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EDITORIAL

# What is changing in medical mycology?

## ¿Qué está cambiando en la micología médica?

*Alexandro Bonifaz** and *Javier Araiza*

*Servicio de Dermatología, Micología, Hospital General de México "Dr. Eduardo Liceaga," Mexico City, Mexico*

Medical mycology as an infectious-disease field is influenced by a range of factors, which raises several questions: What is changing in mycoses? Is there a real increase, or are we simply diagnosing more? Are patterns of fungal disease shifting? What is happening with antifungal resistance across different fungi? These and other issues are posed in this editorial; while many lack definitive answers, they are left for the reader's consideration.

Undoubtedly, one of the shifts we are observing involves the effects of climate change. Although this phenomenon has been studied across many disciplines, mycology remains comparatively underexplored. In 2022, a book titled *The Impact of Climate Change on Fungal Diseases*, led by Frías-de-León et al.[1], was published in Mexico by Springer – one of the first international compendia to provide broader and more precise information. Among all mycoses, which ones are already changing? In our view, the first is coccidioidomycosis, fundamentally a pulmonary disease but one with major dermatologic impact. We have seen a significant rise in reports, although under-reporting and non-reporting remain problematic because notification is not mandatory – except in the state of Sonora (Mexico). Many communications suggest that sandstorms increase case numbers, but recent investigations indicate the opposite; rather, hydroclimatic fluctuations drive increases. We are now regularly seeing "super-rainstorms" that churn water and soil, prolonging environmental exposure to the fungus. Thus, climate-change–related shifts in coccidioidomycosis are a reality[2].

The second disease worth highlighting is histoplasmosis, also primarily pulmonary, but with increasingly frequent cutaneous manifestations among people living with HIV/AIDS. With rising global temperatures, *Histoplasma capsulatum*(s.l.) is being established and isolated more often in areas not considered tropical or bat-associated. A decade ago, disseminated cutaneous histoplasmosis was considered rare; it can now be regarded as an AIDS-defining illness. This is another clear example of how warming fosters fungal emergence across diverse geographic zones, including major cities[3].

Human coexistence with pets is ancient – most commonly dogs and cats – which is why zoophilic dermatophytes such as *Microsporum canis* have long been linked to ringworm. However, people periodically bring new animals into the home in attempts to domesticate them. Two to three decades ago there was a rise in pets such as guinea pigs and hamsters, which brought with them variants of *Trichophyton mentagrophytes* that caused highly inflammatory tinea – not because the fungi were more virulent but because humans recognize their antigenic variants less effectively. More recently we have seen *Trichophyton erinacei* – another variant of the same complex and part of hedgehog microbiota – causing tinea of the hand and other sites. Why is this increasingly common? A worldwide K-pop trend popularized hedgehogs as pets after certain groups showcased them, leading many fans – mostly adolescents – to purchase hedgehogs and, in turn, to a surge of inflammatory tinea of the hands and

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elsewhere[4]. Some European countries promptly banned breeding and sale of these animals, but in many Latin American countries, including Mexico, sales remain unrestricted – even via online platforms. In time this will wane, only to be replaced by new pets with their own microbial shifts.

Another major issue is rising antifungal resistance across fungi. Fluconazole – one of the most widely used agents for candidiasis – was employed indiscriminately, often in short courses and heavily advertised (even on public transport). Over time this induced resistance in multiple *Candida* species, with growing azole resistance in *Candida parapsilosis* and the emergence of the highly virulent, multidrug-resistant *Candida auris*. Moreover, triazoles have been used in the food industry and for mold and plant control; although not identical to human drugs, they share the triazole core, enabling selection of fungi with acquired resistance even before clinical use (e.g., *Cyp51A* mutations that alter the triazole-binding site). This is strikingly evident in systemic mycoses such as aspergillosis, where resistance in Europe has reached up to 20%.

Terbinafine, an allylamine introduced in the early 1990s, quickly became a first-line option – especially against *Trichophyton* species, the most common dermatophytes in tinea. Recently, however, terbinafine resistance has risen. It is now linked to mutations in the squalene epoxidase gene, the key target enzyme, with increased MICs in species such as *T. mentagrophytes* and *T. interdigitale*, and the emergence of the fully resistant *Trichophyton indotineae*. How many more resistant species will appear in the years ahead?[5].

Antimicrobial resistance is a race against time – driven by constant, sometimes indiscriminate use, poor prescribing, or prolonged courses. Resistance is inevitable; the greater problem is that the pipeline for new agents is sparse and slow.

## References


1. Frías-De-León MG, Brunner-Mendoza C, Reyes-Montes MR, Duarte-Escalante E, editores. The impact of climate change on fungal diseases. Cham, Switzerland: Springer; 2022.
2. Camponuri SK, Heaney AK, Cooksey GS, Vugia DJ, Jain S, Swain DL, et al. Recent and forecasted increases in coccidioidomycosis incidence linked to hydroclimatic swings, California, USA. Emerg Infect Dis. 2025;31:1028-32. doi: 10.3201/eid3105.241338.
3. Bonifaz A, GarcíaSalazar E, FríasDe-León. Climate change exacerbating fungal disease disparities. Curr Fung Infect Reports. 2024;18:1-12. doi. org/10.1007/s12281-023-00479-9
4. Bonifaz A, Lumbán-Ramírez P, Frías-de-León MG, Vidaurri de la Cruz H. Tineas caused by hedgehogs due to Trichophyton erinacei an ascending agent of dermatophytosis. Expert Rev Anti Infect Ther. 2024;22:721-4. doi: 10.1080/14787210.2024.2378980.
5. Mori Y, Yamada T, Ban S, Yoshioka I, Yaguchi T. Survey on the prevalence of terbinafine-resistant Trichophyton spp. with squalene epoxidase mutations. Med Mycol J. 2025;66:125-30. doi: 10.3314/mmj.25-00009.

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ORIGINAL ARTICLE

# Social media platform videos as a source of information in the Spanish language for patients with atopic dermatitis: an observational study and concordance analysis

## *Redes sociales como fuente de información en español para paciente con dermatitis atópica: un estudio observacional con análisis de concordancias*

*Tatiana V. Camayo-Vásquez[1,2]*, Daniela Melendrez-Vásquez[1,2], Óscar Muñoz-Velandia[1,3], Daniel G. Fernández-Ávila[1,4], and Daniela Chaparro-Reyes[1,2]*

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## Abstract

**Background:** *Patients and caregivers increasingly rely on social media platforms (SMPs) for health-related information, including content about dermatoses, such as atopic dermatitis (AD).* **Objective:** *To evaluate the reliability, completeness, and quality of Spanish-language videos about AD available on SMPs.* **Materials and methods:** *We conducted an analytical observational study analyzing Spanish-language videos from three SMPs: Instagram®, Facebook®, and YouTube®. General characteristics, user interactions, and source types were described. Standardized tools were used to assess reliability (modified DISCERN), completeness, and overall quality. Two independent evaluators reviewed all videos, and inter-rater reliability was assessed. A total of 300 videos were included, with 100 from each platform.* **Results:** *The median number of views was 6,804 (interquartile range 1,098.3-33,300), and the average duration was 2.18 min. Most videos were generated by independent users. Overall, 79.4% of the videos were rated as reliable sources of information, with Instagram having the highest proportion of reliable videos (84%). Videos on Instagram also showed a significantly higher popularity index. Inter-rater reliability (Cohen's Kappa) was nearly perfect for all tools used (k > 0.8). Findings suggest that Spanish-language SMP videos on AD generally have good reliability and quality, with slightly higher reliability observed in videos produced by academic organizations and independent users, particularly dermatologists. Most patient-perspective videos were also rated as useful.* **Conclusions:** *Instagram was found to be the most popular platform with the highest popularity index, likely due to the shorter length of its videos.*

***Keywords:*** *Atopic dermatitis. Eczema. Social media platforms. Observational study.*

## Resumen

**Antecedentes:** *Los pacientes y cuidadores utilizan las plataformas de redes sociales (PRSs) como fuente de información sobre diversos temas relacionados con la salud, incluyendo algunas dermatosis como la dermatitis atópica (DA).* **Objetivo:** *Evaluar la confiabilidad, exhaustividad y calidad de los videos en español sobre DA disponibles en PRSs.* **Material y métodos:** *Se realizó un estudio observacional analítico que incluyó videos en español de tres PRSs: Instagram®,*

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*Facebook® y YouTube®. Se describieron las características generales, las interacciones de los usuarios y los tipos de fuente. Se utilizaron herramientas estandarizadas para evaluar la confiabilidad (DISCERN modificado), la exhaustividad y la calidad general (GQS). Dos evaluadores independientes revisaron todos los videos y se evaluó la confiabilidad interevaluador. Se incluyeron un total de 300 videos, 100 de cada plataforma.* **Resultados:** *El promedio de vistas fue de 6,804 (RIC 1,098.3–33,300), y la duración promedio fue de 2.18 minutos. Los usuarios independientes generaron la mayoría de los videos. En general, el 79.4% de los videos fueron calificados como fuentes confiables de información, siendo Instagram la plataforma con mayor proporción de videos confiables (84%). Los videos en Instagram también mostraron un índice de popularidad significativamente mayor. La confiabilidad interevaluador (Kappa de Cohen) fue casi perfecta para todas las herramientas utilizadas (k > 0.8). Los hallazgos sugieren que los videos en PRSs en español sobre DA presentan en general buena confiabilidad y calidad, con una confiabilidad ligeramente mayor en videos producidos por organizaciones académicas y usuarios independientes, en particular dermatólogos. La mayoría de los videos con perspectiva del paciente también fueron considerados útiles.* **Conclusiones:** *Se encontró que Instagram fue la plataforma más popular con el índice de popularidad más alto, probablemente debido a la menor duración de sus videos.*

***Palabras clave:*** *Dermatitis atópica. Eccema. Plataformas de redes sociales. Estudio observacional.*

## Introduction

Atopic dermatitis (AD) is the most common chronic dermatological disease in children and adolescents[1]. AD can significantly impact the quality of life of patients and caregivers[2]. Due to the burden this disease carries for the patients and caregivers, most will seek alternative sources of information, such as support groups, communities, and unconventional treatments, among other resources[3].

In the era of networking through social media, patients and caregivers increasingly take advantage of the readily available information found on these platforms. Social media platforms (SMPs), such as Facebook, YouTube, and Instagram have taken on a role as decision-making aids across many areas of daily life, including health-related matters. This is particularly significant given that patients and caregivers may turn to SMPs for health advice even before consulting a healthcare professional.

A cross-sectional study performed by Marar et al.[4] in Saudi Arabia found that nearly 80% (78.3%) of adults aged 18-39 sought health information through social media, with approximately 28% using SMPs for direct medical consultations. These findings align with similar studies in the United States and Colombia, where around 60% and 45% of patients and caregivers reported seeking health information on SMPs[5,6].

In a 2022 U.S.-based study by Rowe et al.[7], 210 patients diagnosed with atopic dermatitis (DA) and their caregivers were evaluated. The study revealed that most web pages accessed lacked scientific validation. Notably, half of the respondents believed that YouTube videos provided more detailed information than that offered by specialist physicians. As Finnegan et al.[8] reported, patients and caregivers also tend to seek information on controversial topics, such as "corticophobia," a fear of steroid treatments, widely circulated on SMPs, and potentially harmful. In 2018, Freemyer et al. assessed 211 YouTube videos and found that those centered on patients' personal experiences with DA received the highest number of views[9].

This study aims to evaluate the information found in Spanish-language videos on Instagram, YouTube, and Facebook using standardized evaluation tools to determine their quality, reliability, and completeness.

## Methods

A cross-sectional analytical observational study was conducted, with concordance analysis included. We evaluated the content and quality of Spanish-language AD videos available on three SMPs: YouTube, Instagram, and Facebook. Duplicate videos were excluded. The study was approved by the Research and Institutional Ethics Committee of the Pontificia Universidad Javeriana.

### *Data extraction*

We used naïve accounts and incognito mode in the Google Chrome web browser to reduce the risk of bias based on previous searches. The search was conducted on April 8 and 9, 2024, and included all videos from the three SMPs. The Spanish terms used in the search process were "atopic dermatitis" and "atopic eczema." No filters were applied. Since previous studies have shown that 90% of users consume only the information displayed on the first three pages of search results on each SMP, only the first 100 videos from

each platform were selected[10], only the first 100 videos from each platform were selected. We reviewed the first pages of each platform's search results until we reached 100 items/platform. Items were excluded if they were off-topic and appeared due to paid sponsorships from unrelated brands. We also excluded reuploads from non-original sources, content lacking verbal or written information, and material in languages other than Spanish that appeared mistakenly in the Spanish-language search. In these cases, the search continued until the target of 100 items/platform was met. No exclusions were made based on video length; included content ranged from a few seconds to an hour (Fig. 1).

Data extraction was performed by one of the researchers (TCV) and included general information, such as the Uniform Resource Locator, publication date, video duration, number of views, number of "likes," and source of origin. All data were collected on the same date the search was conducted and recorded in Microsoft Excel (Version 16.85).

"Time on the Internet" or "time online" was defined as the number of days between the video's publication and the date of data extraction[11]. The popularity index was defined as the "number of views" divided by the "time on the Internet" or "time online." Videos were categorized based on their source into one of the following: (1) independent users, (2) government or news agencies, (3) professional organizations/academic channels (e.g., universities), (4) health information websites, or (5) advertisements/for-profit companies (e.g., pharmaceutical companies).

Videos were also classified into the following categories: "reliable for health personnel," "misleading for health personnel," "reliable opinion for the patient," and "misleading opinion for the patient." This classification considered the intended audience and an overall assessment of the content. A video rated as a "reliable or trustworthy opinion" contained scientifically accurate information about any aspect of the disease, while a "misleading opinion" rating indicated scientifically unproven or inaccurate content.

## Evaluation of information

The selected videos were evaluated independently by two researchers (TCV, DMV), both dermatology resident physicians. The reliability, completeness, and quality of the videos were assessed using standardized tools[11-16].

Reliability was defined as the presentation of correct data and accurate information on any aspect of the disease, from a scientific point of view. The modified 5-point DISCERN tool, a scale that evaluates five key aspects: clarity, reliability, balance, referencing, and discussion of risks and benefits, was used[17]. This assessment includes five questions and is scored from 0 to 5 based on the criteria of the reviewers, these scores were defined as follows: 0-1: lacks clarity, unreliable, biased, no references, omits risks/benefits, 2: major shortcomings in most criteria, 3: moderate quality, partial fulfillment of criteria, 4: mostly clear, reliable, referenced, and balanced, and 5: fully meets all five criteria with high quality and transparency.

Completeness assesses whether the video addresses all pre-defined essential topics, based on the presence or absence of key information[18]. This was assessed using a content score with five domains, which scored from 0 to 5, these scores were defined as follows: 0: none of the key topics addressed, 1-2: minimal coverage of essential content, 3: moderate inclusion of pre-defined topics, 4: most essential topics included, 5: fully comprehensive; all key elements clearly addressed.

Quality was defined as the usefulness of the information presented to a patient. This was assessed using the global quality score (GQS) tool, a 5-point subjective scale measuring the overall quality, flow, and usefulness of health-related content for the general public, which scored from 1 to 5 in the global quality ratin; these scores were defined as follows: 0-1: poor quality, low flow, not useful, 2: generally poor, limited use, 3: moderate quality, some value, 4: good quality, useful and mostly clear, and 5: excellent quality, highly useful and well-structured[11-16].

## Statistical analysis

Qualitative variables are reported as absolute and relative frequencies. Quantitative variables, including reliability (DISCERN), completeness (content score), and quality (GQS) scores, are reported as means and standard deviations when normally distributed; otherwise, they are presented as medians and interquartile ranges [IQRs]. Normality was assessed using the Shapiro-Wilk test.

Concordance between raters was determined by dichotomizing the scores from each of the three tools into two categories: "good or superior" (scores of 3-5) and "poor" (scores of 1 or 2). Cohen's Kappa coefficient was calculated for each tool to assess interrater reliability, showing near-perfect agreement across all scoring systems. Specifically, reliability scores (based on the DISCERN tool) had 95% agreement (Kappa = 0.88,

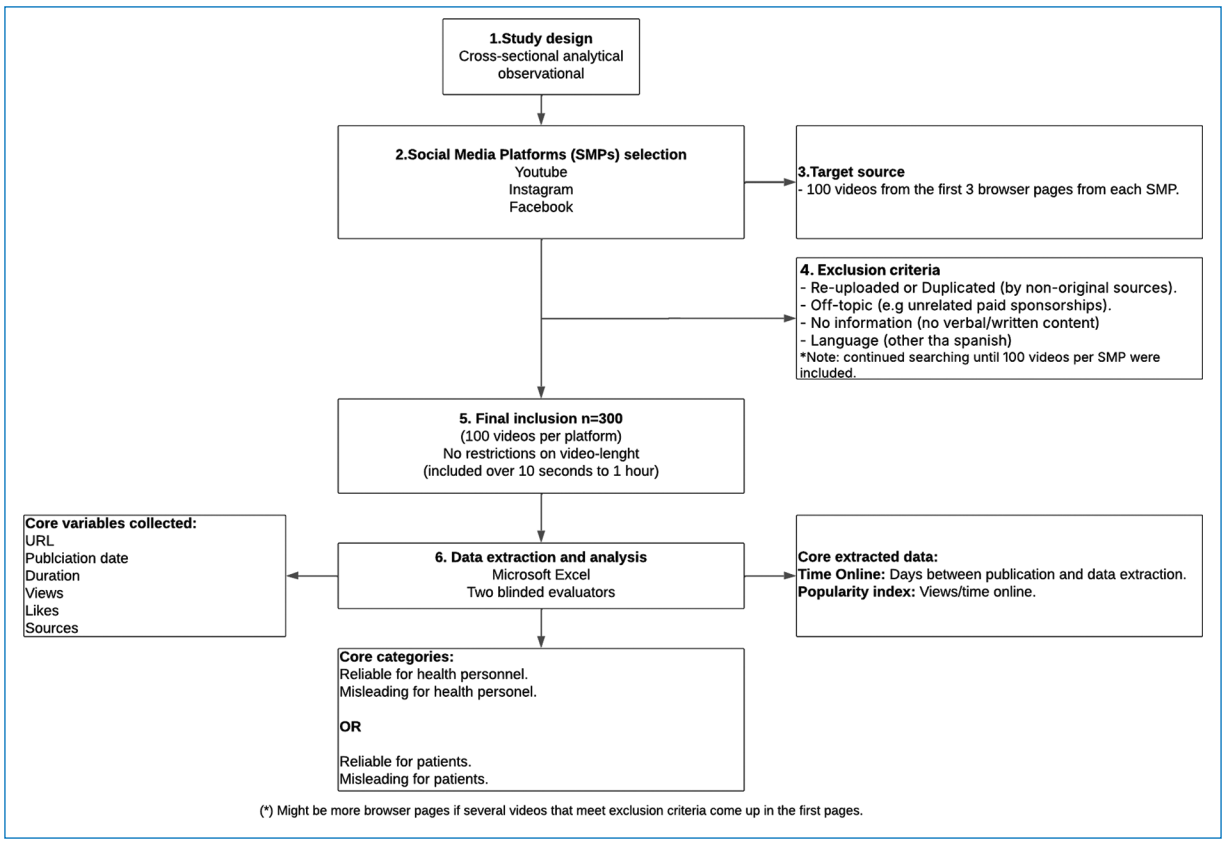


**Figure 1.** Video selection process for Spanish-language atopic dermatitis (AD) videos on YouTube, Facebook, and Instagram.

95% confidence interval [CI]: 0.5-1), completeness scores (based on the content score) had 97% agreement (Kappa = 0.93, 95% CI: 0.5-1), and quality scores (based on the GQS) had 98% agreement (Kappa = 0.96, 95% CI: 0.5-1).

Group comparisons were performed using chi-square tests for categorical variables and non-parametric tests (Mann-Whitney U or Kruskal-Wallis for multiple comparisons) for continuous variables. A $p < 0.05$ was considered statistically significant. Statistical analyses were conducted using Stata software (version 16.1, StataCorp LLC, College Station, TX).

## Results

A total of 300 videos were included. 100 from each SMP: Instagram, Facebook, and YouTube. The general characteristics are summarized in table 1. The median number of views was 6,804 (IQR: 1,098-33,300), with a median duration of 2.18 min (IQR: 0.93-7.44) and a median time online of 573 days (IQR: 210-1,372.5). Most videos were created and published by independent users (55.33%), including patients, caregivers, and healthcare professionals, primarily dermatologists, with private practices and professional social media accounts (Fig. 2).

The median scores were as follows: DISCERN 3 (IQR: 2-4), completeness (content score) 3 (IQR: 2-4), and quality (GQS) 3 (IQR: 2-4). In addition, most of the videos (61%) aimed at patients were considered useful.

### *Facebook versus YouTube versus Instagram*

A comparative analysis of the three SMPs showed that Instagram had the highest number of views, shortest video durations, the least time online, the highest number of likes, and the highest popularity index (median 101.8; IQR: 24.8-550.6), compared to YouTube (14.1; IQR: 1.9-75.6) and Facebook (1.42; IQR: 0.36-8.08) ($p < 0.001$).

