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Case Report

Onychomatricoma Associated with Tuberous Sclerosis: A Case Report

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Abstract: Onychomatricoma (OM) is a rare benign fibroepithelial tumor originating from the nail matrix that often mimics onychomycosis both clinically and mycologically, leading to delayed diagnosis. We report the case of a 23-year-old male with a history of tuberous sclerosis who developed a solitary nail lesion on the second toe of the left foot. The lesion was initially misdiagnosed as onychomycosis and treated unsuccessfully. Dermoscopic findings suggested onychomatricoma, which was later confirmed by histopathological examination following total nail avulsion. The patient showed favorable evolution without recurrence. This represents the second reported case of an association between onychomatricoma and tuberous sclerosis. The case highlights the importance of comprehensive nail evaluation in patients with genodermatoses.

Keywords: Onychomatricoma, Tuberous Sclerosis, Nail Dystrophy, Dermoscopy, Onychomycosis.

INTRODUCTION

Onychomatricoma is a rare benign fibroepithelial tumor of the nail matrix first described in 1992 [1]. Due to its variable clinical presentation, it is frequently underdiagnosed or mistaken for other causes of nail dystrophy, particularly onychomycosis. Dermoscopy is a useful diagnostic tool, especially when the characteristic tetrad is present: nail plate thickening, xanthonychia, increased transverse curvature, and splinter hemorrhages [1]. However, histopathological examination remains the gold standard for diagnosis. The association between onychomatricoma and tuberous sclerosis is extremely rare and poorly understood, making this case clinically relevant.

CASE REPORT

We report the case of a 23-year-old male patient who presented to the dermatology department with a localized dermatosis affecting the left lower extremity, involving the nail apparatus of the second toe of the left foot. The dermatosis was characterized by xanthonychia, whitish streaks, thickening of the nail plate, increased transverse curvature of the nail plate, and deviation of the nail axis (Figure 1 A and B).

On further examination of the skin and its appendages, the patient also presented a dermatosis localized to the head, involving the face at the level of the nose and cheeks, characterized by multiple erythematous hemispherical papules measuring 1 to 3 mm in diameter.

The patient reported a 2-year history of nail changes affecting the second toe of the left foot, initially asymptomatic but later associated with pain when wearing shoes and walking. A previous direct mycological examination was positive, and the patient received both topical and systemic antifungal treatment without improvement.

His medical history was significant for a diagnosis of tuberous sclerosis at 3 months of age, mild intellectual disability, and epilepsy under treatment.

Nail dermoscopy revealed xanthonychia, splinter hemorrhages, honeycomb-like cavities, and digitiform projections emerging from the matrix (Figure 2 A, B, and C).

A radiograph of the left foot was performed and showed no bone abnormalities.

Based on these findings, surgical treatment was decided with a presumptive diagnosis of onychomatricoma. Total nail avulsion was performed, revealing a filamentous tumor arising from the nail matrix with projections (Figure 3A & B).

Histopathological examination showed epidermis with absence of the granular layer and elongated digitiform intraepithelial projections forming dermal islands. On higher magnification, a digitiform pattern with spindle-shaped cells was observed (Figure 4 A and B). The stroma adjacent to the proliferated matrix epithelium was collagenized and hypervascular. Cross-sections of the nail plate showed hyperkeratosis, empty cavities, and others filled with serous material (Figure 5 A and B).

Following surgical treatment, the patient showed no signs of recurrence during follow-up.

