INTRODUCTION

Congenital alopecia universalis is one of the rarest anomaly which involves skin and appendages. It may develop either suddenly or gradually can be superimposed on alopecia areata.\(^1\) It usually affects middle-aged persons although it can occur at any age. Few research studies mentioned that this can be associated with other congenital defects.\(^2\) The inheritance pattern can be autosomal recessive, X-linked recessive, or autosomal dominant. However, the most common is autosomal recessive form and it is the most severe phenotype. Twenty-nail dystrophy refers to the condition in which all the twenty nails are affected in the form of excessive ridging and nail plate roughness leading to unsightly lustureless nails. We report a rare case of two siblings with alopecia universalis congenita with twenty-nail dystrophy. To the best of our knowledge, this case is the first case to be reported with such association in both siblings. This case reports highlights the fact that alopecia areata is an autoimmune disease with a genetic predisposition as in our case both siblings had alopecia universalis and nail dystrophy. There was no evidence of any other ectodermal dysplasia and had normal teeth and seat glands. The skin biopsy ruled out congenital atrichia and was suggestive of alopecia areata.

CASE REPORT

A 10-year-old boy and 6-year-old girl of same parents presented with a history of total absence of scalp and body hair and dystrophic nails since birth. Both were born of nonconsanguineous marriage, at full term, and without fetal anomalies. They did not report any abnormal or delayed dentition, abnormal sweating or anhidrosis, musculoskeletal, ocular, or neurologic symptoms. Their mother reported with the history of dystrophic nails since birth. Physical examination found Alopecia universalis in both children along with dystrophy in all twenty nails, and this has been shown in Figures 1 and 2. Skin biopsy was done to rule out congenital atrichia; however, both transverse and longitudinal sections on H and E staining showed rudimentary hair bulbs with a peribulbar

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lymphocytic infiltrate as shown in Figures 3 and 4. The children had a normal thyroid profile and normal Vitamin D3 levels.

In our case study, the occurrence of hair disorder in two siblings with unaffected parents is suggestive of autosomal recessive inheritance. The patient's parents were counselled for genetic testing; however, they deferred it. At present, both children have been started on oral mini-pulse steroids and topical immunomodulators and topical minoxidil solution.

**DISCUSSION**

Alopecia which presents since birth may include several types of hereditary hair loss disorders in human beings. Congenital alopecia may present soon after birth in the form of either generalized or localized loss of hair. This rare condition of alopecia universalis is either seen with other associated defects or in isolation. Other known syndromes associated with hypotrichia and alopecia are congenital atrichia with nevus flammeus, growth retardation, alopecia, pseudoanodontia, and optic atrophy syndrome, atrichia with papular lesions, and dominant hidrotic epidermo dysplasia. The majority of congenital alopecia cases have an autosomal recessive inheritance but few cases of autosomal dominant or X-linked recessive inheritance have also been reported. Patients with an autosomal recessive disorder who have congenital alopecia manifests as complete loss of hair involving entire scalp and body including eyebrows, eyelashes, axillary, and pubic regions. If it is dominant type, it presents with less severe form of alopecia and often manifests in later years. On performing biopsy, tissue section reveals a normal epidermis and dermis absence of hair follicles. Ectodermal dysplasia is another condition which may present with alopecia and other common anomalies of, teeth, nails, and sweat glands are seen.

Trachyonychia clinically presents as if the nails are sandpapered in longitudinal direction due to which nail plate get a rough and extremely ridged texture. Twenty-nail dystrophy is used only when all the twenty nails are affected. One research study done on children of alopecia observed that about 46% of the children had nail abnormalities and about 12% had twenty-nail dystrophy. In fact, it is seen that the nail dystrophy may further speed up the progression of alopecia and is a marker of poor prognosis. Histopathologically, spongiosis is seen in the...
proximal nail fold and nail matrix with a mild lymphocytic infiltrate. When spongiotic dermatitis is present in the nail matrix and nail bed and column-like parakeratosis in the nail plate, it is suggestive that trachyonychia was due to alopecia areata or atopic dermatitis. However, nail biopsies may be useful to confirm the diagnosis and exclude other conditions. However, the challenging part is treatment of trachyonychia associated with alopecia universalis. Tazorotene, a vitamin A analog given for local application to the nail plate, nail folds, and periungual areas for 3 months has shown results in clinical and functional improvement along with few side effects such as peeling and redness of proximal nail fold skin. Intralungal triamcinolone has been used with some success in idiopathic trachyonychia; however, it is an extremely painful and dissatisfying procedure, especially in the pediatric age group.