In terms of content assessment, Instagram had a slightly higher proportion of videos rated as reliable

**Table 1.** General characteristics and evaluation of information in videos in Spanish about atopic dermatitis in social media platforms

| Variable | All (n = 300) | YouTube (n = 100) | Facebook (n = 100) | Instagram (n = 100) | p |
|---|---|---|---|---|---|
| Audience parameters, median (IQR) | | | | | |
| Number of reproductions | 6804 | 13824 (1325-89504.5) | 1175 (278-5650) | 14350 (3753.5-46850) | < 0.001* |
| Duration (minutes) | (1098-33300) | 4.88 (2.22-13.65) | 4.26 (1.46-11.11) | 0.8 (0.38-1.22) | < 0.001* |
| Time on the internet or online (days) | 2.18 (0.93-7.44) | 1116 (758.5-1994) | 911.5 (289-1883) | 206 (96-330) | < 0.001* |
| Popularity rating (number of views/ internet or online time) | 573 (210-1372.5) 17.35 (1.45-101.8) | 14.1 (1.9-75.6) | 1.42 (0.36-8.08) | 101.8 (24.8-550.6) | < 0.001* |
| Source of information, n (%) | | | | | 0.0001* |
| Independent users | 166 (55.33) | 34 (34.00) | 52 (52.00) | 80 (80.00) | |
| Government/News | 16 (5.33) | 6 (6.00) | 10 (10.00) | 0 (0.00) | |
| Professional organizations/ academic channels | 43 (14.33) | 30 (30.00) | 12 (12.00) | 1 (1.00) | |
| Health Information Websites4 | 25 (8.33) | 14 (14.00) | 10 (10.00) | 1 (1.00) | |
| Ads/For-Profit Companies5 | 50 (16.67) | 16 (16.00) | 16 (16.00) | 18 (18.00) | |
| Number of likes | 124.5 (23.5-511.5) | 210.5 (28-950.5) | 25 (8-88) | 283.5 (89-922.5) | < 0.001* |
| Number of dislikes | 0 (0-0) | 0 (0-0) | 0 (0-0) | 0 (0-0) | < 0.001* |
| Reliability, n (%) | | | | | 0.215 |
| "Reliable" | 238 (79.4) | 77 (77.00) | 77 (77.00 | 84 (84.00) | |
| "Misleading" | 62 (20.6) | 23 (23.00) | 23 (23.00) | 16 (16.00) | |
| Patients' perspective usefulness, n (%) | | | | | 0.001 |
| "Useful" | 36 (61.0) | 9 (9.00) | 4 (4.00) | 23 (23.00) | |
| "Futile" | 23 (39.8) | 4 (4.00) | 7 (7.00) | 12 (12.00) | |
| Assessment scales | | | | | |
| DISCERN, median (IQR) | 3 (2-4) | 3 (2.25-4) | 3 (2-4) | 3 (2-4) | 0.566 |
| "Good," n (%) | 218 (72.6) | 75 (75.0) | 72 (72.0) | 71 (71.00) | 0.804 |
| Completeness, Median (IQR) | 3 (2-4) | 3.75 (2-4.75) | 3 (2-4) | 3 (1-4) | 0.050 |
| "Good," n (%) | 223 (74.33) | 76 (75.0) | 75 (75.00) | 72 (72.00) | 0.797 |
| GQS, median (IQR) | 3 (2-4) | 3 (2.25-4) | 3 (3-4) | 3 (2-4) | 0.695 |
| "Good," n (%) | 224 (74.67) | 78 (34.8) | 75 (33.4) | 71 (31.7) | 0.523 |

*Statistically significant values.
DISCERN: DISCERN instrument (quality criteria for consumer health information); GQS: global quality score; RIC: interquartile range; IQR: interquartile rang.

compared to Facebook and YouTube (84% vs. 77% vs. 77%, respectively), though the difference was not statistically significant. No significant differences were observed in DISCERN scores (YouTube: median 3, IQR 2.25-4; Facebook: median 3, IQR 3-4; Instagram: median 3, IQR 2-4) or GQS scores (YouTube: median 3, IQR 2-4.75; Facebook: median 3, IQR 3-4; Instagram: median 3, IQR 1-4). YouTube videos had slightly better completeness scores (median 3.73; IQR 2.25-4) compared to the others (p = 0.050) (Table 1).

## *Videos by source*

The majority of the videos (n = 166) were produced by independent users (Table 1). Significant differences were found in the number of views per source (p < 0.005), with videos from health and education websites receiving the highest median number of views (11,345; IQR 468-144,906), followed by independent users (median 10,705; IQR 1,283-43,400).

Videos from professional organizations/academic channels and government or news agencies were significantly longer (professional organizations: median 6.8 min, IQR 2.1-13.75; government/news: median 6.6 min, IQR 2.09-10.46; p < 0.001). Time online was longest for videos from health information websites (median 1,224 days; IQR 941-2,040; p < 0.005).

Independent users' videos had the highest median number of likes (62; IQR 9-620; p < 0.001) and the highest popularity index (26.8; IQR 2.2–161.7), followed by videos from non-profit organizations, as shown in table 2.

According to DISCERN ratings, 88.37% of videos from professional organizations/academic channels were rated as "good," followed by government/news agencies (75%) and independent users (72.89%). Videos from for-profit companies scored significantly lower, with only 58% rated as "good" (p = 0.029).

In terms of completeness, 93.02% of professional organization videos were considered "good," along with

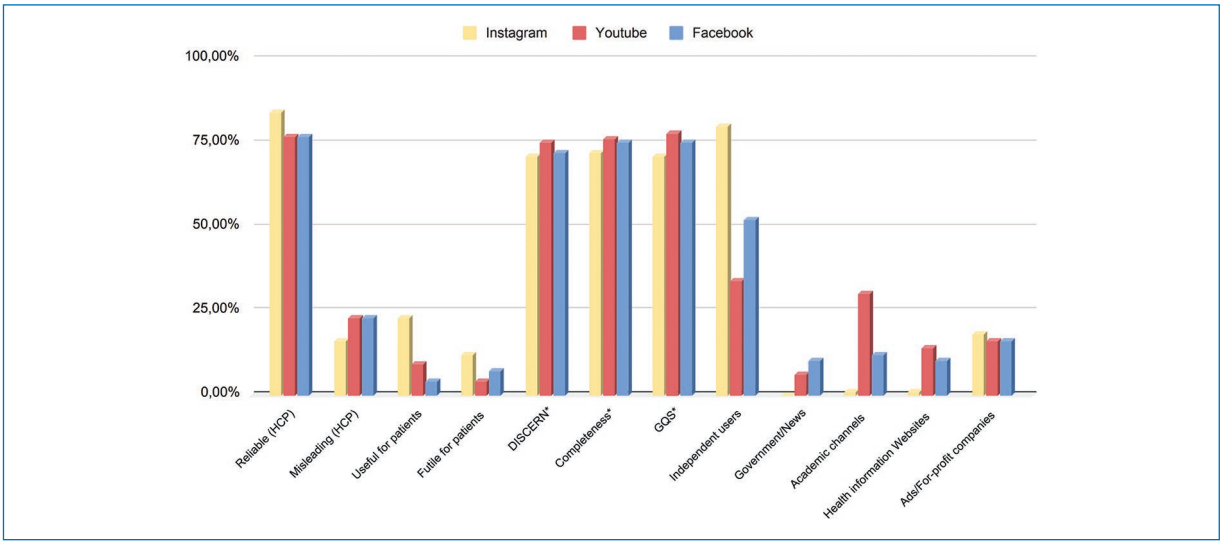


**Figure 2.** Frequency of core findings by source: reliability, patient perspective, and acceptability across scoring tools (GQS, DISCERN, and Completeness) applied to Spanish-language atopic dermatitis videos on social media platforms. *Acceptable ("good") score according to each scoring tool (DISCERN, GQS, and completeness assessment); HCP: health care professional perspective; DISCERN: DISCERN instrument (quality criteria for consumer health information); GQS: global quality score.

over 70% of videos from independent users. For-profit company advertisements had the lowest completeness score (62%) (p = 0.017). Similarly, the GQS ratings showed that while most sources scored above 70%, videos from for-profit companies again had significantly lower scores (60%) (p = 0.015) (Table 2).

### *Videos by reliability*

Based on the reliability assessment (Table 3), 69.3% (n = 238) of the videos were classified as reliable. There were no significant differences in views, duration, time online, popularity index, or number of likes between reliable and unreliable videos.

However, when applying the assessment tools, statistically significant differences were found. Reliable videos had significantly better scores than unreliable ones across all scales: DISCERN: median 3.5 (IQR 3-4) versus 1 (IQR 0.5-1.5), p < 0.0001, Completeness: median 3 (IQR 2-4) versus 0.5 (IQR 0-2), p < 0.0001 and GQS: median 4 (IQR 3-4) versus 1 (IQR 1-2), p < 0.0001.

Finally, 19.6% (n = 59) of all videos were identified as patient-targeted, with 61% of these considered useful. Instagram had the highest percentage of reliable videos considered useful for patients (Table 1).

## Discussion

To date, several studies have been performed to assess the quality of the content of AD videos available in SMPs in English, though only a few have utilized standardized scoring systems, such as DISCERN[19]. At present, there is only one published study that examines Spanish-language AD videos[20]. To our knowledge, this is the first study to comprehensively assess the quality, reliability, and completeness of AD-related video content in Spanish across three major SMPs, Instagram, Facebook, and YouTube, using standardized evaluation tools.

Overall, our findings suggest that most videos demonstrated moderate to high reliability, quality, and completeness. Among the platforms, Instagram stood out with the highest number of views, the highest popularity index, and the shortest video durations and time online. This suggests that even when content quality is similar across platforms, Instagram's dynamic format and brevity (median duration: 0.8 min [IQR 0.3-1.2]) may make it more appealing to users. Notably, of the videos directly aimed at patients, 61% (n = 36) were considered useful, with Instagram hosting the majority of these (63.8%, n = 23).

Our findings align with those of Finnegan et al.[8], who observed that websites promoting alternative treatments and rejecting conventional care often

**Table 2.** Characteristics and evaluation of information in videos in Spanish about atopic dermatitis in social media platforms according to the source of information

| Variable | Independent users (n = 166) | Government/ news (n = 16) | Professional organizations/ academic channels (n = 43) | Health information websites (n = 25) | Announcements for-profit companies (n = 50) | p |
|---|---|---|---|---|---|---|
| Social media platforms, n (%) | | | | | | |
| Instagram | 80 (48.19) | 0 (0.00) | 1 (2.32) | 1 (4) | 18 (36) | 0.0001* |
| YouTube | 34 (20.48) | 6 (37.5) | 30 (69.76) | 14 (56) | 16 (32) | |
| Facebook | 52 (31.32) | 10 (62.5) | 12 (27.90) | 10 (40) | 16 (32) | |
| Audience parameters, median (IQR) | | | | | | |
| Number of reproductions | 10705 (1283-43400) | 2400 (576.5-2400) | 3088 (411-8488) | 11345 (468-144906) | 10284 (3300-33100) | 0.0047* |
| Video length, minutes | 1.5 (0.81-5.41) | 6.6 (2.09-10.46) | 6.8 (2.1-13.75) | 3.38 (2.18-7.53) | 1.23 (0.45-2.86) | 0.0001* |
| Time on the internet or online time, days | 405 (174-1067) | 655.5 (193.5-2098) | 822 (320-1309) | 1224 (941-2040) | 758.5 (205-1904) | 0.0011* |
| Popularity rating, number of views/internet or online time | 26.8 (2.2-161.7) | 4.06 (0.70-19.34) | 3.3 (0.72-14.36) | 10.8 (0.60-134.6) | 23.4 (2.71-110.8) | 0.0001* |
| Number of likes | 62 (9-620) | 28 (4-44) | 34.5 (7-161) | 20 (0-40) | 33.5 (9-76) | 0.0001* |
| Number of dislikes | 0 (0-0) | 0 (0-0) | 0 (0-0) | 0 (0-0) | 32 (64) | 0.0001* |
| Reliability, n (%) | | | | | | |
| Reliable | 137 (82.53) | 13 (81.25) | 38 (88.37) | 19 (76) | 29 (58) | 0.2101 |
| Unreliable | 29 (17.46) | 3 (18.75) | 5 (11.62) | 6 (24) | 21 (42) | |
| Assessment scales | | | | | | |
| DISCERN, median (IQR) | 3. 25 (2-4) | 3 (2-3.75) | 3 (3-4) | 3 (2-4) | 2.75 (1-3) | 0.013* |
| "Good", n (%) | 121 (72.89) | 12 (75.00) | 38 (88.37) | 18 (72.00) | 29 (58.00) | 0.029* |
| Completeness, average (IQR) | 3.25 (1.5-4) | 3.5 (2.5-4) | 3.5 (3-4) | 3.5 (2-5) | 3 (0.5-4) | 0.040* |
| "Good", n (%) | 122 (73.49) | 12 (75.00) | 40 (93.02) | 18 (72.00) | 31 (62.00) | 0.017* |
| GQS, mean (IQR) | 3. 75 (2-4) | 3 (3-4) | 4 (3-4) | 3 (3-4) | 3 (1-4) | 0.031* |
| "Good", n (%) | 122 (73.49) | 13 (81.25) | 39 (90.70) | 20 (80) | 30 (60.00) | 0.015* |

*Statistical significance at the $p < 0.05$ level.
DISCERN: DISCERN instrument (quality criteria for consumer health information); GQS: global quality score; IQR: interquartile range.

**Table 3.** General characteristics and evaluation of information in videos in Spanish about atopic dermatitis in social media platforms according to reliability

| Variable | p |
|---|---|
| Audience parameters, median (IQR) | |
| Number of reproductions | 0.700 |
| Duration (minutes) | 0.871 |
| Time on the internet or online time (days). | 0.950 |
| Popularity rating (number of views/internet or online time) | 0.884 |
| Number of likes | 0.446 |
| Assessment scales | |
| DISCERN, median (IQR) | 0.0001* |
| Mean (SD) | 0.0001* |
| "Good", n (%) | 0.0001* |
| Completeness, Median (IQR) | 0.0001* |
| Mean (SD) | 0.0001* |
| "Good", n (%) | 0.0001* |
| GQS, Median (IQR) | 0.0001* |
| Mean (SD) | 0.0001* |
| "Good", n (%) | 0.0001* |

*Statistical significance at the $p < 0.05$ level.
DISCERN: DISCERN instrument (quality criteria for consumer health information), IQR: interquartile range, SD: standard deviation.

coincided with commercial interests, particularly the marketing of herbal products. Similarly, videos in our study that dismissed evidence-based treatments were consistently rated as unreliable and of low quality[1].

In terms of scoring, 72.6% (n = 218) of all evaluated videos were rated as reliable based on DISCERN. This is in contrast to findings from studies on other medical conditions, such as rheumatoid arthritis and ulcerative colitis[11,12] as well as English-language AD videos. For example, Gorrepati et al.[19] analyzed 30 English YouTube videos using DISCERN and found that even dermatologist-associated content lacked comprehensive information, despite being relatively reliable. While their study had a smaller sample and focused exclusively on YouTube, our study included 300 Spanish-language videos across three platforms, offered content-based reliability scoring, and showed a similar level of interrater agreement (Cohen's Kappa = 0.88).

Iglesias-Puzas et al.[20] of Spanish-language AD content on social media, which similarly highlighted the prevalence and influence of such material but did not assess quality using standardized tools. Voillot et al.[21], analyzing French-language posts, found that patients used SMPs for both health advice and emotional support. Similarly, Mueller et al.[22], reviewing English-language YouTube videos, found only 32% to be useful, largely due to anecdotal or subjective content. While anecdotal content is often undervalued in structured assessments, it may serve an important role for users seeking community or shared experiences between patients and caregivers.

This perspective is supported by Martin et al.[23], who found that while professionally produced YouTube videos achieved higher reliability scores, patient-experience videos had greater engagement, particularly on emotionally charged topics, such as steroid withdrawal. These findings point to the dual role of SMPs in both spreading factual information and allowing patient connection.

Rowe et al.[7] further emphasized that patients are more likely to trust sources recommended by healthcare professionals. This underscores the need for clinicians to engage with social media and recommend high-quality sources, ideally verified medical professionals, such as dermatologists or pediatricians. Creating platform-specific visual aids and lists of reliable content creators may enhance patient education and trust.

In our study, most independent users were healthcare professionals. Dermatologists were the most frequent contributors, reinforcing the key role of medical professionals in online health education. To improve the reliability of SMP content, especially given the unregulated nature of these platforms, fact-checking strategies should be promoted. These include creator-side verification as well as emerging artificial intelligence tools capable of real-time content validation. Although these approaches have traditionally been applied in politics, they are now being explored in medicine as well[24].

Interestingly, no significant differences in video metrics (views, duration, likes) were found between reliable and unreliable videos, although the latter often had slightly higher popularity indices, likely due to engaging visuals and storytelling. Nevertheless, when assessed using validated tools, reliable videos consistently scored significantly higher in reliability, completeness, and overall quality ($p < 0.001$).

This study offers several strengths. It is the first to evaluate Spanish-language AD content across three major SMPs using standardized tools (DISCERN, GQS, content score), assess interrater reliability, and analyze 300 videos – a considerably larger sample than previous studies.

Nonetheless, limitations exist. Despite using validated tools, scoring still involved a degree of subjectivity. However, high interrater reliability (Cohen's Kappa) supports the consistency of our findings. In addition, the exclusion of newer platforms, such as TikTok, now widely used by patients and caregivers, may limit generalizability. At the time of our study, Instagram, Facebook, and YouTube were the dominant platforms for medical information. The cross-sectional design also captures content at a single point in time, which may not reflect the rapidly evolving nature of SMPs. Finally, while our evaluation focused on the medical perspective, incorporating patient-centered outcomes and perceptions will be essential in future research.

## Conclusion

Our study shows that Instagram is the preferred SMP for accessing healthcare information in Spanish on AD, closely followed by YouTube. The high quality of Instagram content is likely related to the fact that most videos on this platform are created by independent users, particularly dermatologists. However, we find most of the videos evaluated with the scoring tools (GQS, DISCERN, and completeness score) on the three SMPs had overall good quality.

In addition, useful content from the patient's perspective can be found on Instagram and should be recognized as a valuable source of information, networking, and support among patients and caregivers when the content is verified as trustworthy. There remains a need for further studies that include other widely used platforms, such as TikTok, especially since most existing research in English has focused solely on YouTube. We emphasize the importance of taking this information into account when guiding our patients in navigating health information on SMPs.

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**Protection of humans and animals.** The authors declare that no experiments involving humans or animals were conducted for this research.

**Confidentiality, informed consent, and ethical approval.** The study does not involve patient personal data nor requires ethical approval. The SAGER guidelines do not apply.

