



Koenen Tumors 20 Nail Involvement: A Rare Case Report

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Abstract

Introduction: Tuberous Sclerosis Complex (TSC) is a multisystem disorder characterized by growth of benign tumors in various organs of the body, caused by mutations of TSC1 or TSC2 gene. Periungual fibromas are a common cutaneous manifestation and a frequent source of cosmetic and functional concern. The purpose of this paper is to report a rare case of Koenen tumours with 20 nail involvement.

Case Report: A housewife aged 55 years presented with complaint of difficulty in doing regular household work due to periungual growths on both hands. She had history of 2-3 episodes of seizures during childhood. Physical examination revealed firm, smooth, skin coloured periungual growths in all 20 nails suggesting 'Koenen tumors'. Multiple facial angiofibromas and ash leaf macules on the back, were seen. We diagnosed the case as TSC on the basis of above findings which fulfilled three major diagnostic criterias. Koenen tumors of bilateral thumbs were ablated by CO2 laser.

Discussion: TSC is a lifelong condition. Symptoms may cause disfigurement and disability, as in this case. So, early diagnosis and regular monitoring is very essential. Till date, a case of Koenen tumors involving 20 nails has not been reported from Nepal.

Keywords: Tubereous sclerosis complex; Koenen tumors; 20 Nail

Introduction

Tuberous Sclerosis (TS) is an autosomal dominant, neurocutaneous syndrome along with multisystem, characterized by growth of benign tumors in various organs such as heart, kidney, central nervous system along with face and nail resulting as Tubereous Sclerosis Complex (TSC) [1]. TSC is caused by mutation on either of two genes; encode for the proteins hamartin TSC1 and encoding tuberin TSC2 respectively. These proteins act as tumors growth suppressor agents that regulate cell proliferation and differentiation [2].

Tuberous sclerosis complex (TSC) or Bourneville's disease, first described by Desiree-Magloire Bourneville in 1880, and the Neurologist Vogt in 1908 established a diagnostic traid of epilepsy, idiocy and adenoma sebaceum. TSC occur in all races and ethnic groups and in both gender. The live birth prevalence is estimated to be between 10 and 16 cases per 100,000 [3]. TSC is diagnosed when either 2 major features (out of a total of 11) or one major feature with 2 minor feature (out of total 9) are present [4].

Peri-Ungual fibroma known as Koenen tumor is one of the major criteria and observed in 20 % of patients of TSC. They are usually asymptomatic, but sometimes cause nail deformity and pain. The purpose of this paper is to report a rare case of Koenen tumors with 20 nail involvement.

Case Presentation

A 55 years housewife present with chief complaint of difficulty in doing regular household work due to periungual growths on both hands specially while peeling of maize. These periungual growth were gradual onset since puberty and were asymptomatic, gradually with time it were present in all twenty nail and especially on bilateral thumb, the growth became more enlarge and painful which restricted her for doing household works. She had history of 2 episodes of seizures during childhood, but was not treated and was not taking any medication. Multiple, tiny, reddish non tender, asymptomatic bums were present in her face since childhood along with multiple hypopigmented, asymptomatic patch on her back. She had got good and happy married life with two sons and three daughters with normal and healthy grand childrens. Psychological assessment and Intelligence quotient (IQ) was equally efficient when compared with other ladies of similar age.

On Examination of patient revealed multiple dome shaped reddish to brown papular lesion less than 0.5 cm over nasolabial fold and cheeks suggestive of facial angiofibromas (adenoma sebaceum)

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Figure 1: Angiofibromas (adenoma sebaceum).



Figure 2: Multiple Hypomelanotic macules: Ash leaf macules.



Figure 3: Bilateral periungual fibroma on Fingers: Koenen tumors.



Figure 4: Bilateral periungual fibroma on Toes: Koenen tumors.



Figure 5: Warty Periungual Koenen tumors.



Figure 6: CO2 Laser Ablation of Periungual fibroma of Thumb.

in a typical butterfly pattern (Figure 1). There were multiple hypopigmented 'ash leaf macules' on the back (Figure 2).

