF1 patients with seizures.

Materials & Methods:

A 23-year-old woman with NF1 (diagnosed via café-au-lait macules and cutaneous neurofibromas) presented with complex partial seizures since age 11. Her history included kyphoscoliosis repair (age 5) and cervical neurofibroma resection (age 15). Family history noted café-au-lait macules without epilepsy. Diagnostic evaluation included:

Clinical examination: Confirmed NF1 stigmata; no exophthalmia or facial asymmetry.

Neuroimaging: MRI/CT revealed sphenoid dysplasia with temporal fossa enlargement, leading to temporal lobe herniation and meningocele formation. EEG: Localized seizure onset to the temporal lobe. Genetic testing for NF1 mutations was not performed due to definitive clinical diagnosis.

Results:

Imaging demonstrated severe sphenoid dysplasia causing herniation of the atrophic temporal lobe into the orbital cavity, compressing adjacent structures. No gliomas or cortical malformations were identified. The seizures were attributed to mechanical irritation from the herniated lobe. Despite antiepileptic drugs, refractory seizures persisted, implicating structural etiology. Surgical intervention for the sphenoid defect was deferred due to anatomical risks.

Sphenoorbital dysplasia, a pathognomonic but rare NF1 complication (<1% of cases), typically presents with pulsatile exophthalmos or facial asymmetry. This case uniquely links sphenoid dysplasia to epilepsy via temporal lobe compression, a mechanism scarcely reported. Unlike NF1-associated epilepsy from gliomas, this structural anomaly underscores the need for detailed craniofacial imaging in seizure evaluation. The absence of cortical malformations or tumors highlights the role of mechanical stress in epileptogenesis.

Conclusion:

This case illustrates an exceptionally rare cause of epilepsy in NF1—temporal lobe herniation due to sphenoid dysplasia. Key takeaways include:

Diagnostic Imaging: Prioritize craniofacial MRI/CT in NF1 patients with refractory seizures, even without typical tumors. Pathophysiological Insight: Mechanical compression from bony defects can drive epilepsy, expanding the etiological spectrum. Research Imperative: Further genetic studies are needed to explore NF1-related bone dysplasia mechanisms and

their neurological sequelae. Clinicians must remain vigilant to atypical epilepsy etiologies in NF1, ensuring multidisciplinary management to address structural and neurological complexities

EADV SYMPOSIUM PRAGUE 22-24 MAY 2025

Abstract N°: 1908

Gorlin-Goltz Syndrome: Diagnosis, Management, and Treatment – Case Report

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Introduction & Objectives:

Gorlin-Goltz syndrome, or basal cell nevomatosis, is a rare, hereditary condition with autosomal dominant transmission. It is characterized by a triad of multiple basal cell carcinomas, odontogenic keratocysts, and rib abnormalities. Caused by mutations in a tumor suppressor gene on chromosome 9, it affects both men and women, especially Caucasians, with an incidence of 1/50,000 to 1/150,000. Early screening and regular monitoring of patients and their families are essential due to its hereditary nature.

Materials & Methods:

A 37-year-old man with a history of uncontrolled hypercholesterolemia and anemia was referred for treatment of multiple facial tumors. Dermatological examination revealed three basal cell carcinomas and several nevi on his face and body. Additionally, he had facial abnormalities, including prominent frontal bosses, hypertelorism, and maxillary prognathism. Radiological tests showed multiple mandibular odontogenic cysts. These signs, along with the basal cell carcinomas and facial anomalies, led to the diagnosis of Gorlin syndrome. Further tests showed no skeletal malformations or family history. The patient underwent excision of the carcinomas, with histopathological confirmation of Gorlin-Goltz syndrome.

Results:

Basal Cell Nevus Syndrome (BCNS) is a rare genetic disorder caused by mutations in the PTCH1 gene, leading to disruptions in the Hedgehog signaling pathway, which regulates cell growth. This results in the early onset of multiple basal cell carcinomas (BCC), often starting in childhood or puberty, and the development of other features like odontogenic keratocysts, facial anomalies, and rib/spinal abnormalities. Patients may also experience rare tumors and other complications. Treatment focuses on regular monitoring, surgery to remove BCCs or cysts, and preventive measures. While there is no cure, ongoing research into genetic therapies offers potential for future treatments.