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## References

1. Moreno MA. Atopic diseases in children. JAMA Pediatr. 2016;170:96.
2. Eichenfield LF, Stripling S, Fung S, Cha A, O'Brien A, Schachner LA. Recent developments and advances in atopic dermatitis: a focus on epidemiology, pathophysiology, and treatment in the pediatric setting. Paediatr Drugs. 2022;24:293-305.
3. Ali F, Vyas J, Finlay AY. Counting the burden: atopic dermatitis and health-related quality of life. Acta Derm Venereol. 2020;100:adv00161.
4. Sumayyia MD, Al-Madaney MM, Almousawi FH. Health information on social media. Perceptions, attitudes, and practices of patients and their companions. Saudi Med J. 2019;40:1294-8.
5. Wang X, Cohen RA. Health Information Technology Use among Adults: United States, July-December 2022. United States: NCHS Data Brief; 2023. p. 1-8.
6. Bartels SM, Martinez-Camblor P, Naslund JA, Suárez-Obando F, Torrey WC, Cubillos L, et al. A characterisation of social media users within the primary care system in Colombia and predictors of their social media use to understand their health. Rev Colomb Psiquiatr (Engl Ed). 2021;50 Suppl 1:42-51.
7. Rowe G, Young GJ, Luu M. Sources of online information utilized by caregivers of pediatric patients with atopic dermatitis. Pediatr Dermatol. 2023;40:476-8.
8. Finnegan P, Murphy M, O'Connor C. #Corticophobia: a review on online misinformation related to topical steroids. Clin Exp Dermatol. 2023;48:112-5.
9. Freemyer B, Drozd B, Suarez A. A cross-sectional study of YouTube videos about atopic dermatitis. J Am Acad Dermatol. 2018;78(3):612-3.
10. iProspect. iProspect Blended Search Results Study; 2008. Available from: https://www.researchgate.net/publication/276454791 [Last accessed on 2023 Jul 26].
11. Singh AG, Singh S, Singh PP. YouTube for information on rheumatoid arthritis--a wakeup call? J Rheumatol. 2012;39:899-903.
12. Mukewar S, Mani P, Wu X, Lopez R, Shen B. YouTube and inflammatory bowel disease. J Crohns Colitis. 2013;7:392-402.
13. Qi J, Trang T, Doong J, Kang S, Chien AL. Misinformation is prevalent in psoriasis-related YouTube videos. Dermatol Online J. 2016 Nov 15;22(11).
14. Delli K, Livas C, Vissink A, Spijkervet FK. Is YouTube useful as a source of information for Sjögren's syndrome? Oral Dis. 2016;22:196-201.
15. Bernard A, Langille M, Hughes S, Rose C, Leddin D, Van Zanten SV. A systematic review of patient inflammatory bowel disease information resources on the world wide web. Am J Gastroenterol. 2007;102:2070-7.
16. Ng CH, Lim GR, Fong W. Quality of English-language videos on YouTube as a source of information on systemic lupus erythematosus. Int J Rheum Dis. 2020;23:1636-44.
17. Charnock D, Shepperd S, Needham G, Gann R. DISCERN: an instrument for judging the quality of written consumer health information on treatment choices. J Epidemiol Community Health. 1999;53:105-11.
18. Lopez-Olivo MA, Suarez-Almazor ME. Digital patient education and decision aids. Rheum Dis Clin North Am. 2019;45:245-56.
19. Gorrepati PL, Smith GP. DISCERN scores of YouTube information on eczema treatments. J Am Acad Dermatol. 2021;85:1354-5.
20. Iglesias-Puzas Á, Conde-Taboada A, Campos-Muñoz L, Belinchón-Romero I, López-Bran E. Social networks and atopic dermatitis: cross-sectional descriptive study. Actas Dermosifiliogr (Engl Ed). 2020;111:665-70.
21. Voillot P, Riche B, Portafax M, Foulquié P, Gedik A, Barbarot S, et al. Social media platforms listening study on atopic dermatitis: quantitative and qualitative findings. J Med Internet Res. 2022;24:e31140.
22. Mueller SM, Hongler VN, Jungo P, Cajacob L, Schwegler S, Steveling EH, et al. Fiction, falsehoods, and few facts: cross-sectional study on the content-related quality of atopic eczema-related videos on YouTube. J Med Internet Res. 2020;22:e15599.
23. Martin A, Thatiparthi A, Liu J, Wu JJ. Atopic dermatitis topical therapies: study of YouTube videos as a source of patient information. Cutis. 2021;108:139-41.
24. Xie Q, Schenck EJ, Yang HS, Chen Y, Peng Y, Wang F. Faithful AI in Medicine: A Systematic Review with Large Language Models and Beyond. Research Square [Preprint]; 2023.

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ORIGINAL ARTICLE

# Prevalence of acne and its impact on quality of life of students from a high school in Montevideo

## *Prevalencia de acné y su impacto sobre la calidad de vida de estudiantes de un liceo de Montevideo*

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## Abstract

**Background:** *Acne is highly prevalent during adolescence, with reported rates reaching up to 96%, being commonly associated with impaired health-related quality of life (HRQoL).* **Objective:** *To know the prevalence of acne in a sample of adolescents from Uruguay.* **Material and methods:** *This study assessed the prevalence of acne and its impact on HRQoL in adolescents aged 12 to 17 years, attending a high school in Montevideo. Students were examined and classified according to acne Severity. Those diagnosed with acne completed the Cardiff Acne Disability Index to assess HRQoL.* **Results:** *A total of 111 adolescents were evaluated (59 females and 52 males). The overall prevalence of acne was 91%, affecting 96.6% of females and 86% of males. Regarding clinical severity, 89% of cases presented mild or moderate acne. On the other hand, 78% of participants with acne showed some degree of HRQoL impairment.* **Conclusions:** *Acne is highly prevalent during adolescence and frequently affects the HRQoL, highlighting the need for comprehensive patient assessments and the adoption of a multidisciplinary therapeutic approach.*

***Keywords:*** *Acne. Health-related quality of life. Cardiff Acne Disability Index. Prevalence. Uruguay.*

## Resumen

**Antecedentes:** *El acné es muy frecuente durante la adolescencia, con reportes de prevalencia de hasta un 96%, siendo comúnmente asociado con compromiso de la calidad de vida relacionado con salud (CVRS).* **Objetivo:** *Conocer la prevalencia de acné en una muestra de adolescentes de Uruguay.* **Material y métodos:** *Se estudió la prevalencia de acné y su impacto en la CVRS en adolescentes de entre 12 y 17 años de un liceo de Montevideo. Los estudiantes fueron examinados y clasificados según grado de severidad de acné. Aquellos diagnosticados con acné contestaron el cuestionario de CVRS, Cardiff Acne Disability Index.* **Resultados:** *Se evaluaron 111 jóvenes (59 mujeres y 52 hombres). La prevalencia global de acné fue del 91%, presentándose en el 96.6% de las mujeres y el 86% de los hombres. Respecto de la severidad clínica, un 89% de los casos presentó acné leve o moderado. Por otro lado, un 78% de los jóvenes con acné evidenció algún grado de compromiso de CVRS.* **Conclusiones:** *El acné es muy prevalente durante la adolescencia y suele afectar la CVRS, hecho que demuestra la importancia de realizar una evaluación integral de nuestros pacientes, adoptando un enfoque terapéutico multidisciplinario.*

***Palabras clave:*** *Acné. Calidad de vida relacionada con la salud. Cardiff Acne Disability Index. Prevalencia. Uruguay.*

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## Introduction

Acne is a chronic, inflammatory, multifactorial dermatosis of the pilosebaceous unit that presents clinically with seborrhea and cutaneous lesions – comedones, papules, pustules, nodules, and scars – predominantly in seborrheic areas of the face, upper back, and chest[1-3].

It is highly prevalent during adolescence, as shown in several studies: a Nottingham study reported a 50% prevalence in 317 youths aged 14-16 years[4], a Singapore study reported 88% in 1045 adolescents aged 13-19 years[5], and a São Paulo study reported 96% in 452 adolescents aged 10-17 years[6].

Multiple scales classify acne severity; notably, the 2016[7] European Dermatology Forum system evaluates the face, back, and chest and uses 4 grades: grade 1, comedonal acne; grade 2, mild to moderate papulopustular acne; grade 3, severe papulopustular or moderate nodular acne; and grade 4, severe nodular acne or acne conglobata.

Acne can affect all areas of life – including work, school, social activities, and personal and sexual relationships – leading to marked impairment in health-related quality of life (HRQoL)[6,8-12], which the World Health Organization defines as "a multidimensional, subjective construct that includes positive and negative feelings, may change over time, and encompasses physical, psychological, and social aspects"[13].

HRQoL can be measured with generic or disease-specific tools; current practice favors disease-specific scales because of their greater sensitivity compared with generic measures[14,15].

The aim of this article is to report the first and only study of acne prevalence and its association with HRQoL impairment in a sample of Uruguayan adolescents.

## Methods

We conducted an observational, cross-sectional, analytic study inviting all students aged 12-17 years at a Montevideo secondary school to participate. Parents or legal guardians provided written informed consent authorizing participation, and adolescents provided written assent indicating willingness to participate. Both documents were required; youths without them were excluded.

Participants were assigned numeric codes to preserve anonymity, reported their age, and were examined by 2 dermatology resident physicians. Acne cases were graded by clinical severity using the European Dermatology Forum classification[7].

Adolescents with acne completed the Cardiff Acne Disability Index (CADI), validated for Uruguay[16], a disease-specific HRQoL instrument with 5 items, each with 4 response options scored 0-3 (total score 0-15). CADI categorizes HRQoL impairment as follows: 0, none; 1-5, mild; 6-10, moderate; 11-15, severe[17,18]. Item content targets different HRQoL domains: items 1-2 assess psychosocial consequences of acne; item 3 addresses psychosocial impact in patients with chest or back lesions; item 4 assesses psychological consequences; and item 5 captures patients' subjective self-assessment of severity.

CADI is the intellectual property of Cardiff University; we obtained a license for this study (registration No. CUQoL2562). The instrument has been translated and validated in multiple languages[19-26] and widely used in HRQoL-and-acne research[27-29], underscoring its relevance and value as a supportive diagnostic tool.

The study was approved by the Ethics Committee of the School of Medicine, *Universidad de la República*, and received the participating school's institutional endorsement.

Statistical analyses were performed using STATA version 12.0. Sociodemographic variables were summarized descriptively. Sample size for prevalence estimation used the Z statistic for proportions, with $\alpha = 0.05$ and an imprecision of 0.1. The association between age and acne presence was tested with the independent-samples Student t test, and associations between categorical variables were assessed with the chi-square $\chi^2$ test.

## Results

First, based on the school's academic records, 348 students aged 12-17 years were potentially eligible for inclusion. The minimum sample size required to estimate prevalence was calculated as at least 81 participants.

Between September 25 and November 25, 2019, a total of 111 adolescents were evaluated; all provided written assent, and their parents/guardians signed informed consent. Of those evaluated, 59 (53%) were female and 52 (47%) were men; the mean age was 13.8 years and the median was 13 years.

An overall acne prevalence of 91% (101 cases) was observed – 86% in men and 96.6% in women. There was a statistically significant association between female sex and the presence of acne ($p = 0.043$). However, this difference tended to disappear among

adolescents older than 14 years, in whom all evaluated individuals had acne.

Among the 101 adolescents with acne, severity was distributed as follows: grade 1 in 57%, grade 2 in 32%, and grade 3 in 11%; no grade 4 cases were observed. There were no significant differences in clinical severity between males and females (p = 0.590).

When acne presence was analyzed by age, those without acne had a mean age of 12.5 years, whereas those with acne had a mean age of 13.9 years; this difference was statistically significant (p < 0.001).

Among the 101 adolescents with acne who completed the CADI, health-related quality of life (HRQoL) impairment was identified in 78%: mild in 95% and moderate in 5%; no severe impairment was found. HRQoL impairment was present in 82.4% of women and 72.7% of men, a difference that was not statistically significant (p = 0.240).

Finally, a total of 67.2% of adolescents with grade 1 acne had HRQoL impairment (always mild), whereas all adolescents with grade 3 acne had HRQoL impairment, with 18.2% showing moderate impairment. Thus, there was a statistically significant association between acne severity and the level of HRQoL impairment (p = 0.002) (Table 1).

**Table 1.** Cross-tabulation of "clinical acne severity" and "health-related quality of life (HRQoL) impairment." A statistically significant association was found (p = 0.002)

| Clinical acne severity | HRQoL) impairment | | | |
|---|---|---|---|---|
| | Absent % (n) | Mild % (n) | Moderate % (n) | Total % (n) |
| Grade 1 | 32.8 (19) | 67.2 (39) | 0 | 57.4 (58) |
| Grade 2 | 9.4 (3) | 84.4 (27) | 6.2 (2) | 31.7 (32) |
| Grade 3 | 0 | 81.8 (9) | 18.2 (2) | 10.9 (11) |
| Total | 21.8 (22) | 74.3 (75) | 3.9 (4) | 100 (101) |

HRQoL: health-related quality of life.

## Discussion

The primary motivation for this study was to explore the epidemiology of acne in a Uruguayan cohort, underscoring the pioneering nature of this work in the country. The high acne prevalence observed closely aligns with prior literature[30-32].

With respect to severity, mild forms (grades 1-2) clearly predominated, and no grade 4 cases were identified, consistent with international series[3-5,30].

Although acne was more prevalent among females, this difference tended to disappear after age 14 – plausibly reflecting later pubertal timing in men[33].

For HRQoL assessment, we selected the CADI (Uruguay Spanish version)[16], given its wide international use[27-29], ease of application and interpretation, and disease specificity – features that confer greater sensitivity than generic questionnaires[14,15]. We found a high prevalence of HRQoL impairment associated with acne, which should prompt routine use of HRQoL tools and the development of multidisciplinary teams to deliver comprehensive, timely care. Item-level review showed no major abnormalities in psychosocial-consequences items (most responses scored 0 on items 1-3), whereas items 4 and 5 – probing psychological consequences and subjective severity – showed more responses scored 1-2. Thus, disability in this cohort appears driven mainly by psychological consequences and adolescents' self-perception of disease severity.

Participants with more clinically severe acne also showed greater HRQoL impairment, which is consistent with international data[29,34-36].

Finally, we note growing recognition of the link between acne and HRQoL deterioration – manifesting as shame, impaired socialization, anxiety, and depression[2,37-39] – which argues for early clinical intervention and routine incorporation of HRQoL measures into our routine clinical practice.

## Conclusions

Acne is very common in adolescence and is frequently associated with HRQoL impairment. Better understanding of acne's psychosocial consequences will help optimize treatment and foster multidisciplinary teams, enabling a comprehensive, integrative therapeutic approach that addresses all domains affecting patients and their families. These findings reinforce the need for early detection and treatment to mitigate HRQoL impact.

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**Confidentiality, informed consent, and ethics approval:** The authors followed institutional confidentiality protocols, obtained informed consent from participants, and received Ethics Committee approval. The SAGER guidelines were followed as appropriate to the nature of the study.

**Declaration on the use of artificial intelligence:** The authors declare that no generative artificial intelligence was used in the writing of this manuscript.

## References


1. Orentreich N, Durr N. The natural evolution of comedones into inflammatory papules and pustules. J Invest Dermatol. 1974;62:316-20.
2. Popovic A, Bojic MD, Djurovic M, Bojic M, Pjescic T. Impact of acne on the quality of life of adolescents in the territory of the Northern and Southern Regions of Montenegro. Iran J Public Health. 2024;53:2341-9.
3. Napolitano M, Ruggiero G, Monfrecola G, Megna M. Acne prevalence in 9 to 14-year-old old patients attending pediatric ambulatory clinics in Italy. Int J Dermatol. 2018;57:1320-3.
4. Smithard A, Glazebrook C, Williams HC. Acne prevalence, knowledge about acne and psychological morbidity in mid-adolescence: a community-based study. Br J Dermatol. 2001;145:274-9.
5. Tan HH, Tan AWH, Barkham T, Yan XY, Zhu M. Community-based study of acne vulgaris in adolescents in Singapore. Br J Dermatol. 2007; 157:547-51.
6. Bagatin E, Timpano D, Guadanhim L, Nogueira V, Terzian L, Steiner D, et al. Acne vulgaris: prevalence and clinical forms in adolescents from São Paulo, Brazil. An Bras Dermatol. 2014;89:428-35.
7. Nast A, Dréno B, Bettoli V, Bukvic Mokos Z, Degitz K, Dressler C, et al. European evidence-based (S3) guideline for the treatment of acne – update 2016 – short version. J Eur Acad Dermatology Venereol. 2016;30:1261-8.
8. Kruglova L, Samushiya M, Talybova A. Mental disorders, social maladaptation and quality of life of patients with acne and post-acne symptoms. Zh Nevrol Psikhiatr Im S S Korsakova. 2018;118:4-10.
9. Sule Afsar F, Seremet S, Demirlendi Duran H, Elif Yildirim F, Mumcu Sönmez N. Social appearance anxiety in adult patients with acne: a cross-sectional study. Acta Dermatovenerol Croat. 2018;26:220-5.
10. Williams H, Dellavalle R, Garner S. Acne vulgaris. Lancet. 2012; 379:361-72.
11. Yolaç Yarpuz A, Demirci Saadet E, Erdi Sanli H, Devrimci Ozgüven H. Social anxiety level in acne vulgaris patients and its relationship to clinical variables. Turk Psikiyatr Derg. 2008;19:29-37.
12. Bez Y, Yesilova Y, Kaya M, Sir A. High social phobia frequency and related disability in patients with acne vulgaris. Eur J Dermatol. 2011; 21:756-60.
13. The WHOQOL Group. World Health Organization Quality of Life Assessment. What quality of life? World Health Forum. 1996;17:354-6.
14. Patrick D, Deyo R. Generic and disease-specific measures in assessing health status and quality of life. Med Care. 1989;27:S217-32.
15. Bowling A. The effects of illness on quality of life: findings from a survey of households in Great Britain. J Epidemiol Commun Health. 1996; 50:149-55.
16. Turra N, Durán C, Otero G, Izuibejeres C, Otero C. Cardiff Acne Disability Index: validación cultural y lingüística al español de Uruguay. Med Cutan Iber Lat Am. 2024;52:75-81.
17. Motley R, Finlay A. Practical use of a disability index in the routine management of acne. Clin Exp Dermatol. 1992;17:1-3.
18. Shams N, Niaz F, Zeeshan S, Ahmed S, Farhat S, Seetlani NK. Cardiff acne disability index based quality of life in acne patients, risk factors and associations. J Liaquat Uni Med Heal Sci. 2018;17:29-35.
19. Jankovic S, Vukicevic J, Djordjevic S, Jankovic J, Marinkovic J, Basra MKA. The Cardiff Acne Disability Index (CADI): linguistic and cultural validation in Serbian. Qual Life Res. 2013;22:161-6.
20. Yap F. Cardiff acne disability index in Sarawak, Malaysia. Ann Dermatol. 2012;24:158-61.
21. Grando LR, Horn R, Cunha VT, Cestari TF. Translation, cultural adaptation and validation for Brazilian Portuguese of the Cardiff acne disability index instrument. An Bras Dermatol. 2016;91:180-6.
22. Gupta A, Sharma YK, Dash K, Verma S. Cultural adaptation of the Cardiff Acne Disability Index to a Hindi speaking population: a pilot study. Indian J Dermatol. 2015;60:419.
23. Dreno B, Finlay AY, Nocera T, Verriere F, Taieb C, Myon E. The Cardiff Acne Disability Index: cultural and linguistic validation in French. Dermatology. 2004;208:104-8.
24. Aghaei S, Mazharinia N, Jafari P, Abbasfard Z. The Persian version of the Cardiff Acne Disability Index. Reliability and validity study. Saudi Med J. 2006;27:80-2.
25. Law M, Chuh A, Lee A. Validation of a Chinese version of the Cardiff Acne Disability Index. Hong Kong Med J. 2009;15:12-7.
26. Chernyshov P. Creation and validation of the Ukrainian version of the Cardiff Acne Disability Index. Lik Sprav. 2012;(5):139-43.
27. Dreno B, Bordet C, Seite S, Taieb C. Acne relapses: impact on quality of life and productivity. J Eur Acad Dermatol Venereol. 2019;33:937-43.
28. Chernyshov P, Petrenko A, Kopylova V. What health-related quality of life factors influence the decision of patients with acne to visit a dermatologist? J Clin Aesthet Dermatol. 2018;11:21-5.
29. Walker N, Lewis-Jones M. Quality of life and acne in Scottish adolescent schoolchildren: use of the Children's Dermatology Life Quality Index (CDLQI) and the Cardiff Acne Disability Index (CADI). J Eur Acad Dermatol Venereol. 2006;20:45-50.
30. Bagatin E, Timpano DL, Guadanhim LR dos S, Nogueira VMA, Terzian LR, Steiner D, et al. Acne vulgaris: prevalence and clinical forms in adolescents from São Paulo, Brazil. An Bras Dermatol. 2014;89:428-35.
31. Dreno B, Poli F. Epidemiology of acne. Dermatology. 2003;206:7-10.
32. Gebauer K. Acne in adolescents. Aust Fam Physician. 2017;46:892-5.
33. Chulani VL, Gordon LP. Adolescent growth and development. Prim Care. 2014;41:465-87.
34. Gupta M, Gupta A. Dissatisfaction with skin appearance among patients with eating disorders and non-clinical controls. Br J Dermatol. 2001;145:110-3.
35. Cusumano D, Thompson J. Media influence and body image in 8-11-year-old boys and girls: a preliminary report of the multidimensional media influence scale. Int J Eat Disord. 2001;29:37-44.
36. Khan N, Khalid S, Khan I, Jabeen M. Impact of today's media on university student's body image in Pakistan: a conservative, developing country's perspective. BMC Public Health. 2011;11:379.
37. Medansky R, Handler R, Medansky D. Self-evaluation of acne and emotion: a pilot study. Psychosomatics. 1981;22:379-83.
38. Krowchuk D, Stancin T, Keskinen R. The psychosocial effects of acne on adolescents. Pediatr Dermatol. 1991;8:332-8.
39. Rocha M, Barnes F, Calderón J, Fierro-Arias L, Gómez CEM, Muñoz C, et al. Acne treatment challenges – recommendations of Latin American expert consensus. An Bras Dermatol. 2024;99:414-24.

