Case Report

Phacomatosis Pigmentokeratotica Associated with Unilateral Toe Walking Due to Short Achilles Tendon

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Abstract

Phacomatosis pigmentokeratotica (PPK) is characterized by the co-occurrence of speckled lentiginous nevus (nevus spilus) and an organoid nevus with or without extracutaneous involvement. The extracutaneous manifestations may vary widely with musculoskeletal, neurologic, ocular, and vascular findings. The PPK is also associated with an increased risk of cutaneous or extracutaneous tumors. Therefore, the patients with PPK should be followed up regularly for possible malignant transformation. Here, we report a 5-year-old boy with PPK associated with toe walking due to short Achilles tendon, which was not previously reported, to our knowledge.

Keywords: Genetic diseases/mechanisms, genodermatoses, neoplasms-benign

Introduction

Phacomatosis pigmentokeratotica (PPK) is characterized by the co-occurrence of speckled lentiginous nevus (nevus spilus, SLN) and an organoid nevus (ON) with or without extracutaneous involvement.[1] PPK is caused by a postzygotic HRAS mutation and thus is a mosaic RASopathy.[2-4] Here, we report a 5-year-old boy with PPK associated with toe walking due to short Achilles tendon, which was not previously reported, to our knowledge.

Case Report

A 5-year-old boy was referred to our clinic for congenital skin lesions. He was born to nonconsanguineous parents and otherwise healthy except for the unilateral toe walking due to short Achilles tendon on the left lower extremity [Figure 1a].

Dermatological examination showed numerous dark brown-to-black macules and papules on a tan background involving the left upper back and left arm and also on the right gluteal region, which was compatible with a diagnosis of SLN [Figure 1b-d]. A Blaschko linear, verrucous plaque affecting the presternal area and extending through the anterior neck and right ear, consistent with an epidermal nevus, was also noted [Figure 2a]. There was also a yellowish-brown, verrucous plaque over the left ear and scalp clinically diagnosed as nevus sebaceous [Figure 2b].

Based on the coexistence of SLN, epidermal nevus, and nevus sebaceous, a diagnosis of PPK was made. Long-term follow-up of the patient was planned for possible malignant transformation.

Discussion

PPK is an epidermal nevus syndrome characterized by the coexistence of SLN and nevus sebaceous, in addition to extracutaneous manifestations including neurologic, ocular, and musculoskeletal involvement.[1] Although it was originally thought to result from twin-spotting phenomenon, the more recent studies have shown that a postzygotic HRAS mutation in a multipotent progenitor cell is the underlying cause.[3-5] Unfortunately, genetic testing in our patient could not be performed due to unavailability.

Submission: 20-10-2020 **Acceptance:** 05-12-2020
Web Publication: 31-12-2020

Access this article online

Quick Response Code:

Website: www.tjdonline.org

DOI: 10.4103/tjd.tjd_106_20

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How to cite this article: Salman A, Yucelten AD, Cakici OA, Unver O. Phacomatosis pigmentokeratotica associated with unilateral toe walking due to short achilles tendon. Turk J Dermatol 2020;14:106-8.

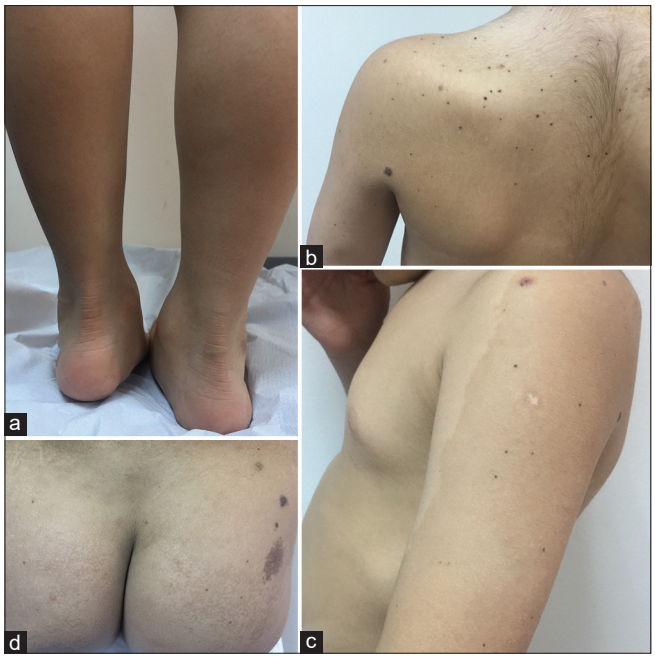

Figure 1: Toe walking on the left side due to short Achilles tendon (a). Nevus spilus over the left scapulae and the right gluteal region (b-d)