Conclusion:

Gorlin and Goltz syndrome is a rare genetic disorder with dominant inheritance, marked by basal cell skin tumors, jaw cysts, and bone malformations. It requires early detection, especially for dentists who can spot jaw cysts on radiographs. The condition, caused by a mutation on chromosome 9, carries a high risk of skin cancer and necessitates long-term monitoring. Treatment is symptomatic and involves an interdisciplinary approach to manage various symptoms.

Abstract N°: 2011

Piecing Together the Puzzle: Follicular Atrophoderma, Hypotrichosis and BCCs

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Introduction & Objectives: Bazex-Dupré-Christol Syndrome (BDCS) is an infrequently encountered genodermatosis exhibiting an X-linked dominant transmission, clinically characterized by the triad of multiple basal cell carcinomas (BCCs), follicular atrophoderma and hypotrichosis. Despite its rarity, BDCS necessitates vigilance due to the potentially aggressive and recurrent nature of BCCs in photoexposed regions. We aim to illustrate a complex presentation of BDCS in a 35-year-old female, emphasizing the importance of high clinical suspicion, dermoscopic evaluation, histopathological confirmation and tailored therapeutic and surveillance strategies.

Materials & Methods: A 35-year-old female presented with multiple, variably pigmented BCCs located on the upper lip, periorbital regions (involving both the superior and inferior eyelids) and the nasal sidewall extending to the nasal tip, as well as the malar eminence. Clinical examination further revealed follicular atrophoderma on the dorsal aspects of her hands, prominent facial milia and overall hypotrichosis. Dermoscopic assessment of the cutaneous neoplasms disclosed classic arborizing telangiectatic vessels, along with multiple blue-gray globules suggestive of pigmented BCCs. Trichoscopy demonstrated miniaturized and dystrophic hair shafts, kinked hairs and a receding frontal hairline.

Her 10-year-old son exhibited follicular atrophoderma over the dorsal hands and extensive facial milia, but no evidence of cutaneous malignancies. Other personal or familial pathologies were unremarkable. Surgical excision of the pigmented lesions was undertaken and histopathological analysis was performed on formalin-fixed, paraffin-embedded tissue specimens.

Results: Histopathology confirmed pigmented basal cell carcinomas with extension into the deep reticular dermis, displaying peripheral palisading and a characteristic mucinous stromal reaction. Excision margins were tumor free in all specimens. Given the presentation with multiple BCCs in photoexposed sites, follicular atrophoderma and hypotrichosis, BDCS was diagnosed. The patient was advised on rigorous photoprotection and a structured follow-up schedule was implemented, including biannual dermatologic examinations and dermoscopy for the prompt identification of new or recurrent BCCs.

Conclusion: This case exemplifies the complexity and clinical heterogeneity of BDCS, underscoring the necessity for meticulous dermatologic assessment of both cutaneous lesions and adnexal structures. Early and accurate recognition facilitates optimal surgical management, lowers morbidity rates, and mandates ongoing surveillance to lower the occurrences of advanced BCCs . Enhanced awareness of this syndrome among dermatologists and genetic counselors is essential, given its potential for hereditary transmission and the critical role of vigilant photoprotection and regular dermoscopic monitoring in improving patient outcomes.

EA DV SYMPOSIUM PRAGUE 22-24 MAY 2025

Abstract N°: 2029

Disseminated Superficial Porokeratosis-A Rare but Recognizable Dermatologic Entity

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Introduction & Objectives:

Porokeratosis is a rare chronic disorder of keratinization which can be aquired or inherited.The risk factors for porokeratosis include genetic factors (inherited or de novo genetic defects) , imunosupression and exposure to UV light. Disseminated superficial porokeratosis is one of the multiple forms that have been described , characterized by erythematous, skin-colored or hyperpigmented, well defined macules or annular papules and plaques with a hyperkeratotic and elevated margin histologically corresponding to cornoid lamella.

Lesions manifest in both sun-exposed and non-sun-exposed areas, typically emerging during childhood, most frequently between the ages of 5 and 10.Treatment options include topical therapy (retinoids, fluorouracil, imiquimod, vitamin D analogs, diclofenac, tacrolimus, keratolytic agents), destructive therapies, surgical excision and oral retinoids, but the response is usually poor and the disease usually progress.

Materials & Methods:

A 39-year-old female patient presented with an eruption consisting of well-demarcated, hyperpigmented and skin-colored macules with a peripheral hyperkeratotic ridge, non-pruritic, distributed on the lower legs, arms, and forearms. The eruption first appeared at the age of 13, and she had not undergone any treatment until the time of consultation. She reported increased hyperpigmentation of the lesions during the summer when exposed to sunlight and with prolonged standing, as well as a progressive increase in the number of lesions with aging.