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REVIEW ARTICLE

# Dermatology of the perianal region

## *Dermatología de la región perianal*

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## Abstract

*Nosology by body topography is an essential exercise in dermatological clinical practice. Many diseases involve the perianal region which due to its characteristics, are difficult to approach in primary care practice or even for experienced specialists. This document aims to present a review of different mucocutaneous pathologies that can affect this anatomical area. An updated bibliographic review was conducted of the most frequently dermatological entities located this area. The results describe the most relevant characteristics of their etiology, morphology and treatment, grouping them by clinical and pathological patterns. In conclusion, we believe it is important to communicate knowledge about skin diseases affecting the perianal region, providing a tool for physicians and consequentl improving patient well-being.*

***Keywords:*** *Dermatoses. Anal. Perianal. Perineum.*

## Resumen

*La nosología por topografía corporal es un ejercicio indispensable en la práctica clínica dermatológica. Un gran número de enfermedades involucran la región perianal y por sus particulares características resultan de difícil abordaje en el análisis médico de primer contacto e incluso para especialistas experimentados. Este documento tiene como objetivo realizar una revisión sobre diferentes patologías mucocutáneas que pueden afectar esta zona anatómica. Se realizó una revisión bibliográfica actualizada de las enfermedades con impacto dermatológico más frecuentemente encontradas en dicha localización. Se describen las características más relevantes de su etiología, morfología y tratamiento, agrupándolas por patrones clínico-patológicos. Como conclusión, nos parece relevante difundir el conocimiento sobre las enfermedades cutáneas que afectan la región perianal, para ofrecer una herramienta al médico tratante y, en consecuencia, aumentar el bienestar de los pacientes.*

***Palabras clave:*** *Dermatosis. Anal. Perianal. Periné.*

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## Introduction

The perianal region is affected mostly by focal dermatologic diseases; however, it may also manifest systemic disorders with perineal, intestinal, gynecologic, and other extensions. For this reason, multiple medical specialties should be involved to ensure accurate diagnosis and timely treatment. Various cultural, social, professional, or health care–access factors may delay care for these conditions, exposing patients to complications, clinical worsening, or self-medication[1].

Perianal dermatoses encompass a large number of globally distributed conditions that can be grouped in different ways according to inflammatory, infectious, immune-mediated, or tumoral origins, among others. They generate diverse signs and symptoms such as pruritus, pain, foreign-body sensation, discharge (secretion), or bleeding (hemorrhage). Signa are observed at all ages, including in newborns (eg, in acrodermatitis enteropathica)[2], but are more frequent in adulthood.

In primary care, knowledge of this anatomic region is usually basic; it is seldom taught and, consequently, poorly recognized. As a result, diagnostic workup is often challenging. Dermatologic processes in this area can extend by contiguity and involve the intergluteal cleft, buttocks, perineum, inguinal folds, external genitalia, and other sites, producing a broad regional process.

This caudal region, bordering the mucosa of the anal verge (approximately 5 cm), is lined by stratified keratinized epithelium with abundant folds generated by the subdermal insertion of the longitudinal and corrugator muscle extensions, among others. It is a frequently pigmented area with pilosebaceous units and a moist environment due to eccrine and apocrine sweat gland activity[3]. The transition to mucosa and submucosa is lined by non-keratinized squamous epithelium, which tends to be congested because of the recurrent presence of dense blood and lymphatic vasculature. The area is subject to friction, occlusion, increased temperature, and exposure to various biologic materials such as sweat, secretions, blood, semen, urine, and feces, and it harbors a distinctive microbiota.

This document aims to provide a general review of the different mucocutaneous conditions that may affect the perianal region so that clinicians have better tools for effective professional performance and, consequently, can offer greater well-being to their patients.

## Clinical approach and nosology

At the initial examination of a patient with a perianal dermatosis, clinicians should consider that, owing to the region's particular characteristics, the classic morphology of lesions may be altered, whether the lesions are solitary or part of a generalized process; the perianal presentation may even represent a sign of internal disease, and thus the remainder of the body must be examined. Proper patient positions for examination include the supine or lithotomy position, Sims position (left lateral), or the genupectoral (knee-chest) position, displacing both buttocks laterally to obtain an optimal field of view. In this way, one can identify color (erythema, pigment, achromia, etc) and the relationship of the lesions to the surface; lesions may be flat or elevated – such as papules, nodules, and tumors – or depressed with loss of continuity, as in excoriations, fissures, and ulcers.

During the medical history, elements may emerge that orient the initial diagnosis, based on signs and symptoms. Pruritus – often together with erythema – may be a constant sign of herpetic, mycotic, bacterial, irritant, or allergic infectious processes. When pruritus becomes chronic, it raises suspicion for other conditions such as atopic dermatitis (often accompanied by lichenification), contact dermatitis, psoriasis, lichen planus, extramammary Paget disease, lichen sclerosus, lichen simplex chronicus, or anal dysplasia. Infection with *Enterobius vermicularis* (pinworms) or other parasites can cause pruritus sine materiae, a term referring to the symptom in the absence of visible dermatosis; treatment consists of mebendazole, albendazole, or pyrantel pamoate and may coexist with other transmissible infections[4].

When the patient reports transient pain or a burning sensation, an anal fissure should be suspected. An anal fissure is a small linear tear caused by constipation or intense straining during defecation, chronic diarrhea, Crohn disease, or trauma. It is often recurrent and is a common reason for consultation with a proctologist. Diagnostic evaluation may include anoscopy, flexible sigmoidoscopy, or colonoscopy. Treatment options are varied and include topical wound-healing agents, analgesics, sitz baths, topical nitroglycerin, topical anesthetics, antihypertensives, botulinum toxin, or – if chronic – surgical procedure[5]. If anal pain is persistent, regional venous thrombosis may be the cause; if there is stabbing pain and systemic symptoms, an abscess may be present, which typically improves after drainage of the purulent collection. Proctitis, in addition to the above, is accompanied by urgency and tenesmus.

Signs of discharge and exudate are identified in the presence of hemorrhoids, proctitis, gonorrhea, lymphogranuloma venereum, condyloma acuminatum, acute dermatitis with eczema, and bacterial infections. Acute bleeding may be due to lacerated hemorrhoids or herpetic exulceration, whereas in the chronic phase (more than 6 weeks), anal carcinoma should be considered. Some patients report a foreign-body sensation; in such cases, common causes include benign tumoral lesions such as fibromas (acrochordons) or malignant lesions such as focal squamous cell carcinoma in a vegetating form (giant Buschke-Löwenstein condyloma) or rectal carcinoma originating from the internal tract. A sense of mass may also be due to cysts, hemorrhoids, and condyloma acuminata.

## Distinction by clinicopathologic patterns

A variety of dermatoses affecting the perianal region can be distinguished by their histologic characteristics, thereby establishing a correct diagnosis – always with proper clinicopathologic correlation – in an approach similar to that used, for example, in the pathology of the vulvar region[6,7] (Table 1).

**Table 1.** Clinicopathologic patterns of lesions affecting the perianal region

| Clinicopathologic pattern | Diseases |
|---|---|
| Spongiotic–Inflammatory | Atopic dermatitis<br>Seborrheic dermatitis<br>Contact dermatitis<br>Infectious dermatoses |
| Acanthotic | Lichen simplex chronicus |
| Psoriasiform | Classic (vulgar) psoriasis<br>Inverse psoriasis<br>Guttate psoriasis |
| Lichenoid | Lichen planus |
| Homogenization–Sclerotic | Lichen sclerosus<br>Radiodermatitis |
| Vesiculobullous | Herpetic infection<br>Pemphigus<br>Pemphigoid<br>Linear IgA disease<br>Darier disease<br>Benign familial pemphigus (Hailey-Hailey disease)<br>Vulvocrural acantholytic disease |
| Granulomatous | Crohn disease |
| Vasculopathic | Behçet disease<br>Plasma cell mucositis-dermatitis |
| Tumoral | Infectious:<br>Condyloma acuminatum<br>Abscesses<br>Benign:<br>Fibromas<br>Hemorrhoids/rectal prolapse<br>Cysts<br>Angiomas<br>Malignant:<br>Squamous cell carcinoma<br>Adenocarcinoma<br>Sarcomas<br>Melanoma<br>Lymphomas |

*Modified and adapted from Lynch et al.*[6,7]

### *Spongiotic–inflammatory pattern*

Acute dermatitis processes characterized by intense erythema, inflammation, vesicles, erosion, and eczema ("weeping" skin) may be triggered by endogenous or exogenous events (Fig. 1). In the chronic phase, erythema, hyperpigmentation, lichenification, and scale are observed[8]. (Fig. 2). As in the genital–perineal region, contact phenomena may be triggered by allergens such as topical anesthetics, neomycin, latex, chlorhexidine, and lanolin, or by irritants such as cleansers, pH-altering secretions, creams, alcohol, drugs, and others[9].

Fixed drug eruption is a drug-induced dermatosis that may appear in this anatomic region, beginning as an inflammatory, red lesion – sometimes with a blister – that is symptomatic and is frequently triggered by antibiotics (sulfonamides) or nonsteroidal anti-inflammatory drugs. The sequela is a residual postinflammatory macule, and re-exposure to the inciting agent produces recurrence at the same site[10] (Fig. 3).

The condition known as granuloma *gluteale infantum* is an irritant contact dermatitis with frequent *Candida* spp overgrowth, favored by occlusion from diaper use, benzocaine exposure, and fluorinated corticosteroids in children and incontinent adults[11,12]. In the differential diagnosis, one must include Sevestre-Jacquet posterior erosive syphilid dermatitis (Fig. 4), a severe and rare condition associated with the use of reusable "eco-friendly" cloth diapers; the most recent case was described in Mexico[13].

Management of contact dermatitis begins with discontinuation of the causal agent – whether contactant or allergen. Astringent wet dressings (eg, aluminum sulfate/calcium acetate), sitz baths, topical corticosteroids, and systemic antihistamines should be recommended.

Perianal streptococcal dermatitis is an infectious condition around the anus caused by group A β-hemolytic


**Figure 1.** Atopic dermatitis: acute **(A)** and chronic **(B)** phases. Contact dermatitis due to cleansing wipes **(C)** and hemorrhoid drugs **(D)**.

streptococcus, common in childhood but also possible in adults. It is characterized by well-demarcated erythematous plaques, edema, isolated pustules, pruritus, fissures, bleeding, and pain. Treatment should include antimicrobials such as penicillin, amoxicillin-clavulanate, cephalosporins, or macrolides for 5 to 10 days; antifungal therapy, topical or systemic, is frequently added to the antibiotic regimen[14]. Appropriate follow-up should be established, management individualized, and recurrences avoided[15].

Within this disease spectrum, seborrheic dermatitis should also be considered. It presents as erythema covered by fine white or yellowish scale in hair-bearing areas (Fig. 5). Affected individuals may have mild to moderate burning or pruritus, but the condition causes psychological distress, diminished self-esteem, and embarrassment. It should be made clear that seborrheic dermatitis is neither contagious nor the result of poor hygiene. Proliferation and hypersensitivity to fungal strains of the genus *Malassezia* are identified. Treatment should include topical agents – and, on occasion, systemic therapy – such as nystatin, clioquinol, imidazoles, piroctone olamine, keluamide, and even low-potency corticosteroids.

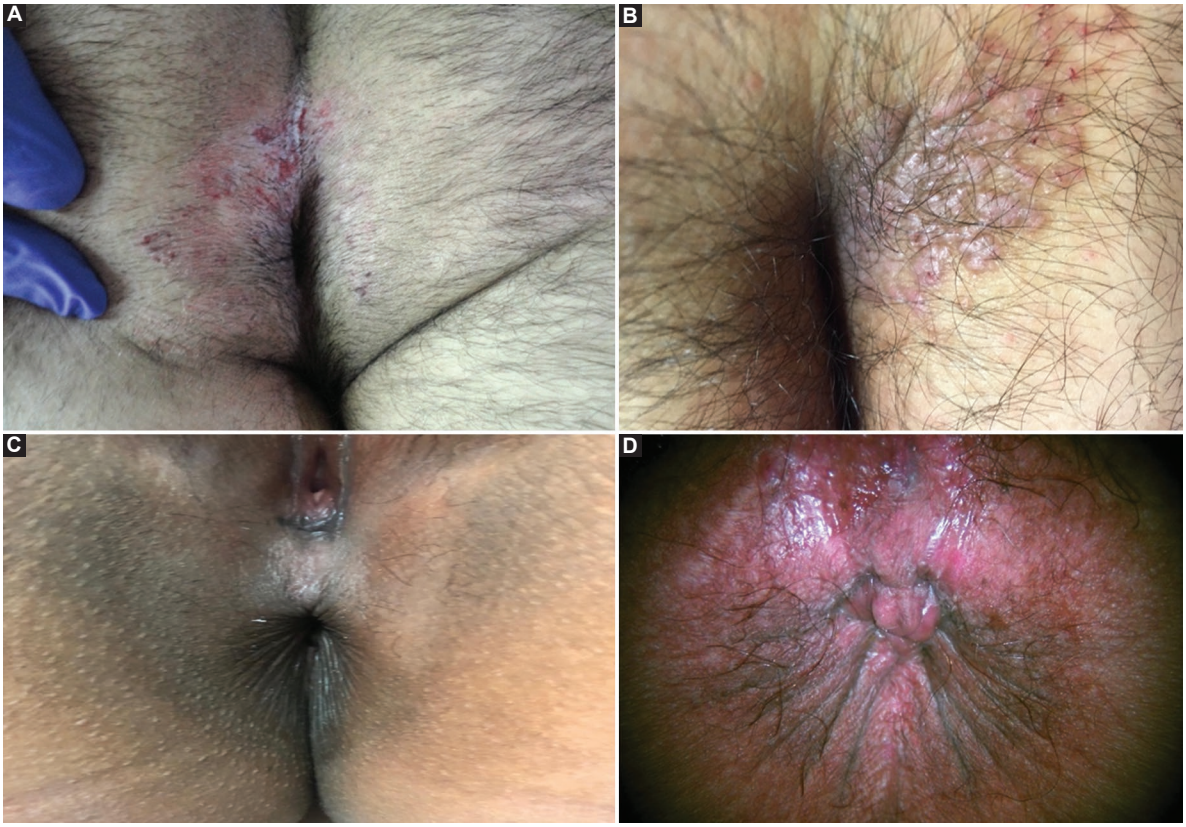


**Figure 2. A** and **B:** chronic dermatitis, lichen simplex phase with scratch marks. **C** and **D:** Residual postinflammatory hyperpigmentation.

Atopic dermatitis is a chronic-recurrent inflammatory skin disease, highly pruritic and affecting all ages. It is one of the most common reasons for dermatology consultation and the leading cause in pediatrics; it may precede other conditions such as allergic rhinitis, conjunctivitis, or asthma and is difficult to control. It is a multifactorial disease with immune dysregulation and genetic predisposition leading to impaired skin-barrier function, influenced by environmental and patient-specific factors. Beginning in infancy – as part of the diaper-area dermatitis complex – it may involve the perianal region; in adulthood it may produce lichen simplex chronicus, as occurs in the vulva and scrotum, with sometimes intractable pruritus (Figs. 1 A and B).

Another relevant sign in this body region is Langerhans cell histiocytosis (formerly histiocytosis X), a rare disease characterized by clusters of these cells together with lymphocytes, eosinophils, and macrophages. Clinically it produces inflammatory dermatitis plaques on the scalp and trunk but may involve mucocutaneous sites and even present initially with ulcers or exulcerations in the perianal region with secondary superinfection[16]. Diagnosis is achieved by biopsy, and treatment is nonspecific; corticosteroids and vinblastine have been recommended[17].

## *Acanthotic pattern*

Within this context, several conditions display an acanthotic pattern, characterized by orthokeratotic hyperkeratosis, focal parakeratosis, irregular acanthosis, vertical dermal vessels, and vertical collagen orientation.

This group includes lichen simplex chronicus, which shows squamous cell hyperplasia and is considered a localized form of atopic dermatitis. This condition generates a vicious itch-scratch-itch cycle and has been associated with irritant or sensitizing agents as well as emotional stress (Figs. 2 A and B).

## *Psoriasiform pattern*

Psoriasis of the perianal region exhibits a psoriasiform histologic pattern, with regular acanthosis,


**Figure 3. A:** fixed drug eruption (sulfonamide-induced). **B:** vulvar and perianal vitiligo. **C:** perineal lichen sclerosus. **D:** inverse psoriasis.

absence of the granular layer, parakeratotic hyperkeratosis, suprapapillary thinning, neutrophilic microabscesses in the stratum corneum, and dilated, tortuous vessels. Psoriasis is a chronic, systemic, immune-mediated inflammatory disease that affects the skin, nails, and joints. It has a recognized genetic predisposition and may be triggered or exacerbated by environmental and emotional factors. Clinically, it presents with well-defined erythematous plaques covered by thick, whitish, glossy scales, widely appearing over bony prominences, the scalp, and the lumbar region. The so-called inverse form occurs in flexural areas such as the antecubital

**Figure 4.** Sevestre-Jacquet posterior erosive syphilid in diaper dermatitis.

folds, groin, intergluteal, and perianal regions (Fig. 3D). Treatment includes soap substitutes, emollients, topical and systemic corticosteroids, calcipotriol, oral retinoids, and interleukin-23 and interleukin-17 inhibitors, among others, with variable results[18]. Among newer therapeutic options for plaque psoriasis are JAK inhibitors, such as topical ruxolitinib, which also provide benefits in other immune-mediated dermatoses[19,20].

## Lichenoid pattern

Clinical entities with a lichenoid pattern show histopathologic findings of orthokeratotic hyperkeratosis, hypergranulosis, interface involvement, vacuolization of basal cells, and a band-like lymphocytic infiltrate. This group includes lichen planus, an inflammatory dermatosis with a predilection for mucosal surfaces, typically chronic, self-limited, and relapsing. Lesions are papular, polygonal, flat-topped, purpuric, and pruritic, and may sometimes be pigmented. The condition may be localized or disseminated, and occasionally erosive. Without appropriate treatment, there is some risk of progression to carcinoma[21]. It has been reported that up to 25% of women with oral lichen planus present anogenital lesions. In mucosal sites, the disease manifests as erosions and whitish striae, giving a characteristic "lace-like" appearance[22].

## Homogenization pattern

The homogenization/sclerosis pattern is characterized microscopically by orthokeratotic hyperkeratosis, epidermal atrophy, hydropic degeneration of the basal layer, edema, and collagen homogenization with a lichenoid inflammatory infiltrate. These features are found in lichen sclerosus, a chronic lymphocyte-mediated disease triggered by autoimmune mechanisms. It presents as whitish, atrophic plaques in the anogenital region of both sexes. This is a debilitating condition that may progress to complications such as squamous cell carcinoma[23] (Fig. 3C).

Diagnosis of lichen planus and lichen sclerosus is confirmed by histopathologic examination. The treatment of choice consists of high-potency corticosteroids; surgery should only be considered in cases associated with malignancy or advanced scarring that cause dyspareunia, phimosis, or other forms of regional functional restriction[24]. These disorders are generally non-curable, but good medium- and long-term control can be achieved.

Vitiligo is the most common depigmenting disease and a relevant differential diagnosis of lichen sclerosus. It affects up to 1% of the global population and has a significant impact on quality of life. It manifests as achromic macules on the skin and mucous membranes; although it predominates on the extremities, acral zones, and eyelids, periorificial forms can occur around the mouth, genitalia, and perianal area (Fig. 3B).

## Vesiculobullous pattern

In the vesiculobullous pattern, histologic examination reveals blisters at different levels within the epithelium, each with distinctive characteristics. Ballooning degeneration is seen in herpetic diseases, including herpes simplex, herpes zoster, and Kaposi varicelliform eruption. At a deeper level – the dermoepidermal junction or subepidermis – antigenic deposits appear in anchoring fibrils, giving rise to conditions such as bullous pemphigoid, gestational pemphigoid, mucous membrane pemphigoid, and linear IgA disease. Acantholysis is common in pemphigus vulgaris and pemphigus foliaceus, which produce anogenital lesions in 2%-27% of cases[25]. This acantholytic pattern, with loss of keratinocyte cohesion, isolated rounded cells, clefts, and blisters, is also identified in Hailey-Hailey disease, Darier disease, and vulvocrural acantholytic dermatosis – all of which may involve the genital and perianal areas.