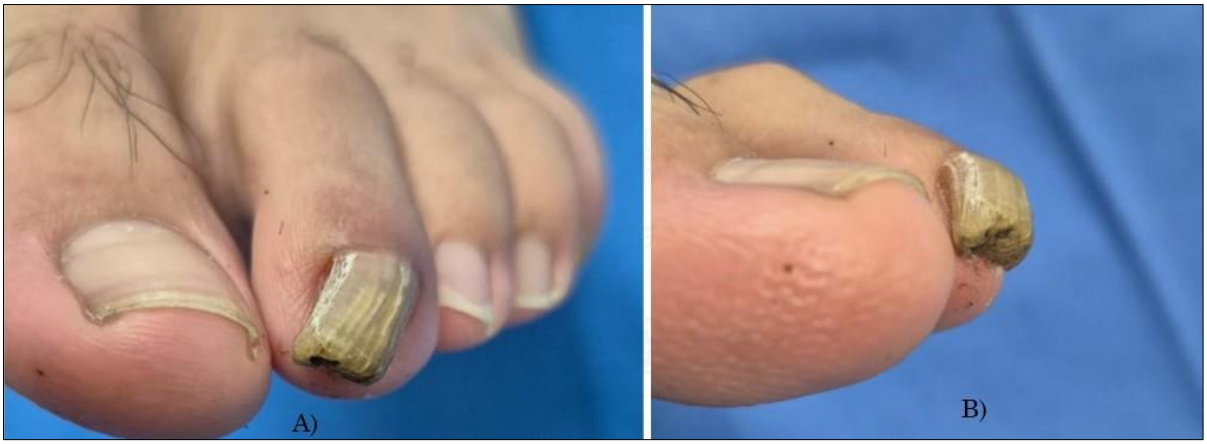

Figure 1 A and B: Dermatosis localized to the left lower extremity, involving the second toe at the level of the nail unit. A) Deviation of the nail axis, xanthonychia, splinter hemorrhages, and whitish and yellowish longitudinal streaks. B) Thickening of the nail plate, xanthonychia, woodworm-like cavities, transverse overcurvature of the plate, and whitish longitudinal streaks

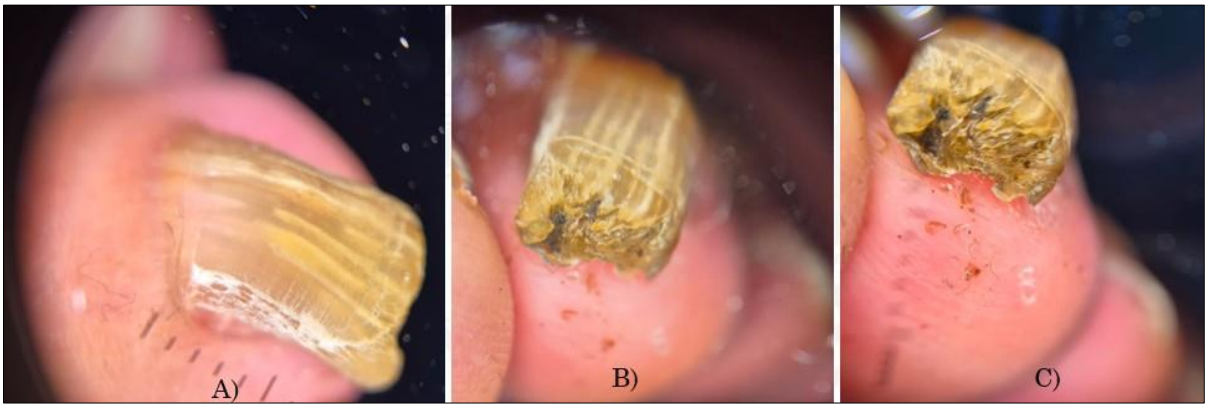

Figure 2 A and B: Dermoscopy showing: A) Xanthonychia and whitish and yellowish longitudinal streaks. B) Splinter hemorrhages and digitiform projections. C) Honeycomb-like cavities with yellow and black coloration, in addition to digitiform projections