The consistent clinical features of PPK are SLN and ON with sebaceous differentiation. The distribution of SLN and ON may be contralateral, ipsilateral, or bilateral.[6] In the present case, both SLN and ON had a bilateral distribution which was previously reported to occur rarely.[7] The ON in PPK tends to involve the head-and-neck region, as in our patient. Although PPK with isolated cutaneous involvement exists, the extracutaneous manifestations are common and may vary widely with musculoskeletal, neurologic, ocular, and vascular findings. Kyphoscoliosis, hemiatrophy, or hemihypertrophy of the musculoskeletal system in PPK have been reported previously; however, short Achilles tendon, as seen in our case, has not been reported before, to our knowledge.[1,5,8]

The PPK is also associated with an increased risk of cutaneous or extracutaneous tumors. Development of malignant melanoma on SLN and basal cell carcinoma on nevus sebaceous has been reported. Visceral tumors such as leiomyoma of the bladder and vaginal rhabdomyosarcoma have been reported as well.[5,7,9,10] Our patient had no evidence of cutaneous or internal malignancies at the time of diagnosis. However, the patients with PPK should be followed up regularly for possible malignant transformation.

The differential diagnosis should include Schimmelpenning syndrome and SLN syndrome. The presence of mental retardation, seizures, and coloboma in addition to nevus sebaceous in Schimmelpenning syndrome; the association of SLN with sensory neuropathy and hyperhidrosis in SLN syndrome may help to differentiate PPK from these conditions.[1]

In conclusion, this report extends the clinical phenotype of PPK with adding short Achilles tendon to extracutaneous manifestations. We believe that early recognition of PPK is critical for proper monitoring of the patients for malignant transformation and counseling of the parents.

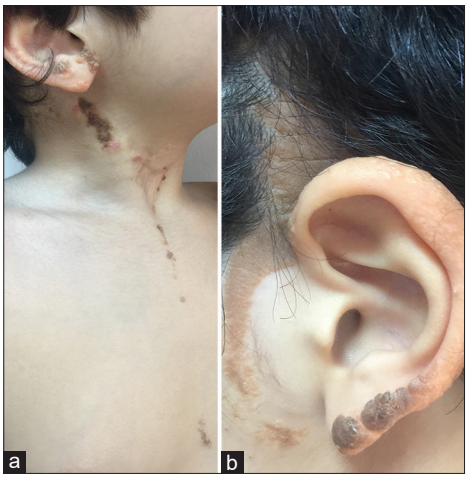

Figure 2: A Blaschko linear epidermal nevus over the anterior neck-and-right ear (a). Nevus sebaceous involving the left ear and the scalp (b)

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

References

1. Boente Mdel C, Asial RA, Happle R. Phacomatosis pigmentokeratotica: A follow-up report documenting additional cutaneous and extracutaneous anomalies. Pediatr Dermatol 2008;25:76-80.
2. Wollenberg A, Butnaru C, Oppel T. Phacomatosis pigmentokeratotica (Happle) in a 23-year-old man. Acta Derm Venereol 2002;82:55-7.
3. Groesser L, Herschberger E, Sagrera A, Shwayder T, Flux K, Ehmann L, *et al*. Phacomatosis pigmentokeratotica is caused by a postzygotic HRAS mutation in a multipotent progenitor cell. J Invest Dermatol 2013;133:1998-2003.
4. Happle R. Phacomatosis pigmentokeratotica is a pseudodidymosis. J Invest Dermatol 2013;133:1923-5.
5. Om A, Cathey SS, Gathings RM, Hudspeth M, Lee JA, Marzolf S, *et al.* Phacomatosis pigmentokeratotica: A mosaic RASopathy with malignant potential. Pediatr Dermatol 2017;34:352-5.
6. Happle R, Hoffmann R, Restano L, Caputo R, Tadini G. Phacomatosis pigmentokeratotica: A melanocytic-epidermal twin nevus syndrome. Am J Med Genet 1996;65:363-5.
7. Gruson LM, Orlow SJ, Schaffer JV. Phacomatosis pigmentokeratotica associated with hemihypertrophy and a rhabdomyosarcoma of the abdominal wall. J Am Acad Dermatol 2006;55:16-20.
8. Chantorn R, Shwayder T. Phacomatosis pigmentokeratotica: A further case without extracutaneous anomalies and review of the condition.

Pediatr Dermatol 2011;28:715-9.
9. Bouthors J, Vantyghem MC, Manouvrier-Hanu S, Soudan B, Proust E, Happle R, *et al.* Phacomatosis pigmentokeratotica associated with hypophosphataemic rickets, pheochromocytoma and multiple basal cell carcinomas. Br J Dermatol 2006;155:225-6.
10. Prieto-Barrios M, Llamas-Martin R, Velasco-Tamariz V, Calleja-Algarra A, Ruano Y, Ortiz-Romero PL, *et al.* Phacomatosis pigmentokeratotica: A case of HRAS mosaicism causing rhabdomyosarcoma. Br J Dermatol 2018;179:1163-7.