The presence of grouped vesicles on an erythematous base is the characteristic image of a herpetic infection (*herpes simplex virus* types 1 and 2 or *varicella zoster virus*), which, in this anatomic region, is often sexually transmitted; however, it must be distinguished from perineal herpes zoster. When bullous lesions rupture, they may leave denuded, highly symptomatic areas

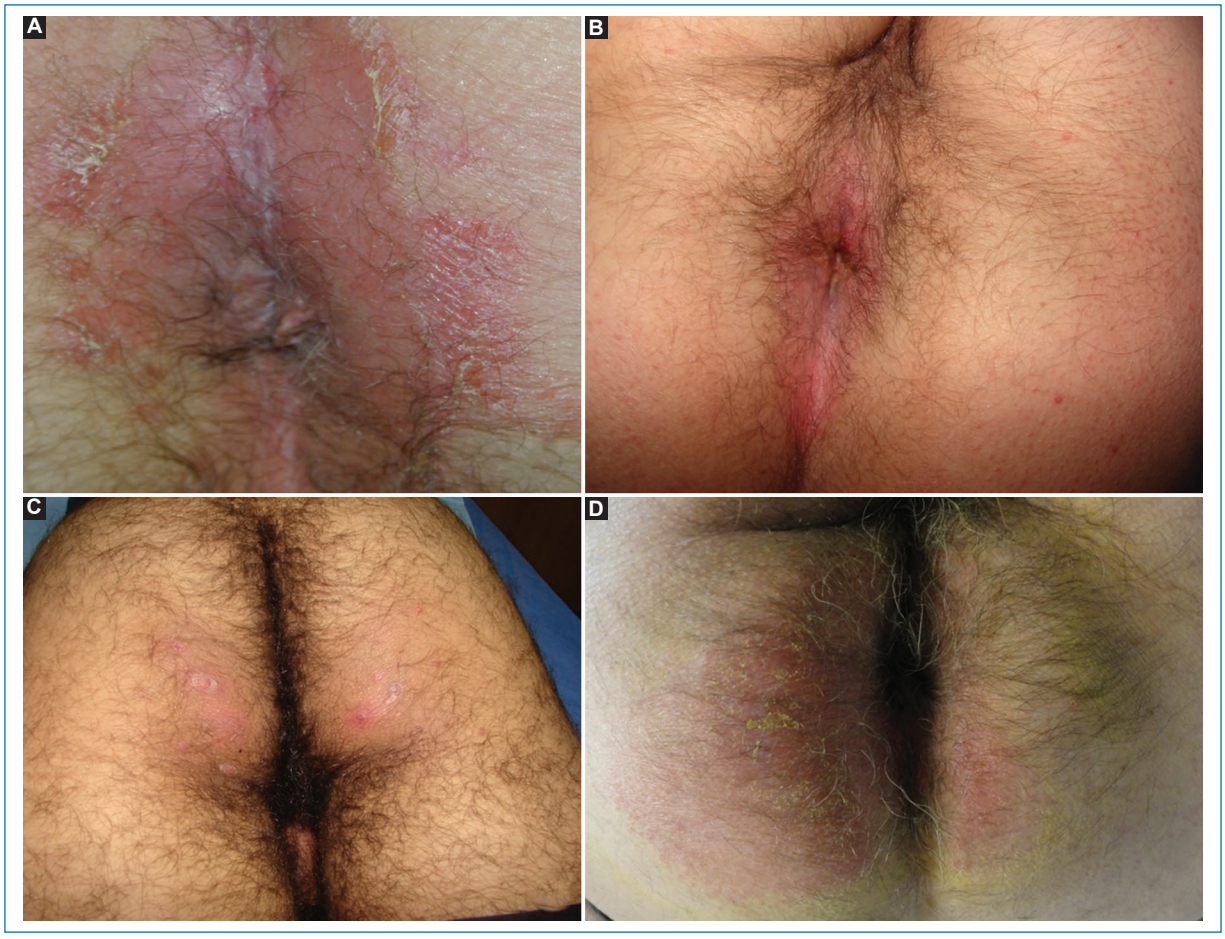


**Figure 5.** **A** and **B:** acute dermatitis eczema. **C:** bacterial folliculitis. **D:** seborrheic dermatitis.

(Fig. 6). Special attention should be paid to immunosuppressed patients, in whom the condition may be more severe. Usual treatment includes local measures and systemic therapy with acyclovir, valacyclovir, or famciclovir, among others[26].

For isolated ulcerations, other differential diagnoses must be considered. Syphilis, in its primary stage, produces the classic, well-defined ulcer known as a chancre; without appropriate therapy, the natural course of the disease may lead to irregular, vegetating masses characteristic of secondary syphilis (Fig. 7). Other conditions that can favor the development of local ulcers include Crohn disease and viral infections caused by herpes simplex virus or cytomegalovirus, particularly severe in immunosuppressed patients[27-29].

## *Granulomatous pattern*

When tissue shows a granulomatous infiltrate without evidence of infection, this represents the granulomatous patterncharacteristic of Crohn disease, a chronic inflammatory bowel disease of unknown cause. In the anal region and the skin-mucosal junction, as occurs in the genital area, it produces edema, pain, erythema, ulcers, fistulas, lacerations, and recurrent abscesses[30]. Treatment is based on the use of corticosteroids, immunosuppressants, and agents directed against tumor necrosis factor, such as infliximab, etanercept, or adalimumab, among others.

Hidradenitis suppurativa (also called acne inversa) is a chronic, inflammatory–granulomatous disease with relapsing phases that produces painful, suppurative nodular lesions due to follicular and apocrine gland obstruction. It appears in the axillae, submammary region, groin, and buttocks, extending to adjacent areas. It is more common in women, with a 3:1 ratio; however, in men, perineal and perianal lesions are more frequent[31]. Associated factors include obesity, smoking, tight clothing, chronic friction, and sedentary lifestyle. The condition can be classified according to

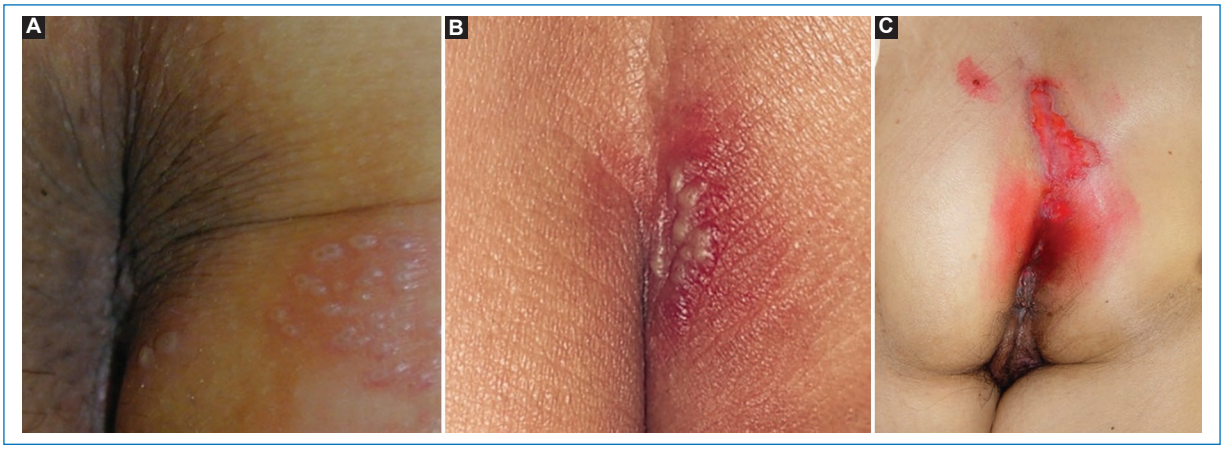


**Figure 6.** Herpes simplex: **A:** primary infection. **B:** recurrence. **C:** denuded areas.

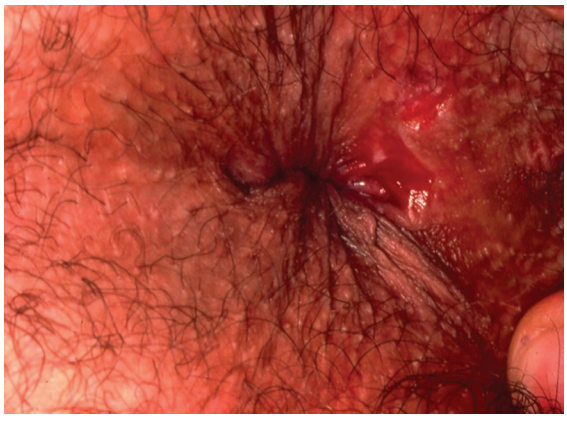

**Figure 7.** Syphilitic chancre.

its extent or severity. Multiple therapeutic options have been described – medical, pharmacologic, and surgical – but all yield partial or refractory results. Differential diagnoses include recurrent bacterial folliculitis and furunculosis (Fig. 5C).

The humidity and temperature of this region favor the proliferation of various fungal genera. When surface and immune conditions permit, infections may occur due to *Candida* spp, dermatophytes causing tinea corporis, or folliculitis caused by *Malassezia* spp (Fig. 8).

Deep cutaneous infections producing a granulomatous histologic pattern must also be considered in this region. These include a rare form of extrapulmonary tuberculosis that predominates in men in the fourth decade of life and presents with nodular, congestive lesions[32]. Another example is mycetoma, an infection by aerobic actinomycetes (predominantly *Nocardia brasiliensis*), which rarely affects the perianal region. In our country, Chávez et al[33] reported a series of 20 cases in this anatomic site, with greater incidence in rural male workers in their 5th decade of life. It is a condition associated with poor hygiene and poverty (Fig. 9).

## Vasculopathic pattern

This pattern is characterized by the presence of angiocentric and eosinophilic inflammatory infiltrates, vasculitis, and hemorrhage. It is constant in Behçet disease, an autoimmune vasculitis with predilection for specific organs, associated with human leukocyte antigen (HLA)-B51 and highly prevalent in the Middle East and Central Asia. Clinically, painful lesions appear with the classic triad of oral ulcers, genital ulcers, and ocular lesions; however, articular, cardiac, cutaneous, and central nervous system involvement has also been described. Therapy includes high-dose corticosteroids, immunosuppressants, tumor necrosis factor inhibitors, interferon, and thalidomide[34].

## Tumoral pattern

This particular anatomic region comprises multiple cellular lineages and tissues that predispose to diverse neoplasms. As in other cutaneous areas, tumoral lesions of the perianal region can be classified according to their lineage as benign or malignant. Among benign lesions, the most frequent are fibromas, nevi, cystic lesions, and angiomatous lesions. Elevated, vegetating masses such as condyloma acuminatum, secondary to human papillomavirus (HPV) infection, are common (Fig. 10). Infantile perianal pyramidal protrusion is a soft, pink, smooth lesion – congenital or acquired – relatively frequent in

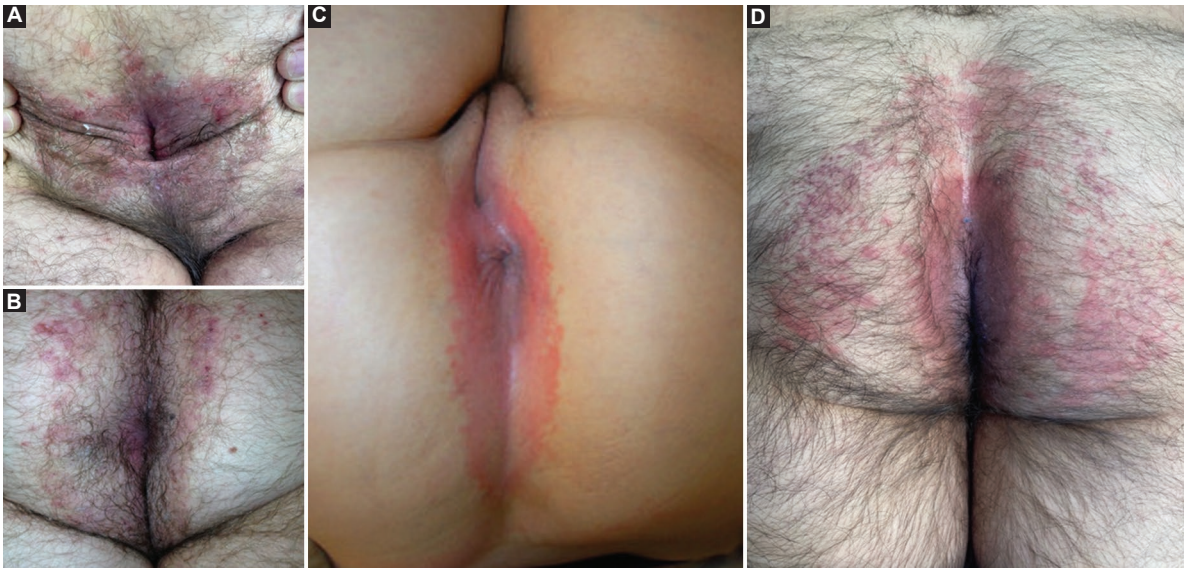


**Figure 8.** Mycology: **A:** dermatophytic tinea. **B:** *Malassezia* folliculitis. **C:** infantile candidiasis. **D:** adult diabetic candidiasis.

**Figure 9.** *Nocardia* spp mycetoma (courtesy of Alexandro Bonifaz, MSc).

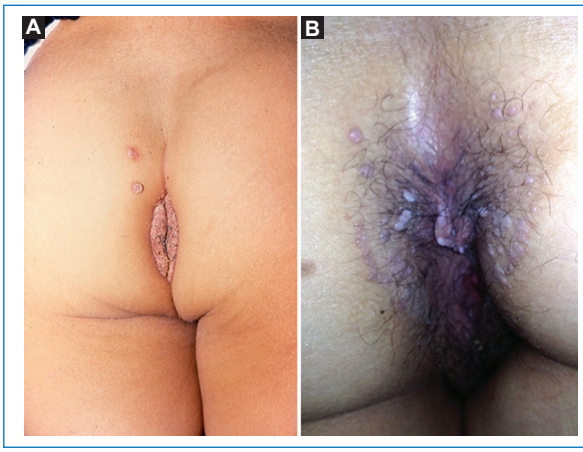


**Figure 10.** Condyloma acuminatum in an infant **(A)** and an adult **(B)**.

infants, often underdiagnosed, and must be differentiated from condyloma acuminatum[35]. Some fibromatous lesions in adults can have a similar pyramidal appearance. Other lesions that produce a pseudotumoral, vasculopathic–varicose, or congestive aspect – such as hemorrhoids or rectal prolapse – may present with recurrent, sometimes painful, congestive episodes and require evaluation and treatment by a proctologist[36] (Fig. 11).

Malignant tumors of the perianal area appear as localized masses in the skin or by infiltration extending from the anal canal or adjacent structures. The most frequent is squamous cell carcinoma, accounting for up to 80% of

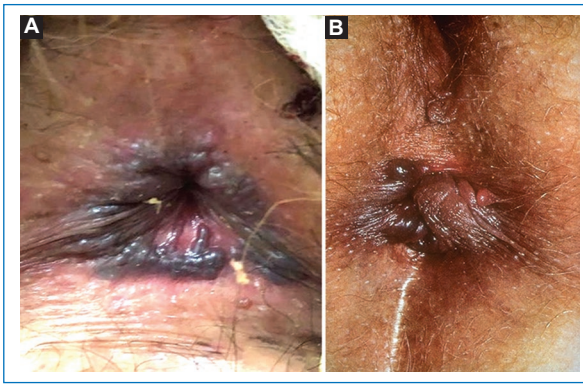


**Figure 11.** Vascular lesion: **A:** varicose process. **B:** hemorrhoidal disease.

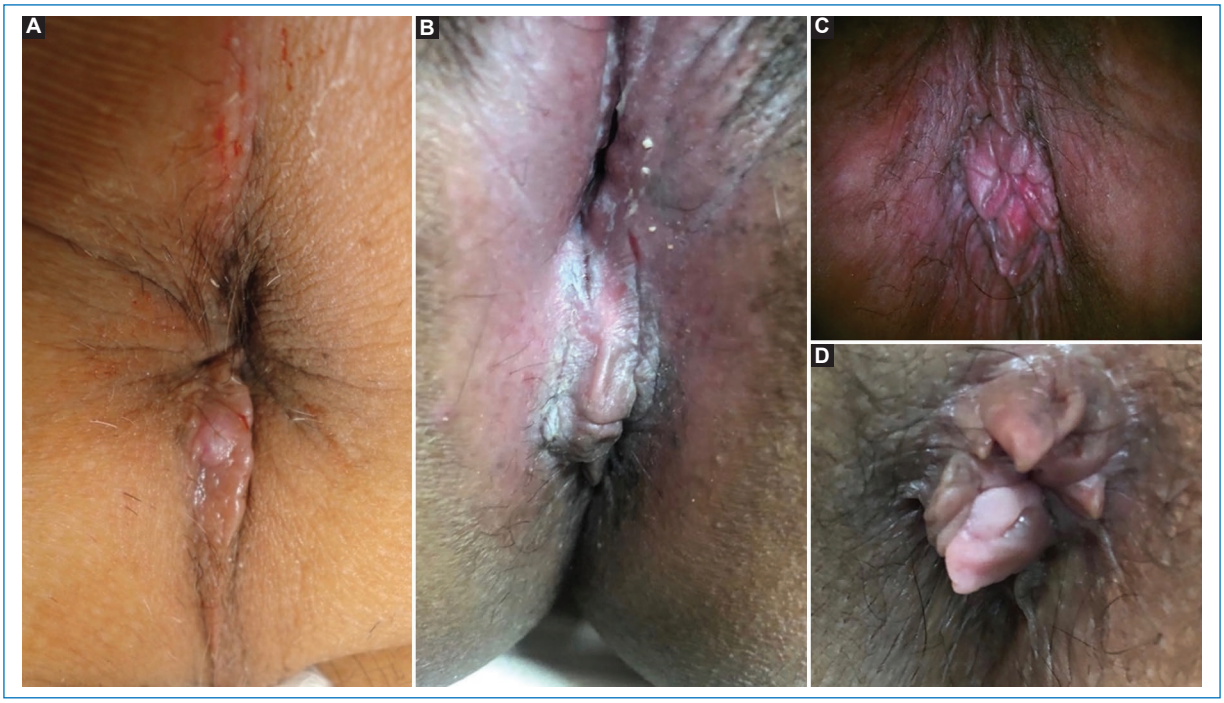


**Figure 12. A** and **B:** squamous cell carcinoma of skin and mucosa. **C** and **D:** anal intraepithelial neoplasm.

cases and occurring mainly in adults in the 6th decade of life. A common variant in this region is anal intraepithelial neoplasm (Fig. 12). The main inducers are HPV and human immunodeficiency virus (HIV) infections, and the lesion has metastatic potential[37]. Anal adenocarcinomas may be mucinous or nonmucinous, representing approximately 15% of cases. Cutaneous melanoma in this location is rare and often presents as a polypoid lesion, generally diagnosed histologically. Other uncommon tumors include neuroendocrine neoplasms, leiomyosarcomas, basal cell carcinoma, anal intraepithelial neoplasm (formerly Bowen disease), Paget disease, and lymphomas[38]. All these conditions require a comprehensive and multidisciplinary approach with appropriate complementary studies and should be managed by specialized medical personnel.

## Conclusions

Perianal skin disorders are generally localized, benign, and highly variable. They may significantly affect patients' quality of life, compromising comfort and daily functional capacity. The most frequent symptom is pruritus, associated with acute or chronic multifactorial inflammatory processes. The most widely used therapies are emollients and corticosteroids of varying potency, usually with good results, although some conditions can only be partially controlled. We consider it important to promote awareness of the mucocutaneous diseases of the perianal region to improve diagnostic capacity and provide better relief for affected patients.

## Funding

The authors declare that this work was conducted with their own resources.

## Conflicts of interest

The authors declared no conflicts of interest whatsoever.

## Ethical considerations

**Protection of humans and animals:** The authors declare that no experiments were performed on humans or animals for this study.

**Confidentiality, informed consent, and ethics approval:** The study does not involve patient data or require ethics committee approval. The SAGER guidelines do not apply.

**Declaration on the use of artificial intelligence:** The authors declare that no generative artificial intelligence was used in the preparation of this manuscript.