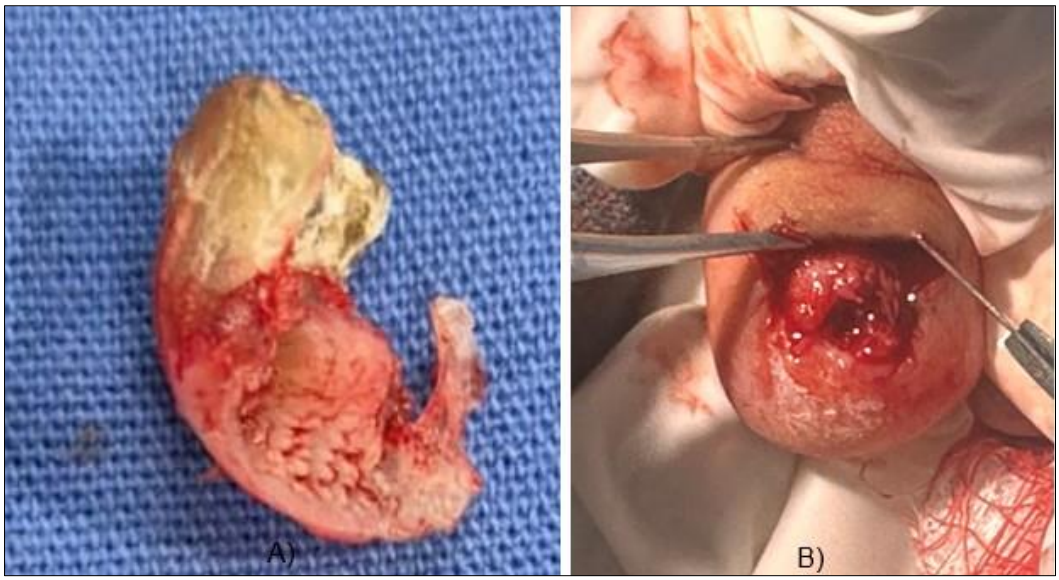

Figure 3 A and B: Intraoperative findings. A) Total nail avulsion revealing a filamentous tumor of the nail matrix with projections. B) Digitiform projections of the tumor are observed

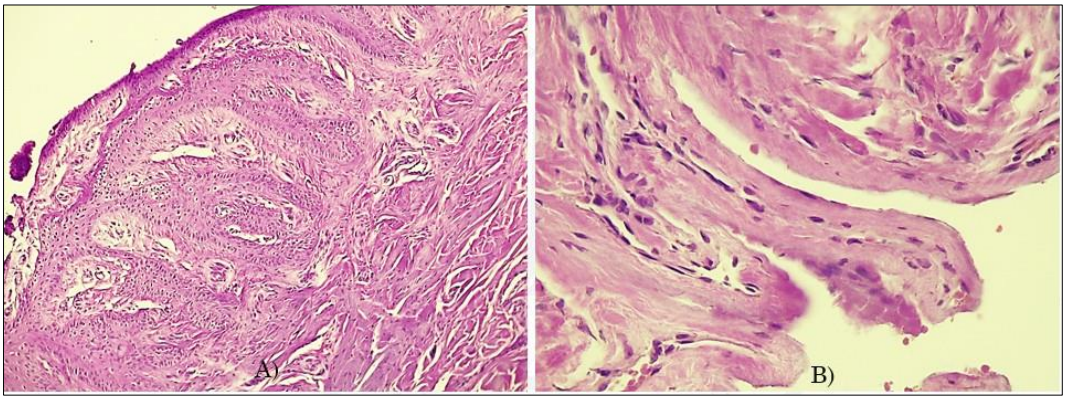

Figure 4 A and B: Histology with H&E showing: A) Epidermis with absence of the granular layer and elongated digitiform intraepithelial processes leaving dermal islands. B) On closer view of the intraepithelial processes, the digitiform appearance with spindle-shaped cells is better appreciated