Regarding her family history, it was noted that her father exhibits a clinically similar eruption, though no histopathological confirmation has been obtained to establish a definitive diagnosis.

Routine blood tests were within normal limits,with no evidence of systemic disease. A biopsy was performed from one of the lesions, and histopathological examination revealed hyperkeratotic, angulated epidermal invaginations with surface parakeratosis—cornoid lamellae—with underlying areas of agranulosis and vacuolated and/or dyskeratotic keratinocytes, findings consistent with a diagnosis of disseminated superficial porokeratosis.

Results:

Given the extent of the lesions, a treatment plan was initiated, consisting of topical therapy with trifarotene cream, applied once every two days, in combination with a keratolytic emulsion containing 10% urea. Additionally, systemic therapy with vitamin A was prescribed in two courses of 20 days per month.After two months, the patient returned for follow-up, and only mild improvement was observed, particularly in the lesions on the forearms and arms, which showed slight depigmentation. The therapeutic regimen was adjusted by introducing tacrolimus 0.1% ointment, applied once every two days, alternating with trifarotene cream. Furthermore, the patient began narrowband UVB phototherapy (NB-UVB) three times per week, leading to additional lesion improvement.

Conclusion:

Although the risk is low, patients with porokeratosis should be regularly monitored and seek medical attention if they notice any changes in the clinical appearance of their lesions, in order to prevent malignant transformation. The most common malignancy associated with porokeratosis is squamous cell carcinoma, followed by basal cell carcinoma and

Bowen’s disease.In this context, the use of sunscreen creams and UV-protective clothing is recommended as preventive measures.

EADV SYMPOSIUM PRAGUE 22-24 MAY 2025

Abstract N°: 2066

The Multiple Faces of RASopathies

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Introduction & Objectives: Legius syndrome (LS) is a rare RASopathy that is caused by heterozygous variants of the SPRED1 gene located on the 15q14 chromosome.. LS is characterized by café-au-lait macules, skin freckling, neurobehavioral issues, macrocephaly being sometimes present. In rare cases, it can be associated with Noonan-like facial features, multiple lipomas, short stature and pectus deformities. Neurofibromas, Lisch nodules, gliomas or malignancies are not characteristic for LS. Since it bears overlapping symptoms of neurofibromatosis type I (NF1), around 2% of the patients suspected to have NF1present actually a mutation of the SPRED1 gene.

We report a case that illustrates the fact that Legius syndrome should be taken into account when encountering a milder phenotype of RASopathies.

Materials & Methods: A 5-year-old girl was reffered for a suspicion of NF1 because of multiple café-au-lait spots with a diameter over 5 mm on the upper limbs, chest, abdomen and lower limbs, associated with axillary and inguinal freckllling, as well as mild thoracolumbar scoliosis. Thorough clinical examination showed no cutaneous neurofibromas, no macrocephaly. There was no family history of NF1 and none of the family members had any clinical signs of NF1.

Results:

The Multiplex Ligation-dependent Protein Amplification detected no duplication or deletion of the neurofibromin 1 gene and no mutation was found with Next-Generation Sequencing. A Whole Exom Test was performed identifing a heterozygous missense variant SPRED1 c.305C>T, p.(Thr102Met) which is suggestive for LS.

Conclusion: Even though there are patients fulfilling NF1 diagnostic criteria based on the presence of more than five CALMs and possible freckling, the differential diagnostic with RASopathies like the Legius syndrome should be taken into consideration. Thorough genetic testing plays an important role in the confirmation of the diagnosis, but also regarding the further management of the pathology.

EADV SYMPOSIUM PRAGUE 22-24 MAY 2025

Abstract N°: 2069

Neurofibromatosis Type 1 and the Birt-Hogg-Dubé Syndrome. A Rare Association

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Introduction & Objectives: Neurofibromatosis type I (NF1) is an autosomal inherited RASopathy that appears with a prevalence of 1:3000. It is caused by mutations in the neurofibromin gene characterized by cutaneous signs such as café-au-lait macules (CALMs), neurofibromas, hyperpigmentation, soft skin touch or vascular lesions. Extra-cutaneous signs include ocular, neurological, skeletal, vascular, cardiologic manifestations, but also a proneness to cancer forms like brain tumors, leukemias, pheochromocytomas, rhabdomyosarcoma, mammary cancer, melanoma.