## References


1. Dietrich A, Ruzicka T, Hermans C. [Differential diagnosis of anal eczema]. Hautarzt. 2015;66:400-7.
2. Corbo MD, Lam J. Zinc deficiency and its management in the pediatric population: a literature review and proposed etiologic classification. J Am Acad Dermatol. 2013;69:616-24e1.
3. Palma-Rodríguez F, Palma-Carazo P, Leyva A, Palma F. Anatomía de la región perianal. Sem Med. 1998;50:65-76.
4. Schadelbauer E, Tripolt-Droschl K, Sadoghi B. [Urethritis due to coinfection with Neisseria gonorrhoeae and Enterobius vermicularis in a 27-yearoldman]. Urologie. 2023;62:53-5.
5. Salati SA. Anal fissure – an extensive update. Pol Przegl Chir. 2021;93:46-56.
6. Lynch PJ, Moyal-Barracco M, Scurry J, Stockdale C. 2011 ISSVD Terminology and classification of vulvar dermatological disorders: an approach to clinical diagnosis. J Low Genit Tract Dis. 2012;16:339-44.
7. Lynch PJ, Moyal-Barracco M, Bogliatto F, Micheletti L, Scurry J. 2006 ISSVD classification of vulvar dermatoses: pathologic subsets and their clinical correlates. J Reprod Med. 2007;52:3-9.
8. Weyandt G, Breitkopf C, Werner RN, Zidane M, Furtwängler A, Jongen J, et al. German S1 guidelines for the diagnosis and treatment of perianal dermatitis (anal eczema). J Dtsch Dermatol Ges. 2020; 18:648-57.
9. Margesson LJ. Vulvar disease pearls. Dermatol Clin. 2006;24:145-55.
10. Valeyrie-Allanore L, Lebrun-Vignes B, Bensaid B, Sassolas B, Barbaub A. [Fixed pigmented erythema: epidemiology, physiopathology, clinical features, differential diagnosis and therapeutic management]. Ann Dermatol Venereol. 2015;142:701-6.
11. Ramos Pinheiro R, Matos-Pires E, Baptista J, Lencastre A. Granuloma glutaeale infantum: a re-emerging complication of diaper dermatitis. Pediatrics. 2018;141:e20162064.
12. Robson KJ, Maughan JA, Purcell SD, Petersen MJ, Haefner HK, Lowe L. Erosive papulonodular dermatosis associated with topical benzocaine: a report of two cases and evidence that granuloma gluteale, pseudoverrucous papules, and Jacquet's erosive dermatitis are a disease spectrum. J Am Acad Dermatol. 2006;55(5 Suppl):S74-80.
13. Dithurbide-Hernández M, Del Río-Martínez CJ, Serrano-Vázquez IC, Rodríguez-Patiño G. [Sevestre-Jacquet posterosive syphilitic dermatitis associated with the use of reusable diapers: A clinical case]. Rev Med Inst Mex Seguro Soc. 2021;59:242-7.
14. Šterbenc A, Točkova O, Lah LL, Kamhi Trop T, Seme K, Švent-Kučina N, et al. A retrospective analysis of clinical characteristics and management of perianal streptococcal dermatitis in children and adults. Acta Dermatovenerol Alp Pannonica Adriat. 2021;30:99-104.
15. Serban ED. Perianal infectious dermatitis: an underdiagnosed, unremitting and stubborn condition. World J Clin Pediatr. 2018;7:89-104.
16. Poppe LM, Müller PA, Poppe H, Bröcker EB, Ugurel S, Weyandt GH. Chronic perianal ulceration as the initial symptom of Langerhans cell histiocytosis in adults. Eur J Dermatol. 2013;23:551-2.
17. Tinsa F, Brini I, Kharfi M, Mrad K, Boussetta K, Bousnina S. Perianal presentation of Langerhans cell histiocytosis in children. Gastroenterol Clin Biol. 2010;34:95-7.
18. Mastorino L, Dapavo P, Macagno N, Ortoncelli M, Verrone A, Stroppiana E, et al. Clinical characteristics and response to biological therapies for inverse psoriasis: a real-life comparison between the therapeutic effects of anti-IL-23 and anti-IL-17 agents. Int J Dermatol. 2024;63:1735-9.
19. Dogra S, Sharma A, Mehta H, Sarkar R. Emerging role of topical Janus kinase inhibitors in dermatological disorders: a review. Clin Exp Dermatol. 2023;48:1102-12.
20. Spadafora M, Morsia S, Di Lernia VG, Kaleci S, Pellacani G, Longo C. Off-label use of topical ruxolitinib in dermatology: a systematic literature review and current perspectives. Exp Dermatol. 2025;34:e70095.
21. Sharma N, Malhotra SK, Kuthial M, Chahal KS. Vulvo-vaginal ano-gingival syndrome: another variant of mucosal lichen planus. Indian J Sex Transm Dis AIDS. 2017;38:86-8.
22. Pecoraro V, Romano Boix E. [Anal manifestations of lichen planus]. Med Cutan Ibero Lat Am. 1984;12:339-44.
23. Pugliese JM, Morey AF, Peterson AC. Lichen sclerosus: review of the literature and current recommendations for management. J Urol. 2007;178:2268-76.
24. Chung ASJ, Suarez OA. Current treatment of lichen sclerosus and stricture. World J Urol. 2020;38:3061-7.
25. Núñez-Rocha RE, Andrade-Restrepo J, Gómez-Carrillo D, Rolón M, Duperly RG, Del Pilar López-Panqueva R. Anal involvement in patient with pemphigus vulgaris: case report. Int J Surg Case Rep. 2024;114:109185.
26. Groves MJ. Genital herpes: a review. Am Fam Physician. 2016;93:928-34.
27. Simon E, Steinhagen E. Nonfistulizing perianal Crohn's Disease. Clin Colon Rectal Surg. 2024;38:141-7.
28. Ko BK, Wei X, Ong WM, Mohd Rosli R, Wong A. An unusual case of genital herpes simplex virus (HSV) infection with severe non-necrotizing soft tissue infection. Cureus. 2025;17:e82583.
29. Ginwala A, Gaikwad S, Pujari M, Patel D, Khirid V, Pujari S. Cutaneous cytomegalovirus ulcers in an individual with acquired immunodeficiency syndrome: a case report. J Assoc Physicians India. 2024;72:e43-4.
30. Boxhoorn L, Stoof TJ, de Meij T, Hoentjen F, Oldenburg B, Bouma G, et al. Dutch Initiative on Crohn and Colitis. Clinical experience and diagnostic algorithm of vulval Crohn's disease. Eur J Gastroenterol Hepatol. 2017;29:838-43.
31. Hagerman GF, Silva-Velazco J, Molina-López JF. Miscellaneous perianal afflictions. Clin Colon Rectal Surg. 2019;32:394-402.
32. Salves D, Podestá F, Ferreyra D, Debuck M, Galván F. Tuberculosis anal como diagnóstico diferencial de lesión ulcerada anal. Rev Argent Coloproct. 2018;29:25-7.
33. Chávez G, Estrada R, Bonifaz A. Perianal actinomycetoma: experience of 20 cases. Int J Dermatol. 2002;41:491-3.
34. Haller C, Guenot C, Odman M, Bruttin JM, Rosso R. [Recurrent anal abscess and cecal perforation as a first presentation of Behçet's disease]. Gastroenterol Clin Biol. 2003;27:940-3.
35. Castro N, Vivoda JL, Belleli A, Barabini LN, Grees SA. Protrusión piramidal perianal infantil en una niña lactante. Dermatol Arg. 2018;24:106-7.
36. Davis BR, Lee-Kong SA, Migaly J, Feingold DL, Steele SR. The American Society of Colon and Rectal Surgeons clinical practice guidelines for the management of hemorrhoids. Dis Colon Rectum. 2018;61:284-92.
37. Augustine A, Issac R, Lakhani A, Kanamathareddy HV, John R, Simon B, et al. Anal and perianal masses: the common, the uncommon, and the rare. Indian J Radiol Imaging. 2024;34:688-701.
38. Cruz-Ramos PA, Nguyen S, Hayman AV. Uncommon anal cancers. Surg Oncol Clin N Am. 2025;34:103-13.

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CLINICAL CASE

# Plasmablastic lymphoma: atypical cutaneous presentation

## *Linfoma plasmablástico: presentación cutánea atípica*

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## Abstract

*Plasmablastic lymphoma is a variant of non-Hodgkin lymphoma, a primary cutaneous diffuse B-cell lymphoma, other type large cell. Uncommon, it mainly affects patients infected with the human immunodeficiency virus (HIV). It tends to occur in the oral cavity. Occasionally it is also described in patients not infected with HIV and in locations other than the oral cavity. It is a diagnostic challenge for the dermatologist due to the low incidence and clinical polymorphism associated with HIV infection. We present the case of a 23-year-old patient with an atypical presentation of this disease associated with HIV infection.*

***Keywords:*** *Plasmablastic lymphoma. Hallux. HIV. Chemotherapy.*

## Resumen

*El linfoma plasmablástico es una variante de linfoma no Hodgkin, un linfoma cutáneo primario de células B difuso, de células grandes tipo otro. Es poco frecuente y afecta principalmente a pacientes infectados por el virus de la inmunodeficiencia humana (VIH). Tiende a presentarse en la cavidad oral. Ocasionalmente también se describe en pacientes sin infección por el VIH y en localizaciones distintas de la cavidad oral. Es un reto diagnóstico para el dermatólogo por su baja incidencia y el polimorfismo clínico asociado a la infección por el VIH. Exponemos el caso de un paciente de 23 años con presentación atípica de este linfoma asociado a infección por el VIH.*

***Palabras clave:*** *Linfoma plasmablástico. Hallux. VIH. Quimioterapia.*

## Introduction

Plasmablastic lymphoma (PBL) is a rare and highly aggressive subtype of non-Hodgkin lymphoma[1] that is predominantly observed in patients infected with the human immunodeficiency virus (HIV). This lymphoma shows a particular predilection for manifesting in the oral cavity[1], although it may also appear in other locations in patients without HIV infection. From a diagnostic perspective, PBL is characterized by a distinctive immunophenotype of activated B-cells that lose the typical markers of mature B-cells and acquire those associated with plasma cells[2]. Concomitant infection with the Epstein-Barr virus is common in these cases. Histologically, PBL shows hyperplastic cells with asymmetrical and eccentrically displaced nuclei (Table 1). Immunohistochemically, lesions are negative for CD45 and for B-cell markers[3] (such as CD20, CD19, and CD79a), and positive for plasma cell markers (CD38, MUM1, CD138, BLIMP-1, XBP-1), with a high Ki-67 index (Table 2). The aggressive nature of this lymphoma and its particular clinical presentation pose significant challenges in both diagnosis and treatment, underscoring the

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**Table 1.** Characteristics of plasmablastic lymphomas according to HIV status[1]

| | HIV-Positive | HIV-Negative |
|---|---|---|
| Frequency in the literature, n (%) | 70 | 30 |
| Risk factors | Low CD4 + lymphocyte counts<br>Absence of prior ART | Older age<br>Post-transplant<br>Transformation of prior lymphoma |
| Mean age, years | 39 | 58 |
| Over 60 years old, n (%) | 1 | 56 |
| Male to female ratio | 4-1 | 2-3:1 |
| EBV positive, n (%) | 75-80 | 50-67 |
| MYC gene alterations, n (%) | 50-80 | 40-44 |
| Extranodal presentation, n (%) | 95 | 70-90 |
| Primary site, n (%) | Oral cavity (48-58)<br>Extraoral (42-48) | Oral cavity (16-40)<br>Extraoral (56-84) |

ART: antiretroviral therapy; EBV: Epstein-Barr virus; HIV: human immunodeficiency virus.

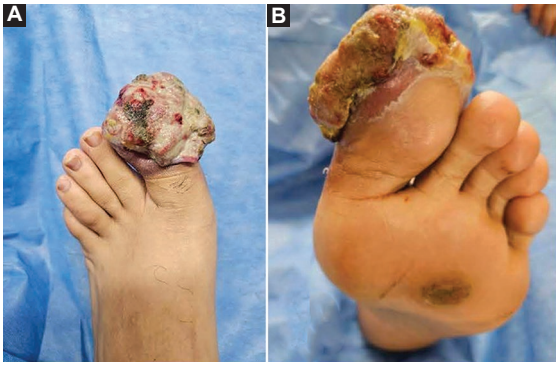


**Figure 1.** Tumor on the left hallux. **A:** exophytic tumor with necrotic areas, hemorrhagic zones, rapid growth, and foul odor. **B:** posterior view of the left foot.

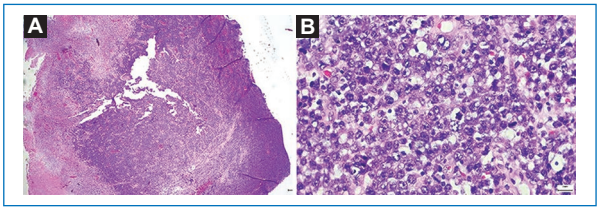


**Figure 2.** Biopsy of the lesion stained with hematoxylin-eosin. **A:** panoramic view showing the dermis occupied by a proliferation of atypical cells. **B:** at higher magnification, moderate nuclear pleomorphism, coarse chromatin, and prominent nucleoli.

need for specific therapeutic approaches and a deeper understanding of its biology and epidemiology.

## Case report

A 23-year-old man presented with a 3-month history tumor on the left hallux. The patient's past medical history included an HIV diagnosis 1 year prior without antiretroviral treatment and recurrent onychocryptosis, for which he had undergone 3 matricectomies on the left hallux.

On physical examination, an exophytic tumor was observed on the left hallux with areas of necrosis and hemorrhage, rapidly growing, approximately 5 cm × 6 cm in size (Fig. 1). The lesion was painful on palpation and emitted a foul odor.

Lab test results showed a viral load of 28,100 copies/mL and a CD4 count of 257 cells. Positron emission tomography showed no bone involvement. A skin biopsy was performed for histopathology (Fig. 2) and immunohistochemistry (Fig. 3).

Initial treatment included combination antiretroviral therapy with dolutegravir (50 mg), lamivudine (300 mg), and tenofovir (300 mg). Additionally, a chemotherapy regimen was initiated with dose-adjusted DA-EPOCH (etoposide, prednisone, vincristine, cyclophosphamide, and doxorubicin). By the third chemotherapy cycle, the patient showed a good response (Fig. 4).

## Discussion

PBL represents a significant challenge in dermatology due to its rarity and aggressiveness, especially in HIV-infected patients[4]. The immunophenotypic characterization of PBL, which shows loss of typical mature

**Table 2.** Differential diagnosis of plasmablastic lymphoma[1]

| | Location | Clinical context | Immunophenotypic markers | | Infection | | |
|---|---|---|---|---|---|---|---|
| | | | Positive markers | Negative markers | HIV | EBV | HHV8 |
| Plasmablastic lymphoma | Extranodal (oral cavity, mucosae) | HIV infection | CD138 | CD45 | 70% | Yes | No |
| | | Post-transplant | MUM1/IRF4 | CD20 | | | |
| | | Older age | MYC | PAX5 | | | |
| Multiple myeloma | Bone marrow and extranodal | Paraproteinemia | CD138 | CD20 | No | No | No |
| | | Renal disease | cIg | PAX5 | | | |
| | | Bone disease | | | | | |
| ALK + DLBCL | Lymph nodes | Young patients | ALK | CD20 | No | No | No |
| | | Advanced stages | CD4 | CD30 | | | |
| | | B symptoms | CD45 | MYC | | | |
| HHV8-positive large B-cell lymphoma | Lymph nodes and spleen | Multicentric Castleman disease | CD20+ / – MUM1/IRF4 | CD79a CD138 | Yes | No | Yes |
| | | HIV infection | cIgM | | | | |
| Primary effusion lymphoma | Serous cavities (pleura, pericardium, peritoneum) | HIV infection | CD45 | PAX5 | Yes | Yes | Yes |
| | | Kaposi sarcoma | CD330 | CD20 | | | |
| | | No tumor masses | MUM1/IRF4 | CD138 | | | |
| | | | | Ig | | | |

ALK: *anaplastic lymphoma kinase*; DLBCL: diffuse large B-cell lymphoma; EBV: Epstein-Barr virus; HHV8: human herpesvirus 8; HIV: human immunodeficiency virus.

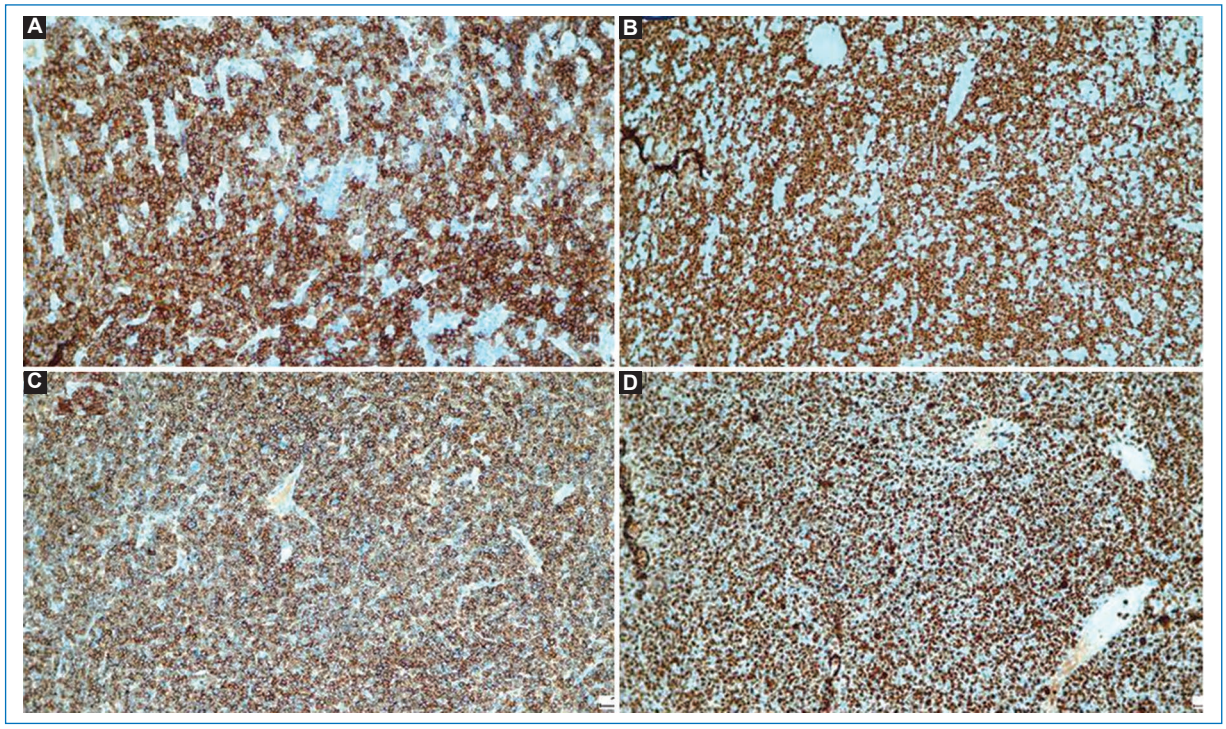


**Figure 3.** Immunohistochemical staining with positive markers. **A:** EMA. **B:** MUM-1. **C:** CD138. **D:** KI67, with a positivity index of 98%.


**Figure 4.** Patient's evolution after 3 treatment cycles. **A:** anterior aspect of the left hallux. **B:** posterolateral aspect of the left hallux.

B-cell markers and acquisition of plasma cell features, highlights its diagnostic complexity. This distinctive immunophenotype requires a rigorous diagnostic approach to differentiate PBL from other lymphomas and plasma cell neoplasms[5]. The strong association of PBL with HIV infection, and in many cases with Epstein-Barr virus[6], suggests a significant role of immunosuppression and viral infection in its pathogenesis. These etiologic factors raise important questions about the underlying molecular mechanisms and the possible vulnerability of certain individuals to this type of lymphoma[7].

From a therapeutic standpoint, treating PBL remains a challenge. Intensive chemotherapy has proven to be the cornerstone of treatment, although outcomes remain limited in terms of long-term survival[8]. Historically, the CHOP combination chemotherapy regimen (cyclophosphamide, doxorubicin, vincristine, and prednisone) has yielded poor results[1]. More recently, dose-adjusted EPOCH (DA-EPOCH: etoposide, prednisone, vincristine, cyclophosphamide, and doxorubicin) has been proposed as potentially more effective than CHOP for treating HIV-associated lymphomas. The addition of antiretroviral therapy to chemotherapy improves the prognosis in these patients[1]. The introduction of new therapeutic agents, such as bortezomib[9], a proteasome inhibitor, has opened new possibilities, though more studies are needed to establish its long-term efficacy and safety in PBL patients.

The prognosis is poor, with a relapse rate of 60% within the first year of treatment[10]. Without treatment, however, the median survival is only 3-4 months, while with treatment survival it ranges from 5 to 15 months, and an estimated one-quarter of patients achieve a cure[1]. The high mortality associated with PBL[11], especially in immunocompromised patients, underscores the urgent need for further research to improve management strategies and develop more effective therapies[2]. Studying the molecular pathways and mechanisms of resistance to current treatments could offer new opportunities for therapeutic interventions.

In conclusion, PBL is a complex neoplasm that requires a multidisciplinary approach for its management[12]. The combination of accurate diagnosis, personalized treatments, and a deeper understanding of its biology could significantly improve the prognosis for patients affected by this aggressive lymphoma.

## Funding

The authors declare that this work was carried out with their own resources.

## Conflicts of interest

The authors declare no conflicts of interest.

## Ethical considerations

**Protection of humans and animals.** The authors declare that the procedures followed conformed to the ethical standards of the responsible human experimentation committee and in accordance with the World Medical Association and the Declaration of Helsinki. The procedures were approved by the institution's Ethics Committee.

**Confidentiality, informed consent, and ethical approval.** The authors followed the confidentiality protocols of their institution, obtained informed consent from the patients, and obtained approval from the Ethics Committee. The recommendations of the SAGER guidelines were followed, according to the nature of the study.

**Declaration on the use of artificial intelligence.** The authors declare that no generative artificial intelligence tools were used in the writing of this manuscript.

## References

1. Fernández-Álvarez R, Sancho JM, Ribera JM. Linfoma plasmablástico. Med Clin (Barc). 2016;147:399–404.
2. García NM. Linfomas asociados a la infección por el virus de inmunodeficiencia humana (VIH): una serie de casos en La Paz, Bolivia. Gac Med Boliv. 2022;45:142–6.
3. Flümann R, Hansen J, Pelzer BW, Nieper P, Lohmann T, Kisis I, et al. Distinct genetically determined origins of Myd88/BCL2-driven aggressive lymphoma rationalize targeted therapeutic intervention strategies. Blood Cancer Discov. 2023;4:78–97.
4. Hashimoto M, Inaguma S, Kasai K, Kuwabara K, Noda N, Hayakawa M, et al. Plasmablastic lymphoma of the stomach in an HIV-negative patient. Pathol Int. 2012;62:763–70.

