CASE REPORT

Total dystrophic onychomycosis of all 20 nails: A case in a patient with down syndrome – Case report

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SUMMARY

The prevalence of onychomycosis in adults ranges from 2% to 14%, with dermatophytes being the primary etiologic agents. In patients with Down syndrome (DS), a higher prevalence of this condition has been documented compared to the general population. DS patients exhibit both cellular and humoral immune dysfunction, which facilitates the colonization and proliferation of pathogens in the nails, increasing susceptibility to fungal infections. We present the case of a 21-year-old female patient diagnosed with trisomy 21, who exhibited thickening of all 20 nails with subungual hyperkeratosis. Mycological examination with potassium hydroxide (KOH) revealed the presence of hyphae, and the culture tested positive for Trichophyton rubrum. The treatment of onychomycosis in DS patients requires a comprehensive approach, considering immune system dysfunction and the need for prolonged therapeutic management to achieve an effective response and prevent recurrence.

INTRODUCTION

Onychomycosis is a nail disease primarily characterized by subungual hyperkeratosis and onycholysis.[1] The prevalence of onychomycosis in the general adult population is estimated to range from 2% to 14%, whereas it is less common in children (0.2% to 2.6%).[2] The main etiologic agents include dermatophytes (60-70%), primarily Trichophyton rubrum and Trichophyton mentagrophytes; non-dermatophyte molds (Scopulariopsis brevicaulis, Acremonium spp., Aspergillus spp., Fusarium spp., and Neoscytalidium); and yeasts such as Candida spp.[3] In patients with Down syndrome, a multisystem disorder caused by trisomy of chromosome 21, a higher prevalence of onychomycosis has been observed compared to the general population.[4]

CASE PRESENTATION

A 21-year-old female patient presented with thickened, yellowish nails on all 20 digits, along with subungual hyperkeratosis (Figure 1). She has had a diagnosis of trisomy 21 (Down syndrome) since birth. The patient reported the onset of the current condition at the age of 10, with no prior treatment.

A mycological study was performed: direct examination with potassium hydroxide (KOH) was positive, revealing hyphae (Figure 2), and culture showed white, cottony colonies with slight reddish pigmentation on the reverse side (Figure 3). A diagnosis of total dystrophic onychomycosis caused by Trichophyton rubrum was established.

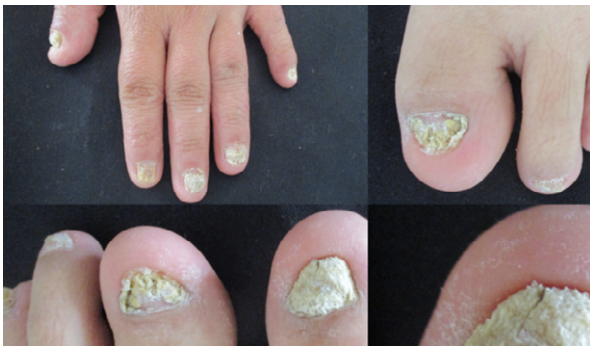

Figure 1. Total dystrophic onychomycosis affecting all 20 nails

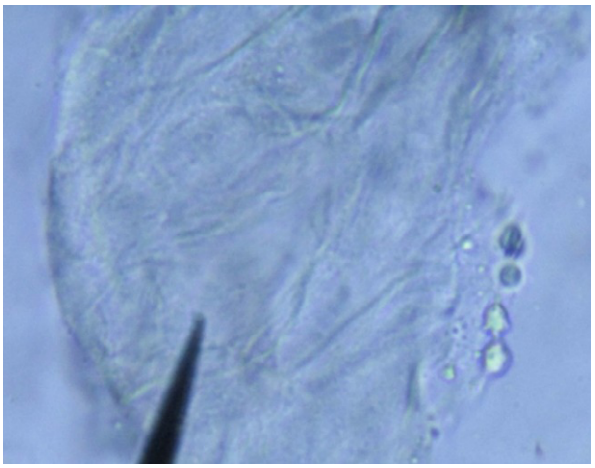

Figure 2. Direct examination showing the presence of hyphae.

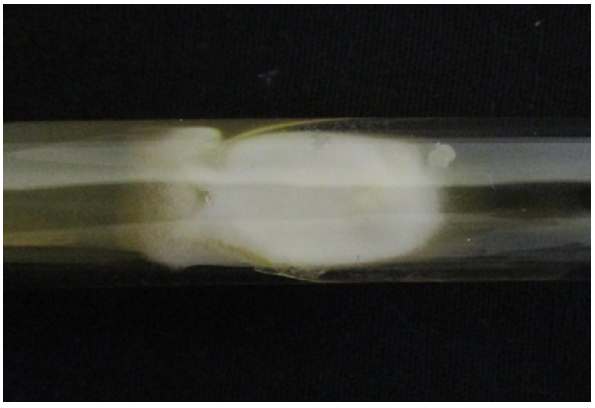

Figure 3. T. rubrum culture.

