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Monilethrix

Improvement with Acitretin

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Abstract

Monilethrix is a rare hereditary disorder that affects the hair and is characterized by shaft anomaly. There is no known treatment that successfully cures the condition. In this report we present a case of monilethrix in a 7-year-old girl treated with oral acitretin. A very good clinical and cosmetic result was obtained while treatment was continued. However, clinical symptoms recurred after discontinuation of acitretin therapy.

Monilethrix is a rare genetic disease that affects hair, and is specifically characterized by hair shaft anomaly. Fusiform nodes and narrow segments are seen in the hair shafts. Although several autosomal recessive cases have been reported, autosomal dominant inheritance is more common.[1-4] It has been reported that the disease develops because of mutations in genes (chromosome 12q13) that code hHb1 and hHb6, which are basic hair keratins in humans.[2,5-8] Lanugo hair in the neonatal period is normal. Clinical signs appear when terminal hair characteristics begin to form. Alopecia may be seen in the neonatal period in severe cases.[2] The scalp in particular is affected, but the eyebrows, eyelashes, axillary hair, pubic hair, and even body hair follicles may be involved. There is no known successful treatment of the disease. However, some treated cases have been reported.[1] Here we present a case of monilethrix that responded to acitretin treatment.

1. Case

A 7-year-old girl presented complaining of fragile hair, apparent sparseness, and lack of hair growth. Problems with her hair had first been noticed 3 months after birth. Specifically, the hair that grew in place of the lost hair did not grow long, it broke when it reached a certain length and her hair was thinning out in certain areas. Her general history was unremarkable, except for premature birth and iron deficiency anemia. Her family history was also unremarkable. Physical examination revealed that all systems, including growth and development, were normal. Dermatologic examination revealed sparse, stiff, dry, short, and irregular hair at bilateral temporo-parietal areas (figure 1). Hair and skin appendages at other areas were normal. The only dermatologic sign in the fingernails was slight longitudinal lines. Oral mucosa examination was normal.

Laboratory investigations showed that the whole blood count, peripheral blood smear, all biochemical values, liver function, serological parameters, and thyroid function were normal. The erythrocyte sedimentation rate was 14 mm/h.

Biopsy from hairy skin revealed regularly structured hair in the scalp. Microscopic examination of spontaneously falling hair and trichography of pulled out hair showed that the hair shafts were completely free of medulla and had elliptical nodes at about 1mm

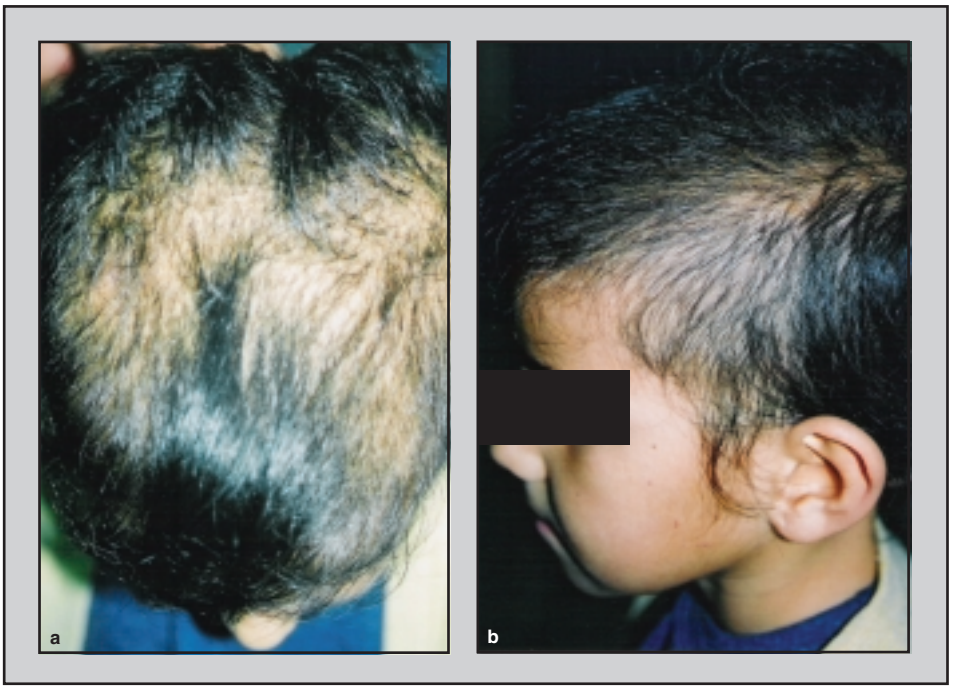

Fig. 1. Short hairs on the temporo-parietal area.

Fig. 2. Microscopic examination of monilethrix hair shafts showing that the shafts were completely free of medulla and had elliptical nodes at approximately 1mm intervals (original magnification × 40).

intervals (figure 2). The patient was diagnosed with monilethrix on the basis of these findings.