5. Inayat F, Virk HUH, Cheema AR, Saif MW. Plasmablastic lymphoma mimicking acute pancreatitis. Case Rep Oncol Med. 2016;2016: 9751736.
6. Assing K, Nielsen C, Jakobsen M, Andersen CB, Skogstrand K, Gaini S, et al. Potential anti-EBV effects associated with elevated interleukin-21 levels: a case report. BMC Infect Dis. 2020;20:878.
7. Joseph AA, Pulimood S, Manipadam MT, Viswabandya A, Sigamani E. Extramedullary plasmacytoma: an unusual neoplasm in a HIV-positive patient. Int J STD AIDS. 2016;27:909–11.
8. Khera R, Ahmed F, Murthy SS, Mallavarapu KM. Plasmablastic lymphoma of small intestine: a rare case report with review of literature. Indian J Hematol Blood Transfus. 2016;32:130–4.
9. Hashimoto N, Ueda T, Hiraiwa S, Tajiri T, Nakamura N, Yokoyama K. Clonally related plasmablastic lymphoma simultaneously occurring with diffuse large B-cell lymphoma. Case Rep Hematol. 2020;2020:1–6.
10. López A, Abrisqueta P. Plasmablastic lymphoma: current perspectives. Blood Lymphat Cancer. 2018;8:63–70.
11. Pérez-Mendoza A, Zárate-Guzmán AM, Lázaro-Pacheco IB, Navarrete-Pérez JJ. Plasmablastic lymphoma of the rectum, a rare cause of lower gastrointestinal bleeding: a case report. Rev Gastroenterol Mex. 2019;84:519–20.
12. Grijalva-Santana C, San Marcos-Romero D, Jiménez-Bobadilla B, Mendoza-Ramírez S, Reyes-Hansen D, Cosme-Reyes C. Linfoma plasmablástico anal: reporte de dos casos. Cir Gen. 2016;38:78–82.

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# Combined treatment of collagenase and the novel use of Nokor® needling in Dupuytren's contracture: a case report

## *Tratamiento combinado de colagenasa y el uso novedoso de la aguja Nokor® en la contactura de Dupuytren: a propósito de un caso*

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## Abstract

*Dupuytren's contracture (DC) significantly impacts hand function and quality of life in those affected. Common treatments include collagenase injections, percutaneous needle fasciectomy, and open surgical fasciectomy, but complications and recurrences are frequent. This article presents the first documented use of the Nokor™ needle, to prevent skin tearing during DC treatment with collagenase. The Nokor™ needle's precise subcision technique offers a less invasive alternative to fasciotomy, highlighting its potential role for dermatologists in managing DC effectively.*

***Keywords:*** *Dupuytren's contracture. Palmar fibromatosis. Collagenase. Admix needle. Subcision.*

## Resumen

*La contractura de Dupuytren (CD) impacta significativamente la función de la mano y la calidad de vida de quienes la padecen. Los tratamientos comunes incluyen inyecciones de colagenasa, fasciectomía percutánea con aguja y fasciectomía quirúrgica abierta, pero las complicaciones y recurrencias son frecuentes. Este artículo presenta el primer uso documentado de la aguja Nokor™ para prevenir desgarros cutáneos, durante el tratamiento con colagenasa, de la CD. La técnica de subincisión precisa de la aguja Nokor™ ofrece una alternativa menos invasiva a la fasciotomía, resaltando un papel importante para los dermatólogos en el manejo efectivo de la CD.*

***Palabras clave:*** *Contractura de Dupuytren. Fibromatosis palmar. Colagenasa. Aguja admix. Subincisión.*

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## Introduction

Dupuytren's contracture (DC) is an autosomal dominant disease of the palmar aponeurosis characterized by pathologic myofibroblasts forming nodules and bands due to deposition of collagen into digitopalmar fascia, resulting in flexion and/or deformity of the affected finger. DC's worldwide prevalence is 8%, and men are 3-4 times more affected than women[1]. This condition has a significant impact on a patient's quality of life and can significantly limit hand function. While there is no definitive cure for DC, there are several treatments available, including collagenase, percutaneous needle fasciectomy (PNF), and open surgical fasciectomy (OSF) which have been shown to be effective in relieving symptoms and improving hand function in some cases. However, PNF procedures can be challenging, particularly when severe adhesions between the nodules and overlying skin are present, increasing the risk of skin tears and other complications.

We present the case of a patient with severe long-standing DC, where the use of an admix needle subcision showed effectiveness in separating the nodule from the overlying skin and reduced the risk of the most common complication of the procedure (i.e., skin tearing).

This case highlights the importance of careful planning and technique selection in PNF procedures, particularly in cases of severe adhesions. We hope that sharing our experience will contribute to the broader understanding of effective surgical approaches for the management of DC and guide future treatment decisions in similar cases.

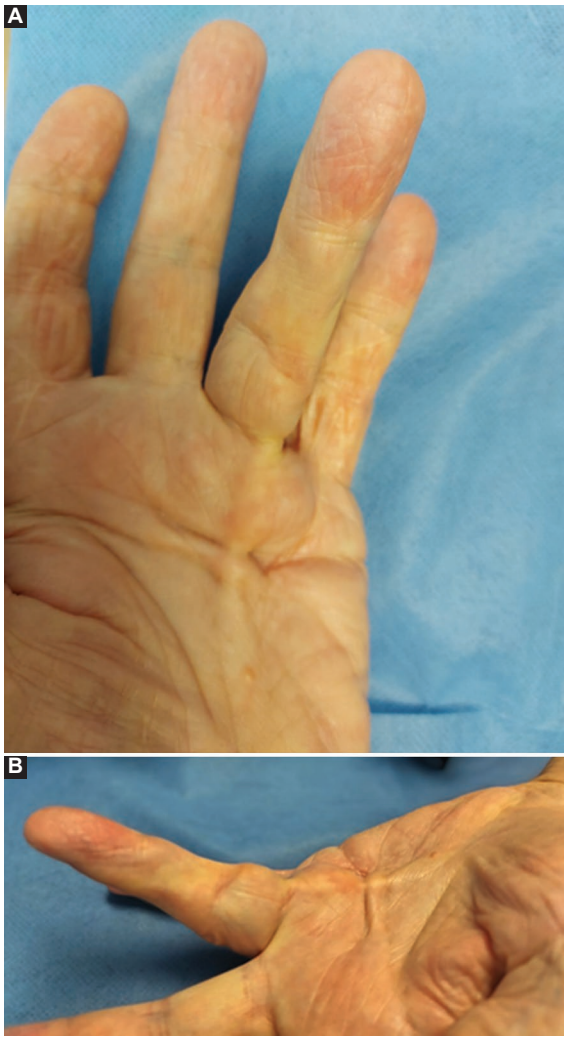


**Figure 1.** Patient's hands with noticeable thickening of the palm, visible nodules beneath the skin, and severe limitation of the extension of the affected fingers. **A:** left hand. **B:** right hand.

## Case presentation

An 84-year-old male patient was seen for a 30-year history of severe DC affecting both hands, particularly the 2nd, 4th, and 5th fingers (Fig. 1). After discussing treatment options, a decision was made to treat with collagenase and perform mechanical rupture of the nodules.

The procedure involved the application of a product containing a combination of 7.36% r-collagenase, 0.92% r-lipase, 0.92% r-lyase, and 0.05% hyaluronic acid (PB serum H.A. 1.5 High™, Proteos Biotech™, Albacete, Spain) 24 h before the rupture of the nodules. The kit was diluted at a minimum of 1.5 mL, and then approximately 0.3 mL of the solution was injected into each nodule. Both hands were bandaged in a neutral position, and the patient was advised not to mobilize them for the next 24 h.

After 24 h, under local anesthesia with 2% lidocaine, rupture was performed as previously described with other collagenase preparations[2]. Although digital nerve blocks are a viable option for percutaneous fasciectomy procedures, in this case, 2% lidocaine local anesthesia was chosen to minimize procedure time and due to the team's prior experience with this technique. It was noted that the local anesthetic was insufficient to completely numb the area, and the patient experienced some 6/10 level pain. In addition, the nodules were severely attached to the overlying skin, and their rupture resulted in a painful skin tear, a common

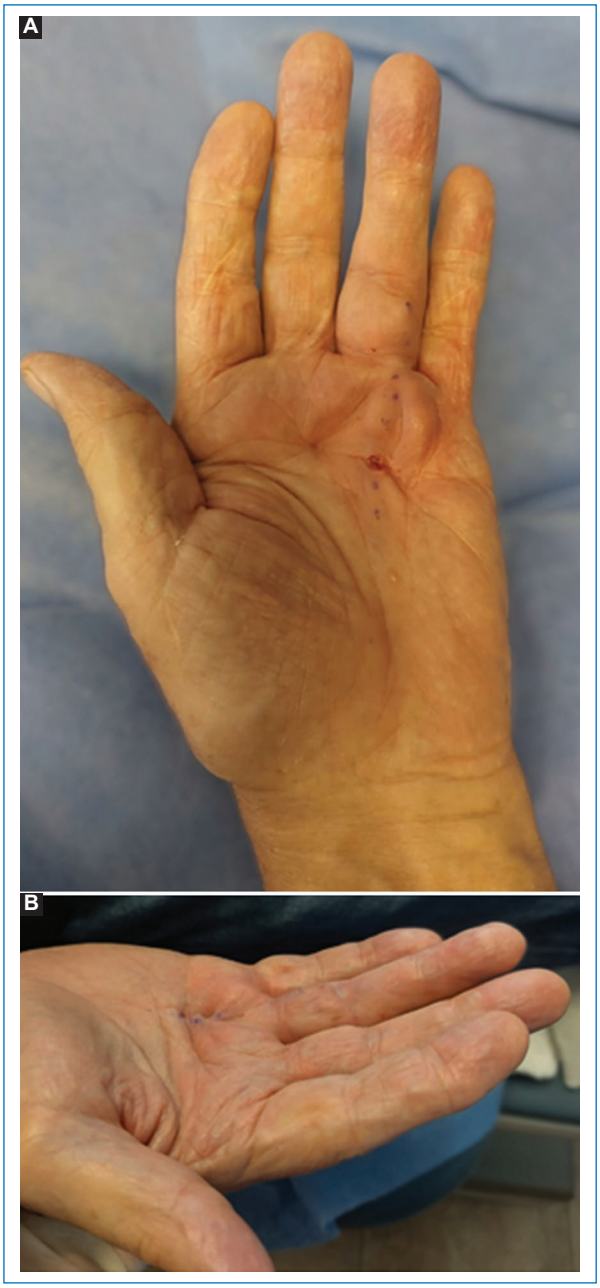


**Figure 2.** Patient's hands after collagenase injection, with 80% improvement in extension after the nodule rupture, and minor skin tear on the palmar aspect of 4th metacarpophalangeal joint. **A:** left hand with small skin tear. **B:** right hand.

complication of the procedure[3]. Despite these adverse events, the patient achieved an 80% improvement in the extension of all the fingers and expressed satisfaction with the results (Fig. 2).

Ten months after the first procedure, the patient returned and requested a second treatment to further improve his fingers' extension and hand mobility. Given the above-mentioned adverse events, it was decided to modify the classic treatment by performing subcision and needling of the nodules with an 18G admix scalpel-tipped needle (BD Nokor™ Filter Needle, Beckton Dickinson and Company, Franklin Lakes, New Jersey) as well as using collagenase. The collagenase preparation was injected as previously described. On the following day, under light sedation and the use of local injections of 2% lidocaine to the nodules and cords, subcision was first performed to separate the plane between the nodule and the overlying skin using the Nokor™ needle, puncturing at least 5 mm proximally to avoid areas of high tension and prevent causing tears. The use of the Nokor™ needle resulted in the complete freeing of the overlying skin. The needle was then used to puncture areas of maximum bowstringing, always communicating with the patient to avoid injury to the digital nerve. Flexing and extending the finger was also necessary to confirm that the needle was not in the flexor tendon. Finally, hyperextension of the joints was carried out by applying gentle extension tension on the cord until the rupture of the nodules was achieved, resulting in 100% release and extension of the fingers without causing skin tears. One week after the procedure the patient showed persistent improvement and was therefore discharged with 100% acceptance and satisfaction (Fig. 3).

## Discussion

DC is a multifactorial condition with a well-documented genetic predisposition, along with environmental and lifestyle factors that may influence its development. Several genetic mutations within aldehyde dehydrogenase 2, dihydrodiol dehydrogenase, and EPDR1 genes have been identified in patients with DC, suggesting an autosomal dominant inheritance in some cases. Other factors, such as alcohol consumption, smoking, and certain metabolic diseases, are also associated with an increased risk of developing DC[4]. It is one of the most prevalent diseases affecting the hand, varying from 5.7% to 11.7% globally[5]. This condition has a significant impact on a patient's quality of life and can significantly limit hand function. DC is characterized by pathologic myofibroblasts that produce a large amount of type III collagen[4], forming nodules and bands in the digitopalmar fascia, resulting in flexion and/or deformity of the affected finger[6].

Treatment of DC may be challenging since there is not a method that is completely effective, recurrences are common, and complications of the procedures may


**Figure 3.** Patient's hands 1 month after collagenase injections and the use of Nokor (TM) needle subcision for PNF, showing 100% release of the cords. **A** and **B:** right hand with 100% extension, **A** and **D:** left hand with 100% extension.

lead to further deterioration[4]. The most widely accepted treatments are collagenase injections, PNF, and OSF.

OSF is the most common treatment, but possible complications include persistent numbness, cold sensitivity, stiffness, and a recurrence rate at 5 years of 12-73%. PNF is a less invasive method, with more rapid recovery and a significantly lower cost, but its disadvantage is the shorter time to progression/recurrence (85%) compared to OSF[1].

*Clostridium histolyticum* collagenase (CHC), approved by the Food and Drug Administration (FDA), offers a non-surgical, office-based treatment option for adult patients with DC. This injectable therapy (Xiaflex®, Endo Pharmaceuticals Inc™, Malvern, Pennsylvania) contains two distinct collagenases that target different sites on the collagen molecule. AUX I, a class I CHC, cleaves the terminal ends of collagen fibers, whereas AUX II, a class II CHC, targets internal sections of the collagen. Together, these enzymes synergistically degrade collagen, thereby weakening the contracted cord and enhancing the elasticity and mobility of the affected tissue[7]. Given that Xiaflex™ is not available in Mexico, we used a product containing recombinant collagenases G (same as AUX I) and H (same as AUX II), but allegedly in a more effective proportion of 1:3, respectively, along with two other enzymes, r-lyase and r-lipase[8]. The effects of these differences are still to be proven in clinical trials. Another difference in this product is that the method of production uses *Escherichia coli* recombinant technology, which may result in a purer form of the enzymes. We tried to contact Proteos Biotech™ to provide the equivalence of H.A. 1.5 High™ with Xiaflex™ and to explain the differences, but up until the submission of this work, we did not receive additional information.

The use of collagenase for treating collagen disorders has been explored for many years. The collagenolytic properties of clostridial filtrates were first described in the 1940s, but it was not until 1965 that Bassot reported on "enzymatic fasciotomy"[9]. This procedure involved an injection of a mixture of trypsin, hyaluronidase, and lidocaine, followed by forced finger extension to rupture the cord, resulting in full finger extension in all patients within 15 min after the injection[10].

CHC injection is not complication-free, and its biggest disadvantage is that patients who receive this treatment have higher rates of recurrence in the longer term[4]. The most common complications of CHC injections are rupture of the skin at the injection site (26-31%) upon finger extension, neurapraxia (4.4%), hematoma (77%), and blood-filled blisters (22%)[1]. In our case, during the first course of treatment, our patient experienced a painful skin tear that took several weeks to heal.

Although CHC injections could enhance the patient's experience, the choice of treatment should be individualized based on case characteristics. The recurrence rate in patients treated with collagenase remains an important area of study, and future research could explore how integrating new techniques, such as the Nokor™ needle's precise subcision, might reduce complications and improve treatment efficacy.

In the case hereby presented, a helpful aid in the treatment to prevent this complication was the use of the Nokor™ needle, an off-label use of a medical device approved by the FDA as a "single-use medical device used for aspiration of fluids from vials and ampoules." The Nokor™ needle has a scalpel tip that makes it useful for performing subcision in many off-label clinical situations such as atrophic acne scars, wrinkles, striae[11], and cellulite[12]. In the case of DC, it allows for very precise detachment of the skin overlying the nodules and by freeing it, allows for soft gliding on top of the cord, rather than forcibly stretching it when the hyperextension is performed.

Although DC is mostly seen by hand surgeons, dermatologists have an important role in the diagnosis of this ailment and, as is seen in this case, may also provide the patient with a much less invasive procedure than fasciotomy. Furthermore, the use of collagenase injections has proven to be a very effective method.

To our knowledge, this is the first published article on the use of the Nokor® needle to prevent skin tearing. It may be a useful aid in the treatment of DC for dermatologists after collagenase injections and nodule fragmentation.

## Conclusion

This case demonstrates the innovative use of the Nokor™ needle to enhance collagenase treatment in Dupuytren's contracture by performing subcision, which differs from traditional needling techniques. This method effectively separates the skin from the nodule, preventing tears and minimizing complications, while achieving significant functional improvement. By offering a less invasive alternative to traditional surgical methods, this approach highlights a potential role for dermatologists in managing this challenging condition. Further exploration of such techniques may improve outcomes and broaden treatment options for patients with severe Dupuytren's contracture.

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The authors declare that this work was carried out with the authors' own resources.

## Conflicts of interest

The authors declare that they have no conflicts of interest.

## Ethical considerations

**Protection of humans and animals.** The authors declare that no experiments involving humans or animals were conducted for this research.

**Confidentiality, informed consent, and ethical approval.** The authors have followed their institution's confidentiality protocols, obtained informed consent from patients, and received approval from the Ethics Committee. The SAGER guidelines were followed according to the nature of the study.

**Declaration on the use of artificial intelligence.** The authors declare that no generative artificial intelligence was used in the writing of this manuscript.

## References

1. Ruettermann M, Hermann RM, Khatib-Chahidi K, Werker PM. Dupuytren's disease-etiology and treatment. Dtsch Arztebl Int. 2021;118:781-8.
2. Hurst LC, Badalamente MA, Hentz VR, Hotchkiss RN, Kaplan RT, Meals RA, et al. Injectable collagenase clostridium histolyticum for Dupuytren's contracture. N Engl J Med. 2009;361:968-79.
3. Legato JM, Gill MK, Coutelle NA, Nydick JA. Outcomes following repeat collagenase treatment of Dupuytren contracture. J Hand Surg Am. 2023;48(10):S0363-5023(23)00180-6.
4. Żyluk A. Dupuytren's disease - what's new: a review. Pol Przegl Chir. 2022;95:53-61.
5. Steppe C, Cinclair R, Lies S. A 10-year review of collagenase versus fasciectomy in the treatment of dupuytren contracture. Ann Plast Surg. 2024;92:642-6.
6. Peimer CA, Hurley D, Latch T, Scott S, Pess GM. Incidence of tendon rupture after collagenase *Clostridium histolyticum* injection for treatment of dupuytren contracture in adults: a postmarketing safety analysis. J Hand Surg Glob Online. 2023;5:33-8.
7. Gilpin D, Coleman S, Hall S, Houston A, Karrasch J, Jones N. Injectable collagenase *Clostridium histolyticum*: a new nonsurgical treatment for Dupuytren's disease. J Hand Surg Am. 2010;35:2027-38.e1.
8. Muñoz-Montaño JR. Uso de la colagenasa G recombinante, colagenasa H recombinante y PZ-peptidasa recombinante, para el tratamiento de enfermedades que cursan con alteraciones del colágeno (Patente No. ES2385239B1). España: Oficina Española de Patentes y Marcas; 2013.
9. Starkweather KD, Lattuga S, Hurst LC, Badalamente MA, Guilak F, Sampson SP, et al. Collagenase in the treatment of Dupuytren's disease: an *in vitro* study. J Hand Surg Am. 1996;21:490-5.
10. Bassot J. Treatment of Dupuytre's disease by isolated pharmacodynamic "exceresis" or "exceresis completed by a solely cutaneous plastic step. Lille Chir. 1965;20:38-44.
11. Khunger N, Khunger M. Subcision for depressed facial scars made easy using a simple modification. Dermatol Surg. 2011;37:514-7.
12. Hexsel DM, Mazzuco R. Subcision: a treatment for cellulite. Int J Dermatol. 2000;39:539-44.

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CLINICAL CASE

# Clouston syndrome: case report and diagnostic approach to pachyonychia in pediatrics

## *Síndrome de Clouston: reporte de caso y abordaje diagnóstico de las paquioniquias en pediatría*

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## Abstract

*Clouston syndrome or hypohidrotic ectodermal dysplasia is a rare autosomal dominant disorder caused by a mutation in the GJB6 gene. Its main clinical features include the triad of palmoplantar keratoderma, nail dystrophy, and hypotrichosis. A pedia- tric patient with nail and hair abnormalities was diagnosed clinically and genetically with this condition, and a diagnostic algorithm for approaching causes of pachyonychia in pediatric patients is presented. The importance of genetic counselling and appropriate differential diagnosis underscore the need for early clinical suspicion to initiate suitable therapeutic and supportive interventions.*

***Keywords:*** *Ectodermal dysplasia. Nail diseases. Hypotrichosis. Genetic counselling.*

## Resumen

*El síndrome de Clouston o displasia ectodérmica hidrótica es una rara enfermedad autosómica dominante, causada por una mutación en el gen GJB6. Sus principales hallazgos clínicos incluyen la tríada de queratodermia palmo-plantar, distrofia ungueal e hipotricosis. Se reporta el caso de una paciente pediátrica con alteraciones ungulares y del pelo, con diagnóstico clínico y genético de esta patología, y se presenta un algoritmo diagnóstico para el abordaje de las causas de paquioniquia en la edad pediátrica. La importancia de la consejería genética y el diagnóstico diferencial adecuado resaltan la necesidad de una sospecha clínica temprana para iniciar intervenciones terapéuticas y de apoyo adecuadas.*

***Palabras clave:*** *Displasia ectodérmica. Enfermedades de las uñas. Hipotricosis. Consejería genética.*

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## Introduction

Hidrotic ectodermal dysplasia, or Clouston syndrome, is a rare autosomal dominant disorder caused by a mutation in the *GJB6* gene[1]. Its main clinical features include the triad of palmoplantar keratoderma, nail dystrophy, and hypotrichosis.