Birt-Hogg syndrome (BHDS) is an autosomal dominant genetic condition caused by germline mutations in the gene FLCN, a tumor suppressor which encodes folliculin and appears in 1:20000 of the population. Dermatological manifestations include the classic triad of fibrofolliculomas in 90% of the patients, trichodiscomas and acrochordons, whilst other features are multiple pulmonary, recurrent spontaneous pneumothorax and renal tumors. Most BHDS patients develop skin lesions, with 90% having skin fibrofolliculomas.

We report a rare case an association of NF1 and BHDS aiming to emphasize the importance of genetic testing and multidisciplinary collaboration in the management of genodermatoses.

Materials & Methods: An 11-year-old male patient presented with multiple CALMs bilaterally, with axillary and inguinal freckling. Other findings include upside-down triangular facias, ocular hypertelorism, broad nasal root, short stature, a moderate form of scoliosis, concentration and language acquisition difficulties. He underwent orchidopexy at the age of 3 years. Genetic testing of 4813 genes was performed.

Results: The genetic panel identified two pathogenitc variantsl, one in heterozygous status in the NF1 gene NM_001042492.3:c.5651T>G, which establishes the diagnosis of Neurofibromatosis type 1 with autosomal dominant transmission and another one in the FLCN gene, NM_144997.7:c.1214dup, with autosomal dominant transmission., establishing an association with the BHDS.

Conclusion: Multidisciplinary management is especially important in cases such as this, where more than 50% of BHDS patients experience pneumothorax at least once as emergency, mostly at the age of 30 years, have skeletal and neurological manifestations and are prone to different malignancies because of the NF1-BHDS association. Although skin lesions increase in size and number with age both in NF1 and in BHDS, early skin evaluation is of uttermost importance as a differential diagnosis and referral to the genetic department for testing in order to prevent complications that might appear.

Abstract N°: 2087

Genetic Testing Criteria of Familial Melanoma: A Retrospective Cross-Sectional Analysis

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Introduction & Objectives:

Hereditary melanoma susceptibility gene testing enables identification of pathogenic variants in familial melanoma (FM) clusters, most commonly the *CDKN2A* gene, which encodes the tumour suppressor proteins p16 and p14. Genetic counselling and testing may facilitate enhanced surveillance, preventative behaviour and early intervention, leading to improved survival rates. This study sets out to evaluate the utility of the UK National Genomic Test Directory (NGTD) eligibility criteria for FM.

Materials & Methods:

We conducted a retrospective cross-sectional analysis of referrals to our regional genetics service in the UK (n = 141) between 2012 and 2023. Descriptive statistics and regression analysis were used to assess the association between age, family and personal history of melanomas and/or melanomas in situ (MMIS) with genetic testing outcomes.

Results:

The cohort included 44 males, and 97 females aged 1 to 86 (mean age = 46.9 ± 17.4). 68.1% (n=96) of referred patients underwent genetic testing, out of which 30.9% (n=30) tested positive for a genetic variant. Of these, all patients (96.7%, n=29) harboured a *CDKN2A* pathogenic variant (PV) aside from one patient who was a carrier of PTEN PV. 55.2% (n=53) of those tested met the UK NGTD eligibility criteria for testing, considering 29 patients were tested before introduction of NGTD in 2018.

Out of the 29 patients with *CDKN2A* PV, at least 15 (51.7%) were part of cascade testing, and 11 (37.9%) probands were index cases. The distribution of age of index case was right skewed, with majority of patients lying in 40s-50s (45.5%, n=5) and above 60s (36.4%, n=4) age groups. This is the reverse in the cascade cohort, where 42.9% (n=6) were below age 40.

58.6 % (n = 17) of those with *CDKN2A* PV had at least one personal history of MMIS, and the mean number of melanomas these patients had was 1.07 ± 1.25 (mode = 0). The mean age of first diagnosis of MMIS was 35.2 ± 15.58. Interestingly, 60.0% (n=9) of the cohort carrying a *CDKN2A* PV as part of cascade testing had no MMIS (mean age 45.4 ± 16.2).

All probands with *CDKN2A* PV have a family history of all-cause malignancy. 62.1% (n=18) has one first-degree relative with MMIS, whereas 20.7% (n=6) has two and 6.9% (n=2) has three first-degree relatives with MMIS respectively. Overall, a family history of MMIS in first-degree relatives was associated with a *CDKN2A* PV (OR = 1.882, CI:1.078-3.484). Two of those with *CDKN2A* PV had a first-degree relative with pancreatic cancer aged <60.