Treatment of alopecia universalis is based on immunomodulation. Topical clobetasol and diphenylcyclopropenone, which is a topical sensitizer may show some efficacy. Systemically, oral, intramuscular, or intravenous pulse glucocorticoids, methotrexate and biologicals such as efalizumab and the recent Jak kinase inhibitors have shown promising results in the management of alopecia areata.

Although alopecia areata is the most prevalent autoimmune disease, affecting more individuals than most other autoimmune diseases combined, the cellular and molecular mechanisms underlying this complex autoimmune disease are still poorly understood, and rational treatments are still lacking. It is currently accepted that alopecia areata is an autoimmune disease that occurs in genetically susceptible individuals and that environmental factors play a role in the development and progression of the disease. The exact molecular pathways are still elusive, and our case report certainly highlights the fact, that genetic factors play a crucial role in causation.

**CONCLUSIONS**

All cases of alopecia universalis should be thoroughly investigated for associations with syndromes and anomalies. Alopecia is an unusual association with trachyonychia. Genetic evaluation of the affected person is essential for better understanding of the disease. This seems to a first reported case of alopecia universalis with twenty nail dystrophy strongly suggesting an autosomal recessive mode of inheritance.

**Limitations**

The siblings could not be subjected to genotyping and further tests are needed to ascertain the genetic basis of alopecia areata in siblings.

**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.

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**Conflicts of interest**

There are no conflicts of interest.

**REFERENCES**

1. Andrews GC. Diseases of Skin. Philadelphia: W. B. Saunders Company; 1932.
2. Gillespie JB. Congenital and Familial Alopecia Totalis. Am J Dis Child. 1937;53(1_Part_I):132-36.
3. Nothen MM, Ciehon S, Vogt IR, Hemmer S, Kruse R, Knapp M, et al. A gene for universal congenital alopecia maps to chromosome 8p21-22. Am J Hum Genet 1998;62:386-90.
4. Scheinfeld NS. Trachyonychia: A case report and review of manifestations, associations, and treatments. Cutis 2003;71:299-302.
5. Grover C, Khandpur S, Reddy BS, Chaturvedi KU. Longitudinal nail biopsy: Utility in 20-nail dystrophy. Dermatol Surg 2003;29:1125-9.
6. Chien P Jr., Kovich OI. Alopecia universalis with twenty-nail dystrophy (trachyonychia). Dermatol Online J 2008;14:24.
7. Bennassar A, Ferrando J, Gimel R. Congenital atrichia and hypotrichosis. World J Pediatr 2011;7:111-7.
8. Tosti A, Morelli R, Bardazzi F, Peluso AM. Prevalence of nail abnormalities in children with alopecia areata. Pediatr Dermatol 1994;11:112-5.
9. Braun-Falco O, Dorn M, Neubert U, Plewig G. Trachyonychia: 20-nail dystrophy. Hautarzt 1981;32:17-22.
10. Soda R, Diluvio L, Bianchi L, Chimenti S. Treatment of trachyonychia with tazarotene. Clin Exp Dermatol 2005;30:301-2.
11. Khoo BP, Giam YC. A pilot study on the role of intralungal triamcinolone acetoniode in the treatment of pitted nails in children. Singapore Med J 2000;41:66-8.
12. Sotiriadis D, Patsatsi A, Lazaridou E, Kastanis A, Vakirlis E, Chrysomallis F. Topical immunotherapy with diphenylcyclopropenone in the treatment of chronic extensive alopecia areata. Clin Exp Dermatol 2007;32:48-51.
13. Bin Saif GA. Oral mega pulse methylprednisolone in alopecia universalis. Saudi Med J 2006;27:717-20.
14. Kurusawa M, Nakagawa S, Mizuishi M, Sasaki Y, Kawamura M, Saito M, et al. A comparison of the efficacy, relapse rate and side effects among three modalities of systemic corticosteroid therapy for alopecia areata. Dermatology 2006;212:361-5.
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15. Assouly P, Reygagne P, Jouanique C, Matard B, Marechal E, Reynert P, et al. Intravenous pulse methylprednisolone therapy for severe alopecia areata: An open study of 66 patients. Ann Dermatol Venereol 2003;130:326-30.

16. Joly P. The use of methotrexate alone or in combination with low doses of oral corticosteroids in the treatment of alopecia totalis or universalis. J Am Acad Dermatol 2006;55:632-6.

17. Kaelin U, Hassan AS, Braathen LR, Yawalkar N. Treatment of alopecia areata partim universalis with efalizumab. J Am Acad Dermatol 2006;55:529-32.

18. Jabbari A, Petukhova I, Cabral RM, Clynes R, Christiano AM. Genetic basis of alopecia areata: A roadmap for translational research. Dermatol Clin 2013;31:109-17.