The nails typically become thicker and more dystrophic over time, which is an essential feature of the syndrome, and in approximately 30% of affected individuals, nail dystrophy may be the only suggestive sign[2]. Very often, such changes may be misdiagnosed as onychomycosis or pachyonychia congenita[1].

## Case report

A 5-year-old girl from Cali, Colombia, with no relevant past medical history, born from a full-term first pregnancy to non-consanguineous parents. The patient was seen at a tertiary referral center pediatric dermatology service with a 2-year history of thickening of all fingernails and toenails. She had been managed by a primary care physician as a case of onychomycosis, without mycological studies, using oral antifungals without improvement. Additionally, the patient presented with diffuse hair loss on the scalp and eyebrows, with no treatment administered for this condition.

The clinical examination revealed phototype II, and involvement of all 20 nail plates as evidenced by thickening, subungual hyperkeratosis, and pincer nail deformity, particularly in the fingernails. Furthermore, sparse blond scalp hair and hypotrichosis of the eyebrows and eyelashes were observed (Fig. 1).

Initially, universal alopecia areata was considered as a differential diagnosis, but due to the clinical findings and chronic nature of the condition, a medical genetics evaluation was requested. A whole exome sequencing (NGS-WES) was performed, revealing a heterozygous variant related to the described symptoms in the *GJB6* gene, categorized as pathogenic according to the American College of Medical Genetics and Genomics criteria. Based on this result, a diagnosis of Clouston syndrome was established. Genetic counseling was provided, and treatment with keratolytics was initiated to reduce the thickening of the nail plates, resulting in an improved aesthetic appearance of the nails.

## Discussion

Ectodermal dysplasias are genetic conditions that affect the development or homeostasis of 2 or more tissues of ectodermal origin, including hair, teeth, and certain glands, with or without involvement of other tissues and organs. Multiple genes and pathways involved in the development of complex molecular structures necessary for the formation, structure, and normal function of ectodermal derivatives are involved[3]. They are considered rare diseases, with an estimated incidence rate of 7 per 10,000 births and a prevalence of 1-9 per 100,000[4].

They can be categorized as hypohidrotic or hidrotic. Hypohidrotic ectodermal dysplasia is characterized by fine, sparse, or absent hair, missing or conical teeth, and marked reduction in sweating, along with normal nails[4]. In contrast, hidrotic ectodermal dysplasia or Clouston syndrome is a rare autosomal dominant disease caused by a mutation in the *GJB6* gene. This gene is located on the long arm of chromosome 13 and encodes connexin 30, which is part of a group of proteins that enable the transport of nutrients, ions, and signaling molecules, and is present in various tissues of the body, especially the palms, soles, hair follicles, and nails. The disease has complete penetrance but highly variable expressivity, even among affected individuals from the same family[5].

Clinical findings include the triad of palmoplantar keratoderma, nail dystrophy, and hypotrichosis. Palmoplantar keratoderma may develop during childhood and worsen with age, being a common but not universal finding[6]. The nails are typically thick and dystrophic over time, representing an essential feature of the syndrome and the only suggestive sign in up to 30% of patients. However, they can be mistaken for onychomycosis or congenital paronychia. Nail abnormalities reported include thickening or shortening (micronychia), onychorrhexis, or triangular nail plates that may resemble pachyonychia congenita. In fact, most cases of Clouston syndrome have thickened and hyperconvex nails. Pincer nails have also been described, as in the present case[7,8]. Hypotrichosis manifests as fine, slow-growing hair, sparse or absent eyebrows and eyelashes, as well as scant pubic and axillary hair[1,2].

Other ectodermal anomalies have also been reported, such as hyperpigmentation of the skin over large joints, ocular abnormalities, skeletal disorders, sensorineural hearing loss, and intellectual disability[2-9]. There are few reports linking this condition to benign neoplasms, such as eccrine syringocystadenoma, or malignant tumors, such as squamous cell carcinoma of the skin, nail bed, or trachea. One report describes a cuniculatum epithelioma associated with conditions involving palmoplantar keratoderma[5-10].

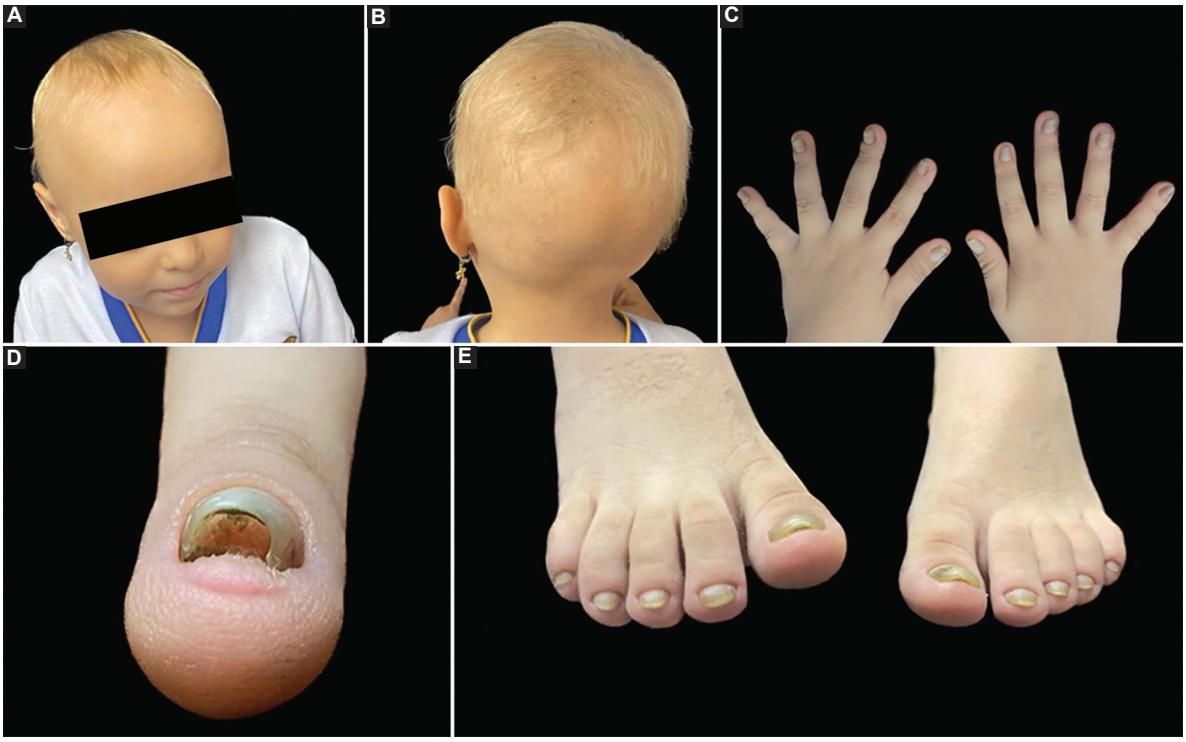


**Figure 1.** Clinical findings in the patient. **A:** hypotrichosis of the scalp, eyebrows, and eyelashes (frontal view). **B:** hypotrichosis of the scalp (posterior view). **C:** thickening and xanthonychia of all 10 fingernails. **D:** detail of pincer nail deformity and subungual hyperkeratosis on one finger. **E:** xanthonychia and thickening of all 10 toenails.

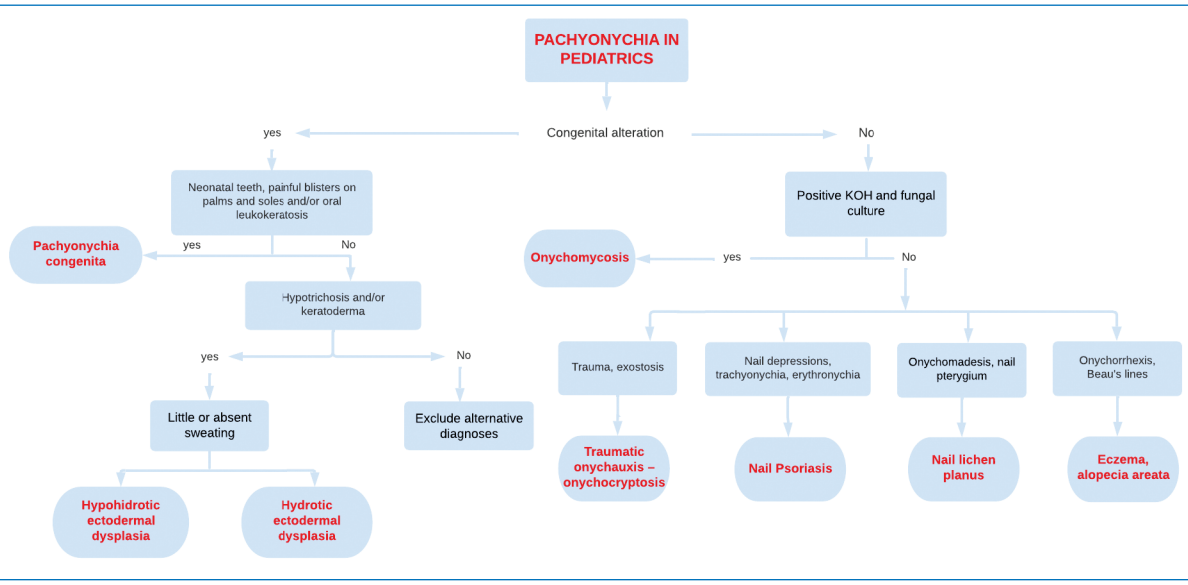


**Figure 2.** Diagnostic algorithm for pediatric patients with pachyonychia *(developed by the authors).*

The degree of alopecia and hyperkeratosis appears to vary, with some families described without the latter. Dental and eccrine gland functions are normal[1].

Diagnosis is clinical, supported by genetic testing to identify variants in the affected gene[2-11]. However, a methodical differential diagnostic process is essential

when evaluating pachyonychia associated with congenital abnormalities or genodermatoses. Figure 2 proposes a practical diagnostic algorithm for the initial assessment and study of pachyonychia in pediatric patients.

Currently, there is no cure for the disease, and management is primarily supportive. A multidisciplinary follow-up approach is important to monitor clinical features and provide emotional and psychological support, along with genetic counseling[2-12].

Treatment options for nail plates and palmoplantar hyperkeratosis include topical emollients and keratolytics (urea- or lactic acid-based), and nail matrix ablation. There are reports of topical minoxidil use with improvement in alopecia[13]. Physical and cosmetic options may also be proposed, such as wigs for scalp hair, eyebrow tattoos, and artificial nails[12-14].

## Conclusions

This is a case of a rare disease known as Clouston syndrome, which may clinically resemble more common pediatric conditions such as alopecia areata, onychomycosis, and other conditions involving cutaneous appendages or genodermatoses. However, awareness of this condition enables dermatologists to suspect it and refer the patient to a geneticist in a timely manner for appropriate studies, allowing early supportive management to improve the patients' quality of life.

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The authors declare that this work was conducted with personal resources.

## Conflicts of interest

The authors declared no conflicts of interest whatsoever.

## Ethical considerations

**Protection of humans and animals.** The authors declare that no experiments on humans or animals were conducted for this research.

**Confidentiality, informed consent, and ethics approval.** The authors have followed their institution's confidentiality protocols, obtained informed consent from the patients, and have approval from the Ethics Committee. The recommendations of the SAGER guidelines have been followed according to the nature of the study.

**Statement on the use of artificial intelligence.** The authors declare that no generative artificial intelligence was used in writing this manuscript.

## References

1. van Steensel MAM, van Geel M, Steijlen PM, Jonkman MF, McLean WH, Smith FJ. Clouston syndrome can mimic pachyonychia congenita. J Invest Dermatol. 2003;121:1035–8.
2. Sanches S, Rebellato PR, Fabre AB, de Campos GL. Do you know this syndrome? Clouston syndrome. An Bras Dermatol. 2017;92:417–8.
3. Wright JT, Fete M, Schneider H, Zinser M, Koster MI, Clarke AJ, et al. Ectodermal dysplasias: classification and organization by phenotype, genotype and molecular pathway. Am J Med Genet A. 2019;179:442–7.
4. García MP, Hernández M, Torrelo A. Displasias ectodérmicas: revisión clínica y molecular. Actas Dermosifiliogr. 2013;104:451–70.
5. de Andrade AC, Vieira DC, Harris OM, Pithon MM. Clouston syndrome associated with eccrine syringofibroadenoma. An Bras Dermatol. 2014;89:504–6.
6. Kantaputra P, Intachai W, Kawasaki K, Ohazama A, Carlson B, Quarto N, et al. Clouston syndrome with pili canaliculi, pili torti, overgrown hyponychium, onycholysis, taurodontism and absence of palmoplantar keratoderma. J Dermatol. 2020;47:230–2.
7. Sukakul T, Yang HS, Onoufriadis A, Hsu CK, McGrath JA. Pterygium and thinning of nails as an unusual manifestation in Clouston syndrome. J Dermatol. 2019;46:329–30.
8. Hu YH, Lin YC, Hwu WL, Lee YM. Pincer nail deformity as the main manifestation of Clouston syndrome. Br J Dermatol. 2015;173:581–3.
9. Arroyo Trídico L, Antonio JR, de Oliveira Pozetti EM, Mendes Rosa AM, Antonio CR. Clouston syndrome: 25-year follow-up of a patient. An Bras Dermatol. 2015;90:897–9.
10. Herrera SP, Santacoloma G. Displasia ectodérmica hidrótica y epitelioma cuniculado. Rev Asoc Colomb Dermatol Cir Dermatol. 2014;22:340–2.
11. Mellerio J, Greenblatt D. Hidrotic ectodermal dysplasia 2. In: Adam MP, Feldman J, Mirzaa GM, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington; 1993–2024.
12. Cammarata SF, Rinelli M, Pisaneschi E, Diociaiuti A, Willoughby CE, Avendaño A, et al. Novel clinical features associated with Clouston syndrome. Int J Dermatol. 2019;58:143–6.
13. Lee HE, Chang IK, Im M, Seo YJ, Lee JH, Lee Y. Topical minoxidil treatment for congenital alopecia in hypohidrotic ectodermal dysplasia. J Am Acad Dermatol. 2013;68:139–40.
14. Murshidi R, Al-Lala H. Clouston syndrome: report of a Jordanian family with GJB6 gene mutation. Case Rep Dermatol Med. 2023;2023:5577379.

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CASE FOR DIAGNOSIS

# Abdominal fistula mimicking a skin tumor

## *Fístula abdominal simulando un tumor cutáneo*

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## Case report

A 48-year-old man with a past medical history of perihepatic abscesses presented to the emergency department with a 2-week history of an exophytic lesion developing over a prior drainage scar in the right costal region. He exhibited no signs of intra-abdominal disease (Fig. 1). Differential diagnosis of the rapidly growing ulcerated tumor included cutaneous neoplasms with similar clinical presentations, such as amelanotic melanoma, squamous cell carcinoma arising from scar tissue, or pyogenic granuloma. Given the history of abdominal abscesses, the possibility of a cutaneous fistula was also considered.

Five days after excision and into antibiotic therapy, the lesion recurred with purulent drainage. Ultrasound and computed tomography identified an abdominal cutaneous fistula with a perihepatic collection (Fig. 2). The patient was, then, referred to general surgery.

Refractory abdominocutaneous fistulas often require surgical exploration and excision, with a reported mortality rate of 11%-21%. To reduce this risk, minimally invasive techniques – such as fibrin sealant injection or collagen plug placement – are increasingly being used[1].

Cutaneous fistulas can present as pseudotumoral lesions[2], requiring imaging modalities for accurate diagnosis, tract evaluation, and treatment planning[3].

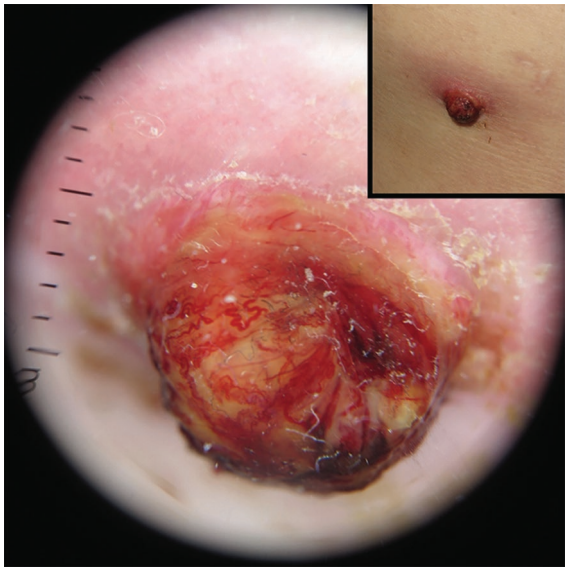

**Figure 1.** Soft exophytic lesion approximately 10 mm in diameter with a collarette border, located over a prior drainage scar. Dermoscopy of the lesion shows spiral vessels and yellowish areas.

## Funding

The authors declare that this work was conducted with their own resources.

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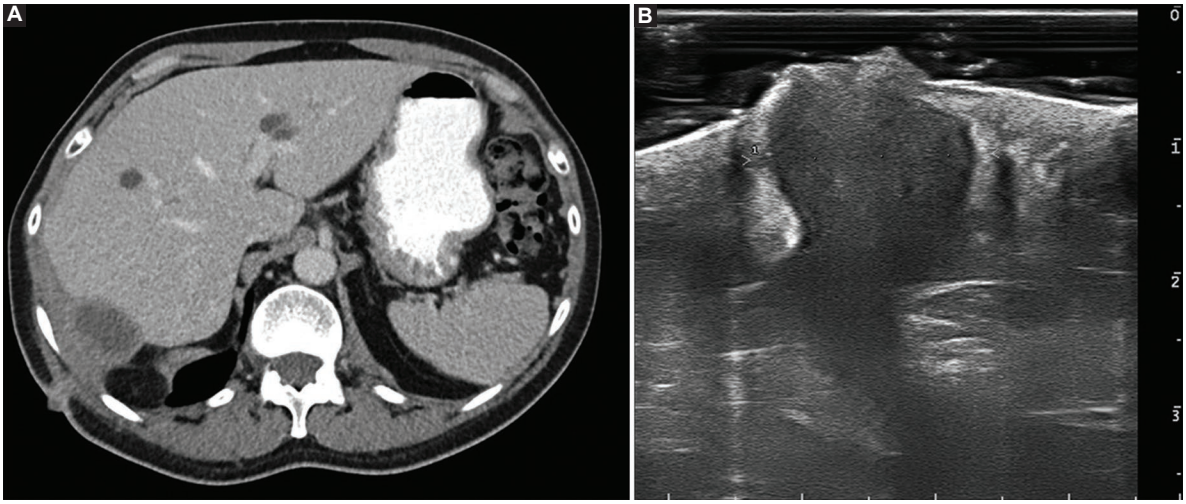


**Figure 2. A:** imagen de tomografía computarizada que muestra tanto el trayecto fistuloso como una colección perihepática posterior derecha de 48 × 34 × 54 mm. La fístula se extiende desde el tejido subcutáneo hasta la piel. **B:** imagen ecográfica que revela la presencia de una fístula cutánea originada en un sitio de drenaje previo, extendiéndose más allá del tejido subcutáneo.

## Conflicts of interest

The authors declare no conflicts of interest.

## Ethical considerations

**Protection of humans and animals.** The authors state that no experiments were conducted on humans or animals for this research.

**Confidentiality, informed consent, and ethical approval.** The authors followed their institution's confidentiality protocols, obtained informed consent from the patients, and have approval from the Ethics Committee.

The authors followed the recommendations of the SAGER guidelines according to the nature of the study.

**Declaration on the use of artificial intelligence**. The authors declare that no generative artificial intelligence tools were used in the writing of this manuscript.

## References

1. Lomis NN, Miller FJ, Loftus TJ, Whiting JH, Giuliano AW, Yoon HC. Refractory abdominal-cutaneous fistulas or leaks: percutaneous management with a collagen plug. J Am Coll Surg. 2000;190:588–92.
2. Luque-Luna M, Alamon-Reig F, Morgado-Carrasco D. Odontogenic fistula. JAMA Dermatol. 2022;158:813.
3. Frick MP, Feinberg SB, Stenlund RR, Gedgaudas E. Evaluation of abdominal fistulas with computed body tomography (CT). Comput Radiol. 1982;6:17–25.