Conclusion:

Our data shows that a family history of MMIS in first-degree relatives is associated with a*CDKN2A* PV, reinforcing its value as a key eligibility criterion in identifying hereditary melanomas. The distribution of age in our cohort with *CDKN2A* PV suggests testing remains relevant for the elderly. In predictive testing, a personal history of MMIS has less weight in

predicting a PV, likely reflecting the younger age of referral and cumulative risk exposure in tumorigenesis, regardless of genetic predisposition. This underscores the role of genetic screening for early risk identification and prevention, as well as risk stratification by age group.

Abstract N°: 2182

Late diagnosis of epidermolysis bullosa pruriginosa: a rare form of the disease

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Introduction & Objectives: Epidermolysis bullosa (EB) is a family of inherited genodermatoses characterized by blistering in response to minor trauma. Blistering level categories are the simplex, hemidesmosomal, junctional, and dystrophic subtypes. Cutaneous involvement varies from localized to widespread blistering depending on subtype. Inheritance can be autosomal dominant or recessive. Clinically, dominant dystrophic EB has the following types: 1) generalized, 2) acral, 3) pretibial, 4) pruriginosa, 5) nails only and 6) bullous dermatolysis of the newborn. EB pruriginosa is a very rare variant of dystrophic EB (DEB) caused by type VII collagen gene mutation, with distinctive clinico-pathological features. It is characterized by nodular prurigo-like lichenified lesions, nail dystrophy, and variable presence of albopapuloid lesions.

Materials & Methods: We report two cases of EB pruriginosa, 71-year-old father and his 44-year-old son, with blisters after minor trauma from the first year of life, mainly on the hands, feet, knees, lower legs and later on the scalp and the trunk. The father had been treated with acitretin as psoriasis, and the son was suspected of having lichen planus and epidermolysis without specification.

Results: On examination, the son had atrophic scars on the scalp, numerous slightly elevated whitish scars on the trunk (albopauloid lesions), and numerous erythematous lichenoid excoriated papules on the forearms, lower legs, dorsum of the hands, and feet, while the father had predominantly hyperkeratotic excoriated papules on the lower legs and dorsum of the feet. Both had intense pruritus. Both had hyponychia on the fingers and toes. There were no lesions in the oral cavity. Skin biopsy showed subepidermal blister filled with fibrin. Direct immunofluorescence was negative. Dermatology profile for autoimmune bullous dermatoses was negative. Sequencing the coding exons and exon-intron boundaries of 4813 clinically relevant genes in both father and son revealed a heterozygous pathogenic variant c.6034G>A p.(Gly2012Ser) in the COL7A1 gene.

Conclusion: EB pruriginosa is a type of dystrophic EB, described by McGrath in 1994. There is a clinical overlap of DEB pruriginosa with pretibial type DEB (lichenoid papules, milia, nail dystrophy, delayed onset). Patients with pretibial DEB often experience localized lesions, pruritus at the site of disease, while DEB pruriginosa patients often experience more intense pruritus and the disease may be more widespread. There are no effective treatments nor cure for DEB pruriginosa. Treatment is symptomatic and is aimed at controlling pruritus and halting the progression of cutaneous lesions. Potent topical steroids and intralesional triamcinolone have been reported to reduce the pruritus in some cases, but do not produce sustained improvement. Systemic therapy with H1 antihistamines and corticosteroids had no sustained effect. However, successful results have been achieved with topical tacrolimus and systemic thalidomide, cyclosporine and dupilumab. **

EADV SYMPOSIUM PRAGUE 22-24 MAY 2025

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A rare Genodermatosis Case report of Dyschromatosis universalis hereditaria

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Introduction :

Dyschromatoses are a group of disorders characterized by the presence of symmetrical distribution of small irregular hyperpigmented and hypopigmented macules. It is a spectrum of diseases which includes dyschromatosis universalis hereditaria (DUH), dyschromatosis symmetrica hereditaria (DSH), a segmental form called unilateral dermatomal pigmentary dermatosis (UDPD).