DISCUSSION

Down syndrome (DS) is the most common chromosomal abnormality among live-born children and is associated with various neurological, cardiac, and gastrointestinal anomalies.[4] From a dermatological perspective, patients with DS have a higher predisposition to develop skin infections such as impetigo and folliculitis, as well as autoimmune dermatoses such as vitiligo and alopecia areata.[5]

Immune dysfunction in DS affects both cellular and humoral immunity, reducing the ability to mount an effective immune response against fungal infections.[4] This facilitates the colonization and proliferation of pathogens in the nails and contributes to other inflammatory skin disorders.

Studies on lymphocyte subpopulations in children with DS have shown a significant decrease in transitional and naïve B lymphocytes, as well as CD3-CD16 and NK 56+ T cells, contributing to a high incidence of dysgammaglobulinemia.[6] This is attributed to various immunological alterations, including T and B lymphocyte dysfunction, immunoglobulin G deficiency, impaired opsonization, decreased interleukin production, and reduced CD3 expression, leading to defective chemotaxis and phagocytosis.[7]

The immune dysfunction observed in DS may be related to the overexpression of certain genes located on chromosome 21, such as SOD1 and RCAN1. Overexpression of the SOD1 (superoxide dismutase) gene may contribute to an imbalance in oxidative stress regulation, negatively affecting immune function. Additionally, overexpression of RCAN1 (regulator of calcineurin 1) may interfere with intracellular signaling required for an adequate immune response. These mechanisms, along with nutritional deficiencies such as zinc deficiency, further exacerbate immunodeficiency in individuals with DS, making them more susceptible to infections like onychomycosis.[4]

In DS patients, an increased prevalence of onychomycosis has been reported, estimated to range between 30-70% compared to 5-10% in the general population.[8] In a retrospective study by Rork et al. (2020) conducted in pediatric DS patients, onychomycosis was identified in 9.9% of cases.[9] A study on the Mexican DS population conducted in 2000 by Córdova and colleagues found that among 55 patients

with a clinical diagnosis of onychomycosis and tinea pedis, 48 cases (87.3%) tested positive via direct examination and culture, with Trichophyton rubrum being the most common causative agent.[10]

The treatment of onychomycosis in patients with Down syndrome (DS) should be approached comprehensively, considering both the selection of the most effective antifungal agent and the patient's immunological comorbidities. Onychomycosis is inherently difficult to treat due to the nature of the fungal pathogen, making prolonged treatment necessary.[11] Terbinafine and itraconazole are the most commonly used therapeutic options, with continuous-dose terbinafine being the treatment of choice due to its high mycological cure rate and lower incidence of adverse effects. A meta-analysis showed a 76% mycological cure rate for continuous-dose terbinafine, compared to 63% for pulse-dose itraconazole and 59% for continuous-dose itraconazole.[1,12]

In addition to systemic antifungals, the management of onychomycosis in DS patients may benefit from adjunctive treatments that enhance immune response. Therapies aimed at disrupting fungal biofilms, such as antimicrobial photodynamic therapy and low-frequency surface acoustic waves, have shown promising results. These techniques may improve the effectiveness of traditional antifungals by destabilizing biofilms that protect fungi from treatments.[12]

The recurrence rate after completing treatment is approximately 20-25%, which may be attributed to multiple factors such as age, genetic predisposition, occupation, living conditions, and climate.[13,14] Additionally, a systematic review by Stewart et al. demonstrated that onychomycosis has a negative impact on patients' quality of life.[15]

It is essential to emphasize education on personal care and hygiene, as proper hygiene practices can reduce the incidence of nail infections. Patients and caregivers should be advised to wear appropriate footwear, keep nails short and dry, and avoid humid environments that promote fungal growth.

CONCLUSION

In conclusion, onychomycosis in DS patients represents a significant clinical challenge due to their underlying immunodeficiency. This case underscores the need for comprehensive and personalized care to address the complexities of onychomycosis in a vulnerable population such as patients with DS.

INFORMED CONSENT

The patient included in this study has signed the informed consent, approving the use of her images and clinical data exclusively for research and scientific publication purposes. It is guaranteed that no personal data has been disclosed and that no photographs allowing her identification have been used.

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