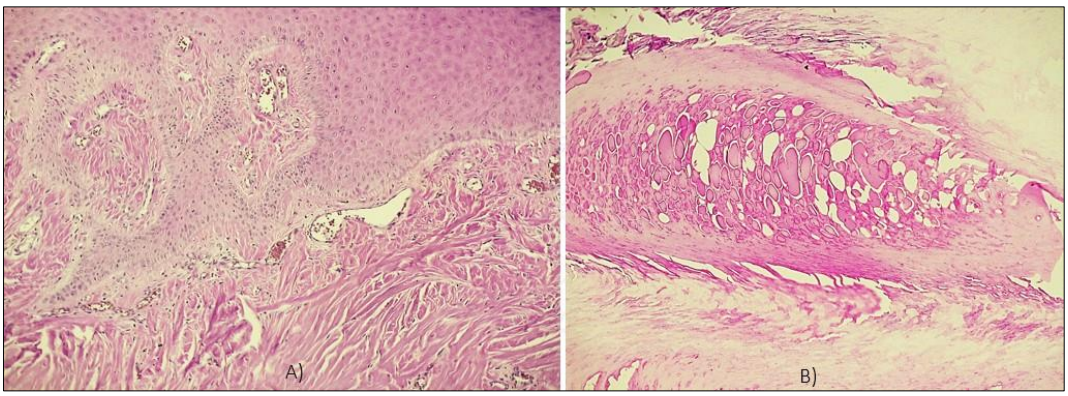

Figure 5 A and B: Histology with H&E showing: A) Collagenized, hypervascular stroma adjacent to the proliferated matrix epithelium. B) Transverse section of the nail plate with hyperkeratosis, empty cavities, and others filled with transudate

DISCUSSION

Onychomatricoma (OM) is a benign fibroepithelial neoplasm of the nail matrix whose rarity and clinical variability have made it a diagnostic challenge for dermatologists since its original description in 1992 [1]. Its diagnosis is based on the correlation between clinical morphology, dermoscopic findings, and histopathological study [1].

The average age at presentation is in the fifth decade of life, with a predominance in women (female-to-male ratio of 2:1) [3]. The case we report involves a male patient, which is less frequent according to the literature, and he was in the third decade of life, which also differs from what is commonly reported.

Fingernails are the most frequently reported location, as in the study by Perrin *et al*., where 19 cases were reported and 11 involved fingernails.2 Di Chiacchio *et al*., reported 30 cases of onychomatricoma, in which the most frequent location was also fingernails with 19 cases and only 11 cases involving toenails. Seven lesions occurred on the thumb, three cases on the second finger, seven on the third finger, and two on the fifth finger. The non-dominant hand was most commonly affected. Of the 11 tumors involving toenails, seven occurred on the great toe, two on the second toe, and two on the third toe [3]. The case we report affected the second toe of the left foot, which is an uncommon presentation.

The most frequently reported clinical features include thickening of the nail plate in 83% to 100% of cases depending on the series,3,6 splinter hemorrhages in 40% to 80% of cases, xanthonychia in 73% to 90% of cases, and increased transverse curvature of the nail in 50% to 60% of cases [3-6]. Our case exhibited these clinical characteristics, which led us to suspect onychomatricoma.

From a clinical standpoint, thickening of the nail plate, xanthonychia, woodworm-like cavities, transverse overcurvature, and longitudinal striations are characteristic changes [3, 4]. Other reported changes include edema of the proximal nail fold (40% to 50%) and melanonychia (23% to 30%) [3, 4]. These findings may be confused with onychomycosis, frequently leading to prolonged and ineffective antifungal treatments [5]. Our case was initially diagnosed as onychomycosis, which is one of the differential diagnoses of onychomatricoma.

The average reported duration of evolution is 12.7 years, likely due to lack of awareness of this entity and because the tumor is usually asymptomatic [3-5]. Our patient had a shorter duration of evolution and, although initially asymptomatic, later developed pain associated with footwear.

A key point in this case, which should alert clinicians, is refractoriness to conventional treatment, even when positive mycological reports exist, which may be incidental [5]. Rushing *et al*., after reviewing the literature, found that out of nearly 80 reported cases of onychomatricoma, 15 were initially misdiagnosed and treated as primary onychomycosis [5]. Our patient received this initial diagnosis and treatment.

In onychomatricoma, digitiform projections penetrate the nail plate and may produce woodworm-like cavities [2], visible on the coronal plane of the free edge of the nail plate, making the nail plate susceptible to fungal invasion, as occurred in our case. Other differential diagnoses include fibrokeratoma, ungual fibroma, subungual verruca vulgaris, and Bowen disease.

Although the exact etiology of onychomatricoma is unknown, trauma to the nail plate and onychomycosis may be predisposing factors [5]. Our case had the second antecedent.

Nail dermoscopy has proven to be a fundamental noninvasive diagnostic tool that helps guide clinical suspicion. The most common features include the presence of longitudinal white lines, proximal and distal splinter hemorrhages, and holes at the distal edge [3].