Multiple firm, smooth, skin colored to mild reddish periungual non tender verrucous growths were seen on the surface in all 20 nails suggesting of 'Koenen tumours' (Figure 3,4 and 5). All the nails were dystrophic. Her family history was negative for TSC. Examination of her eyes (fundoscopy) and cardiovascular and CNS, were in normal limits. Abdominal ultrasound imaging, X- ray of hand and feet were also normal. CT scan and MRI were not significant. Blood investigation for complete blood count, liver function test, Renal function test, within normal limits. The diagnosis of TSC was made on with clinical ground fulfilled with three major criteria, without the requirement of histological confirmation. Koenen tumours of bilateral thumbs were ablated by CO2 laser under aseptic condition (Figure 6). Our case was uncommon as the tumors affecting all fingers and toe nails along with healthy neurological feature at age of fifty.

Discussion

Tuberous sclerosis complex is autosomal disorder characterized by multiple hamatomas of skin, kidney, retina, heart and central nervous system [1]. Only 25% of patients present with classical

triad of disease. The incidence of disease is 1/10,000 and up to 50% of cases result of spontaneous mutation [1]. The diagnosis of TSC can be made by genetic analysis or on the basis of clinical diagnostic criteria as given below (Table 1). Half of the TSC families are linked to chromosome 9q34 with tumor suppressor activity protein tuberin (TSC1) and the other half to chromosome 16p13 with tumor suppressor activity protein hamartin (TSC2) [1].

Koenen tumors are periungual or subungual fibromas appearing as reddish to flesh-colored, smooth, soft to firm papules and nodules emerging from nail folds. They are usually 5–10 mm in length, but may be very large. This can be the only clinically evident abnormality [5-6]. In 50% cases of TSC, it appears at an age between 12 and 14 years and increases progressively in size and number with age. The tumors sometimes become hyperkeratotic, resembling fibrokeratoma [7]. Similar case of multiple periungual verrucous nail was reported from India [6]. Histologically, Koenen tumors are characterized by fibrosis with stellate shaped strands of fibroblasts and dense

Table 1: Diagnostic criteria for tuberous sclerosis complex (TSC) [10].

Major features	Minor features
1. Facial angiofibromas	1. Multiple, randomly distributed pits in dental enamel
2. Hypomelanotic macules (three or more)	2. Hamartomatous rectal polyps
3. Ungula or periungual fibroma	3. Bone cysts
4. Shagreen patch	4. Cerebral white matter radial migration lines
5. Retinal nodular hamartomas	5. Gingival fibromas
6. Cortical tuber	6. Nonrenal hamartoma
7. Subependymal nodule	7. Retinal achromic patch
8. Subependymal giant cell astrocytoma	8. Confetti' skin lesions
9. Cardiac rhabdomyoma	9. Multiple renal cysts
10. Lymphangiomyomatosis	
11. Renal angiomyolipomas	

Definite Diagnosis: Two major feature or one minor feature with > 2 major feature.

Possible Diagnosis: Either one major feature or > 2 minor feature.

collagen with numerous ectatic blood vessels. Multiple Koenen tumors, affecting all the fingers and toe nails, with a bigger size and morphologically resembling periungual wart, have been reported earlier [7,8]. Periungual fibromas are frequent source of cosmetic and functional concern, and were treated with a continuous wave CO₂ laser in sequential focused and defocused modes at 10-15 Watt. As compared to conventional surgery in terms of cosmetic satisfaction, CO₂ laser presents a viable alternative to surgery through significantly reduced treatment time and amount of bleeding and wounds healed well with good cosmetic results. Berlin in 2002 as got good cosmetic result case with CO₂ laser [9]. It is an underestimated figure as many cases of TSC remain undiagnosed due to various clinical presentations. Intervention programs, including special schooling and occupational therapy may benefit individuals with special needs and developmental issues [5] laser treatment surgery; dermabrasion may be useful for treatment of skin lesions. Prognosis of the disease depends on the severity or multiplicity of organ involvement. About a quarter of severely affected infants are thought to die before age 10 years, and 75% die before age 25 years old [5]. However, the prognosis for the individual diagnosed late in life with few cutaneous signs as present in our case, where she present at age of 56 with difficulty for doing her normal household work due to periungual fibroma, without any clinical presentation of neurological and systemic feature.

Conclusion

TSC is a lifelong condition; therefore individuals should be regularly monitored by an experienced clinician. It is not uncommon for patients with TSC to have symptoms or signs that do not lead to immediate diagnosis. In some cases, diagnosis is delayed for prolonged periods of time. Early diagnosis is important to improve quality of life, and to avoid disfigurement and disability.

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