Case Report:

A 20 yr old male presented with multiple disseminated hyperpigmented and hypopigmented macules all over the body since 10yrs of age. was no history of any photosensitivity,usage of topical and systemic medications, no family history,no history of consanguinity among the parents.The lesions started from the trunk and both arms and gradually spread towards extremities with increase in number.The lesions were distributed to over the trunk,neck,both upper and lower limbs and face and with finger and toe nails dystrophy and pterygium formation and mucous membranes were spared and systemic examination didn't reveal any abnormalities.A routine laboratory investigations were within normal limits. Serology was negative. A skin biopsy from hyperpigmentary lesions showed Hyperkeratosis,basal layer hyperpigmentation,melanin incontinence and hypopigmentation lesions showed decrease in pigmentation and dermis showed lymphocytic infiltration.**Exome sequencing** revealed ABCB6(-),location Exon15,variant c.2029A>G. Based on the clinicopathological correlation and gene sequencing,the diagnosis of DUH was finally made. The patient was informed of the benign course of the disease and the possible unsatisfactory response to treatment options

Conclusion:

Our case highlights the clinical presentation of DUH, emphasizing the importance of clinical examination and histopathological correlation for accurate diagnosis. While DUH is primarily a benign condition, its differentiation from other pigmentary disorders, such as dyschromatosis symmetrica hereditaria, xeroderma pigmentosum, and reticulate pigmentary disorders, is crucial.

EA DV SYMPOSIUM PRAGUE 22-24 MAY 2025

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A Case of Classic Epidermodysplasia Verruciformis with Elephantiasis: An Atypical Presentation

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Introduction & Objectives:

Epidermodysplasia verruciformis (EV) is a rare genetic dermatological disorder characterized by an abnormal susceptibility of the skin to human papillomavirus infections. EV typically manifests before the age of 20, with no sex predilection. The disease carries a significant risk of cutaneous dysplasia and malignant transformation, particularly into squamous cell carcinoma.

We report the case of an immunocompetent female patient with epidermodysplasia verruciformis presenting with an atypical clinical manifestation associated with elephantiasis.

Materials & Methods:

We report the case of a 30-year-old female with no medical history presented with multiple raised cutaneous lesions. Dermatological examination revealed numerous verrucous lesions of varying sizes. These lesions appeared hypopigmented on the face and hyperpigmented on the rest of the body, predominantly affecting the extremities. Additionally, elephantiasis of the right lower limb had been present since the age of five. The patient reported no family history of similar cases or consanguinity.

Histopathological examination of a skin biopsy revealed hyperacanthosis, papillomatosis, hyperkeratosis, focal parakeratosis, elongated epidermal ridges, and thickening of the granular layer with large keratinocytes containing irregular, compact keratohyalin granules, consistent with verruca vulgaris.

The clinical and histological findings confirmed the diagnosis of epidermodysplasia verruciformis.

Comprehensive immunodeficiency screening, including HIV serology, was negative. A pre-treatment workup revealed no abnormalities, and the patient was started on acitretin at an initial dose of 10 mg/day.

Results:

Epidermodysplasia verruciformis, also known as “Tree Man Syndrome,” is a rare autosomal recessive genodermatosis associated with mutations in the EVER1/TMC6 and EVER2/TMC8 genes.

Patients with EV develop persistent and widespread cutaneous lesions due to specific HPV infections, particularly HPV-5 and HPV-8, which can lead to squamous cell carcinomas, especially in sun-exposed areas. Histologically, EV lesions resemble flat warts, with mild hyperkeratosis, hypergranulosis, and epidermal acanthosis. The upper epidermal keratinocytes appear enlarged, with vacuolated nuclei, pale blue-gray cytoplasm, and abundant basophilic keratohyalin granules.

Various treatments, including cryotherapy, topical imiquimod, 5-fluorouracil, oral retinoids, interferon-alpha, and photodynamic therapy, have been explored. Recent advances, particularly HPV vaccination, offer potential therapeutic avenues for this rare and often treatment-resistant condition. Surgical excision remains the primary approach for managing squamous cell carcinoma.

Our patient’s case exemplifies the key features of EV, with the absence of a reported family history potentially attributable

to incomplete penetrance or a de novo mutation. Diagnosis relies on clinical features, including characteristic cutaneous lesions, and histopathological findings, which, in our case, were more suggestive of verruca vulgaris than the commonly reported flat wart-like presentation.

Conclusion:

EV is a rare dermatological disorder associated with an increased susceptibility to specific HPV infections. Diagnosis is based on clinical examination, histopathology, and genetic studies. Current treatments remain symptomatic, with no definitive cure.