A classic tetrad is described: longitudinal thickening of the nail plate, xanthonychia, increased transverse curvature, and splinter hemorrhages. In our patient, the identification of cavities with a “honeycomb” pattern and digitiform projections was decisive [5, 6]. These findings are the macroscopic reflection of the tumor architecture: the cavities in the nail plate correspond to the spaces occupied by stromal projections emerging from the matrix [2].

Additional diagnostic methods include radiography, ultrasonography, and magnetic resonance imaging. Radiographic examination does not reveal any underlying bone involvement associated with onychomatricoma [5-7].

Ultrasound may show a hypoechoic, hypervascular tumor located in the nail matrix and projecting toward the nail bed and plate, with linear hyperechoic points within the hypoechoic áreas [7, 8].

Magnetic resonance imaging in onychomatricoma shows low signal intensity in the portion affecting the nail matrix and high signal intensity in the tumor projections [5-7]. Tumoral digitations within the nail plate appear Y-shaped with high-intensity signal, probably due to their higher water content compared with the surrounding nail. Axial images reveal the holes in the nail plate and the tumor digitations [7].

Confocal microscopy shows acanthotic and papillomatous epithelium without identified atypia and numerous spindle cells corresponding to fibroblasts in the surrounding dermal papillae [7].

The gold standard is histopathological examination. In 15 of 19 cases reviewed by Perrin *et al*., [2], the tumor consisted of a connective tissue core covered by epithelium lacking a granular layer and stratum corneum. Deep epithelial ridges appear as small optically empty cavities, which are serum-filled cavities lined by parakeratotic epithelium [2, 3]. Longitudinal sections show a broad pedunculated fibroepithelial tumor. The stroma is organized into a superficial layer composed of thin, wavy fibroblast nuclei arranged randomly, and a deeper layer with thickened collagen bundles oriented along a horizontal axis. The presence of fibroepithelial digitations lined by matrix epithelium and a collagenized hypervascular stroma confirms the entity [2-9]. Our case presented these histological changes.

Immunohistochemical study shows strong and diffuse CD34 expression in all cases, while EMA, S-100 protein, smooth muscle actin, desmin, and CD99 are negative [2]. In our case, immunohistochemistry was not performed.

Management by total nail avulsion not only allows diagnostic confirmation [2], but is also the treatment of choice, showing virtually null recurrence rates when resection is complete.

Although most cases of OM present idiopathically, its association with tuberous sclerosis is of particular interest. The presence of nail matrix tumors in patients with genodermatoses suggests a possible genetic predisposition to the development of hamartomas or fibroepithelial tumors.

Grover *et al.,* reported the case of a 15-year-old male patient with tuberous sclerosis presenting with facial angiofibromas, periungual fibromas, seizures, and onychomatricoma on the right great toenail. The authors proposed a possible relationship between genetic mutations related to tuberous sclerosis and the development of onychomatricoma [9].

Cañueto *et al*., performed genomic analysis of a 36-year-old man with onychomatricoma on the fourth toe of the right foot. Testing revealed 34 genomic alterations, with most genomic losses occurring on chromosome 11. Array-based comparative genomic hybridization showed deletion of 11p15.4, harboring STIM1, 11q14.2 harboring the Cathepsin C gene, 11q14, and 11q21.10.

Deletion of 11p15.4 harboring the STIM1 gene is especially significant, as decreased STIM1 levels have been associated with accelerated progression of tumors related to tuberous sclerosis due to restoration of AKT1 function and increased angiogénesis [9].

Losses involving chromosome 11 and decreased STIM1 levels may explain the association between onychomatricoma and tuberous sclerosis. According to Grover *et al*., p53 mutations identified in onychomatricoma cases suggest an interaction between p53 and tuberous sclerosis contributing to tumorigenesis. Mutations in TSC2 or p53 may impair normal autophagy, leading to tumorigenesis [9].

In our case, genomic study was not performed to corroborate this association.

CONCLUSION

This is the second reported case in the literature describing an association between onychomatricoma and tuberous sclerosis. In this case, OM presented as an isolated lesion and the patient only had facial angiofibromas, highlighting that nail surveillance should be comprehensive in these patients.

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