Treatment was started in the form of 0.5 mg/kg acitretin. In order to monitor early epiphysis shutdown before treatment, hand-wrist x-rays were obtained and growth and development data documented. In the 12th week of treatment, breaking of the hair had stopped, hair growth had started and recovery was observed in alopecic areas. Clinically, very good cosmetic results were obtained during treatment (figure 3), which was continued for 12 months. Symptoms resumed 4 months after discontinuation of treatment.

2. Discussion

Monilethrix is a genetic hair shaft disease that causes fragility in growing hair and alopecia.[1,3] It is characterized by localized or diffuse alopecia.[3] Lanugo hair is normal in the neonatal period. The condition manifests clinically with the formation of terminal hair. Follicular hyperkeratosis can be seen in any part of the body.[1,2,4] It most commonly affects occipital areas and the back of the neck.[4] Our patient's complaints began after birth, when terminal hair characteristics developed, and were localized to temporo-parietal areas.

Monilethrix is most frequently associated with follicular hyperkeratosis and koilonychia and rarely with mental retardation, syndactyly, cataract, tooth, and nail anomalies.[1,2,4,9] Our patient

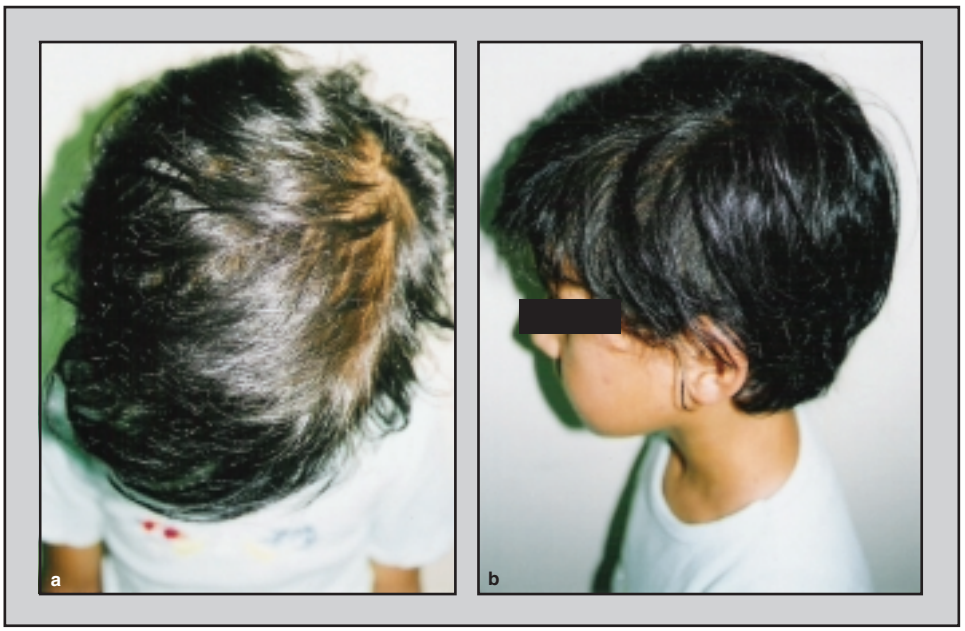

Fig. 3. Good clinical and cosmetic results on the alopecic area after acitretin treatment.

did not have follicular hyperkeratosis and koilonychia. Psychiatric and eye examinations in our patient were normal.

Light microscopy and trichologic examinations are diagnostic in monilethrix. Trichologic examination shows markedly higher rates of anagen hair. The shafts examined reveal 0.7–1mm intervals between nodes.[6] Hair can be visualized better under a polarized microscope, in particular, and the disease can be distinguished from pseudo-monilethrix using this approach.[4] It should be noted that the diagnosis in the present study was made on the basis of light microscopy findings. However, genetic testing for the classic mutations of the keratin genes would be desirable to make the diagnosis of monilethrix certain.[8] We diagnosed our patient using trichologic examination and light microscopy. Pseudo-monilethrix, other hair shaft anomalies, congenital alopecia, and ectodermal dysplasia should be considered in the differential diagnosis.[1,2]

The prognosis of monilethrix is variable.[1] Although it has been reported that the disease remits in adulthood in some cases, in most cases it is persistent.[10] It has been suggested that symptoms improve in summer, with advancing age, and with pregnancy in some cases.[4] Presentation of a patient whose complaints disappeared after the first menstrual period suggests that hormonal effects may play a role in recovery.[1,11] A case of localized monilethrix has also been reported in which the patient recovered after treatment of iron deficiency anemia.[3]

There is no known treatment that successfully cures monilethrix.[1] Although there have been reports of cases treated with systemic corticosteroids, oral retinoids, topical minoxidil, vitamins, and peeling ointments (desquamative oil), the best treatment is protecting the hair against traumatic procedures such as excessive combing, brushing, and friction. In addition, wigs may be used for cosmetic purposes.[1,4,9,10,12,13]

