

Case Report

Congenital true leukonychia totalis

B. M. Vyshak, Snehal B. Lunge*

Department of Dermatology, Venereology and Leprosy, KLE Academy of Higher Education and Research, Belagavi, Karnataka, India

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***Correspondence:**

Dr. Snehal B. Lunge,

E-mail: drsnehallunge@gmail.com

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ABSTRACT

Congenital true leukonychia totalis is very rare hereditary condition characterized by milky white discoloration of all nails. It has mainly autosomal dominant mode of inheritance and autosomal recessive in few cases. It may be inherited or associated with systemic disease or idiopathic. It has been linked to the mutations in *PLCD1* gene on chromosome 3p21.3-p22. Leukonychia totalis is associated with multiple systemic diseases such as congenital hyperparathyroidism, Hodgkin's lymphoma, Leopard syndrome, Epiphyseal dysplasia syndrome, Bart Pumphrey syndrome etc. we report a case of sporadic congenital leukonychia totalis in a 24 years female without any systemic abnormalities.

Keywords: Congenital leukonychia, Leukonychia totalis, White nails, Partial leukonychia

INTRODUCTION

Leukonychia is the white discoloration of nail that loses its normal pink color, with disappearance of the lunula.¹ It is the commonest chromatic abnormality of the nail apparatus.¹ Leukonychia can be classified in several ways. Based on the site of origin of the white discoloration it is classified as True leukonychia (due pathology in the nail matrix), apparent leukonychia (due to pathological changes under the nail bed) and pseudo-leukonychia (due to external factors such as trauma).² Based on the extent of the discoloration it is classified as: leukonychia totalis (complete whitening of the nails), leukonychia partialis (incomplete whitening of the nails), leukonychia punctata (white spots), and leukonychia striata (white bands).²

True leukonychia can either be acquired or hereditary. Acquired true leukonychia occurs due to alteration in the nail matrix by medical conditions or external exposure. Hereditary true leukonychia may be a benign isolated condition or an associated occurrence with a range of systemic diseases.² Hereditary leukonychia is associated

with the mutations in *PLCD1* gene on chromosome 3p21.3-p22 encoding an important enzyme in phosphoinositide metabolism responsible for control of color and growth of nail.³

Here we report a case a 24 years old lady with asymptomatic complete white discoloration of all the nails (congenital true leukonychia totalis) with family history and with absence of associated conditions.

CASE REPORT

A 24 years old female presented with whitish discoloration of the entire nail on all fingers and toenails since birth. The patient was born from non-consanguineous parents and has two children. His family history was significant for similar nail findings in her grandmother with no similar history in her parents or in her children. She denied prolonged illness, pain, decreased sensation to pressure on the nails or any prior history of serious mechanical trauma to the nails. The patient had no other medical conditions. The growth of her nails was otherwise normal.

On cutaneous examination all finger nails and toe nails were opaque, milky, porcelain white in color with smooth surface involving the entire nail plate with normal cuticles (Figure 1 and 2). Lunula was invisible, and there was no subungual debris or onycholysis. Pressure on the nail plate caused no fading of the discoloration. The nails were normal with respect to strength, shape, and texture. No other skin or hair abnormalities were detected. General and systemic examination was unremarkable. Extensive laboratory investigations including complete blood count, blood sugar, urinalysis, liver function test, serum protein and albumin, serum calcium, renal function test, and thyroid function test were performed and were found to be within normal limits. Onychoscopy over the nail plate of all the nails showed diffuse milky white discoloration with normal appearing proximal and lateral nail folds. Nail clippings for potassium hydroxide (KOH) mount and culture on Sabouraud's dextrose agar was negative. Nail biopsy was not performed as the patient did not consent to the procedure. On clinical and laboratory correlation, a diagnosis of idiopathic congenital true leukonychia totalis was made.



Figure 1: Milky white, porcelain colored smooth surfaced finger nails.



Figure 2: Milky white, porcelain colored toe nails.

DISCUSSION

Leukonychia refers to whitish discoloration of the nails. It is derived from the Greek words leuko (meaning white) and onyx (meaning nail).¹ It was first described by Mees in the year 1919 in association with chronic arsenic poisoning.¹ The exact mechanism of whitening is not known. However, it has been proposed that it is attributable to reflection of light from large keratohyalin granules present in the parakeratotic cells of the ventral portion of nail plate thus preventing the visualisation of normal vascular tissue underlying the nail plate.^{2,3}

Unna classified leukonychia into five clinical types based on the distribution of whiteness leukonychia totalis, leukonychia partialis, leukonychia striata, leukonychia transversalis, and leukonychia punctata. However, Butterworth proposed that leukonychia partialis as a phase of leukonychia totalis, and not a distinct entity.³ Leukonychia is also classified into true leukonychia, where the pathology is in the nail plate, apparent leukonychia, where the pathology is in the subungual tissue and pseudo-leukonychia due to external factors such as trauma.³

Leukonychia totalis is a relatively rare condition. It is usually classified into inherited and acquired. Acquired leukonychia totalis is significantly more common and is associated with several medical conditions.² The inherited form of leukonychia, i.e., hereditary leukonychia totalis, however, is extremely rare. In a review article by Kruse et al, the first isolated case of hereditary leukonychia totalis was reported in a 30 years old man and his father in 1913.² And as per the previous literature data only about lesser than fifty cases of congenital leukonychia totalis has been reported and these cases were reported as far back as 1913. Family history was elicited in nearly 79% of the cases.² Although, hereditary leukonychia is predominantly inherited in an autosomal dominant pattern, there are a few case reports of autosomal recessive inheritance as reported by Frydman et al.³ Mutations in *PLCD1* gene on chromosome 3p21.3-p22 was identified as cause of hereditary leukonychia totalis. This *PLCD1* gene is localized to nail matrix and encodes phosphoinositide specific phospholipase C delta 1 subunit which is important enzyme in phosphoinositide metabolism required for molecular control of color and growth of nail.^{1,2}

Leukonychia totalis is associated with multiple systemic diseases such as Bart Pumphrey syndrome, Buschke-Gorlin syndrome, LEOPARD syndrome diabetes, liver cirrhosis, renal failure, cardiac failure, gall stones, renal calculi, duodenal ulcers, pili torti, trichilemmal cyst, sebaceous cyst, congenital hyperparathyroidism, Hodgkin's lymphoma, epiphyseal dysplasia syndrome, mental retardation, etc.^{1,3,4} However, there are very few reports in the literature on sporadic congenital leukonychia totalis without any systemic abnormalities.

The prognosis of leukonychia remains undetermined in cases of congenital leukonychia. Although there is no specific treatment for true leukonychia, proper counselling and advice regarding camouflaging of the nails would be necessary.² Nevertheless, total leukonychia due to systemic disorders appears to have reasonably good prognosis as it seems to be transient and reversible.

CONCLUSION

Isolated congenital true leukonychia totalis is a rare entity and we report this case not only because of its rarity, but also to emphasize that this condition may display itself as an important visible clinical clue to other associated systemic diseases or inherited malformations. Hence it is necessary to conduct a detailed investigation to determine the underlying disorders associated with this condition.

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