Retinoids have anti-keratinizing effects and have been shown to modulate keratin expression in *in vitro* experiments.[14] It has also been proposed that acitretin 0.5 mg/kg/day should be commenced in children with genetically transmitted keratinization disorders such as Netherton syndrome, palmoplantar keratoderma, and Sjögren-Larsson syndrome.[14-17] Some researchers have stated that they obtained 70–80% remission with etretinate 1–2 mg/kg/day in various keratinization disorders.[17,18]

The most frequent adverse effects of retinoids are mucocutaneous effects such as cheilitis, skin dryness, increase in skin fragility, pruritus, dryness of the nose, and epistaxis. Despite these adverse effects, children receiving long-term retinoid treatment did not have considerable bone changes and had normal growth and development.[17] Because early epiphyseal closure and hyperostosis are important adverse effects reported in children, the dermatologist and pediatrician should cooperate in the follow-up of growth and development in children receiving retinoids.[17,19,20]

In light of reports that acitretin 0.5 mg/kg/day should be commenced in children with genetically transmitted keratinization disorders,[14,15] we started this therapy in our patient. In the 12th week of treatment, breaking and splitting of hair stopped and hair growth began. Growth and development of the patient was regu-

larly monitored by the pediatrics department. This follow-up showed that growth, development and epiphyseal cartilages were normal throughout the treatment. No adverse effect of retinoids, except for slight cheilitis and skin dryness, was found during follow-up.

The treatment was stopped in the 12th month, and the patient's symptoms resumed 4 months later. The patient is not currently receiving any treatment.

3. Conclusion

To date, no standard treatment for monilethrix patients is available. Our case report suggests that retinoids are useful for correcting monilethrix symptoms as long as the treatment is continued, but signs and symptoms will return when the treatment is discontinued.

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References

1. Zalka AD, Goldsmith LA, Scott GA, et al. Picture of the month. Arch Pediatr Adolesc Med 1995; 149: 549-50
2. Landau M, Brenner S, Metzker A. Medical pearl: an easy way to diagnose severe neonatal monilethrix. J Am Acad Dermatol 2002; 46: 111-2
3. Karaman GC, Sendur N, Basar H, et al. Localized monilethrix with improvement after treatment of iron deficiency anaemia. J Eur Acad Dermatol Venereol 2001; 15: 362-4
4. Harper J, Orange A, Prose N. Hair disorders. In: Olsen EA, editor. Text book of pediatric dermatology. Vol 2. London: Blackwell Science, 2002: 1463-90
5. Jury JS, Korges BP, Hamm M. Phenotype and prognosis in monilethrix due to mutations of hair keratins hHb 1 and hHb 6. Br J Dermatol 1999; 141 Suppl. 55: 102-7
6. Zlotogorski A, Christiano AM. Mapping of monilethrix and papular atrichia [letter]. Dermatology 1995; 190: 89
7. Smith F. The molecular genetics of keratin disorders. Am J Clin Dermatol 2003; 4: 347-64
8. Winter H, Rogers MA, Langbein L, et al. Mutations in the hair cortex keratin hHb6 cause the inherited hair disease monilethrix. Nat Genet 1997; 16 (4): 372-4
9. Thai K, Sinclair RD, Rodney D. Keratosis pilaris and hereditary koilonychia without monilethrix. J Am Acad Dermatol 2001; 45: 627-9
10. Braun-Falco O, Plewing G, Wolff HH, et al., editors. Diseases of hair. Berlin: Springer, 2000
11. Gebhardt M, Fischer T, Claussen U, et al. Monilethrix improvement by hormonal influences? Pediatr Dermatol 1999; 16: 297-300
12. de Berker D, Dawber RP. Monilethrix treated with oral retinoids. Clin Exp Dermatol 1991; 16: 226-8
13. Tamayo L. Monilethrix treated with the oral retinoid Ro 10-9359 (Tigason). Clin Exp Dermatol 1983; 8 (4): 393-6
14. Brecher AR, Orlow SJ. Oral retinoid therapy for dermatologic conditions in children and adolescents. J Am Acad Dermatol 2003; 49: 171-82
15. Hartschuh W. Successful retinoid therapy of Netherton Syndrome. Hautarzt 1989; 40 Suppl. 7: 430-3
16. Al-Khenaizan S. Papillon-Lefevre syndrome: the response to acitretin. Int J Dermatol 2002; 41 (12): 938-41
17. Traupe H, Happle R. Etretinate therapy in children with severe keratinization defects. Eur J Pediatr 1985; 143: 166-9
18. Ruiz-Maldonado R, Tamayo-Sanchez L, Orozco-Covarrubias ML. The use of retinoids in the pediatric patient. Dermatol Clin 1998; 16: 553-69
19. Prendiville J, Bingham EA, Burrows D. Premature epiphyseal closure a complication of etretinate therapy in children. J Am Acad Dermatol 1986; 15: 1259-62
20. Lacour M, Mehta-Nikhar M, Atherton DJ, et al. An appraisal of acitretin therapy in children with inherited disorders of keratinization. Br J Dermatol 1996; 134: 1023-9

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