A case report of atrichia with papular lesions

Saeid Fallah, MD [1*]
Vahideh Raeisi, MD [2]

1. Health Management and Social Development Research Center, Golestan University of Medical Sciences, Gorgan, Iran
2. Golestan University of Medical Sciences, Gorgan, Iran

*Corresponding author:
Saeid Fallah, MD
Health Management and Social Development Research Center, Golestan University of Medical Sciences, Gorgan, Iran
Email: s.fallah@goums.ac.ir

Received: 31 March 2020
Accepted: 18 July 2020

Atrichia with papular lesions (APL) is a rare autosomal recessive disease caused by a genetic mutation in the human hairless gene's zinc finger domain. It suddenly appears in the first months after birth and causes irreversible hair loss and keratin cysts on the head, face, and other body parts. Our patient is a 31-year-old female of a consanguineous marriage living in a deprived and remote city of Iran. She suffered from total hair loss and papules spreading all over the body. The patient showed no signs of other physical or mental health issues; she also had healthy physical growth and standard eyesight and hearing. Bone and teeth development were developed, and sweat glands were also robust. APL was diagnosed based on the clinical findings (total hair loss, emergence of papules, morbidity over the body from early childhood, and hypopigmented suture-like lines on the scalp), standard laboratory tests rejecting other differential diagnoses, and lack of response to all available treatments.

Keywords: atrichia, alopecia universalis, APL, autosomal recessive disease

Iran J Dermatol 2021; 24: 231-233

DOI: 10.22034/ijd.2021.136581

INTRODUCTION

Atrichia with papular lesions (APL) is a rare autosomal recessive disease, often emerging shortly after birth as sudden, irreversible hair loss. It is caused by a mutation in the zinc finger domain in the human hairless gene of the 8p12 chromosome [1,2].

The differential diagnoses of APL include alopecia universalis congenital (AUC), vitamin D dependent rickets type II, and ectodermal dysplasia. APL patients are physically and mentally healthy, with the bones and teeth developing normally. There are no abnormalities in the eyesight, hearing, and sweat glands. APL is clinically distinguished from AUC, where keratin cysts appear over the head, face, and most body parts within 2-26 years once total hair loss begins [3-5].

Atrichia with papular lesions can be diagnosed by ten clinical and laboratory criteria, the most prominent of which are atrichia, papules, clinical rejection of vitamin D dependent rickets, and a history of consanguineous marriage [2].

CASE PRESENTATION

Our patient is a 31-year-old female with total hair loss that began around six months. 0.25- 0.5 cm papules gradually appeared first on the face and head, then on the arms and legs (Figure 1). The patient lives in Maraveh Tappeh - a small town in the easternmost region and one of the most deprived cities of the Golestan province of Iran, where the socioeconomic status is poor. Furthermore, access to advanced diagnostic laboratories for genetic testing and histology is geographically challenging.

The patient was born of consanguineous marriage (the parents were cousins). Family members (including two brothers, three sisters, and parents), second-degree relatives (grandparents, uncles, and aunts), and third-degree relatives (cousins) showed no symptoms, excluding a 29-year-old brother who endured similar expressions. The patient and her brother were in normal physical and mental health, with no vitamin D deficiency and healthy kidney and liver function tests. The patient also

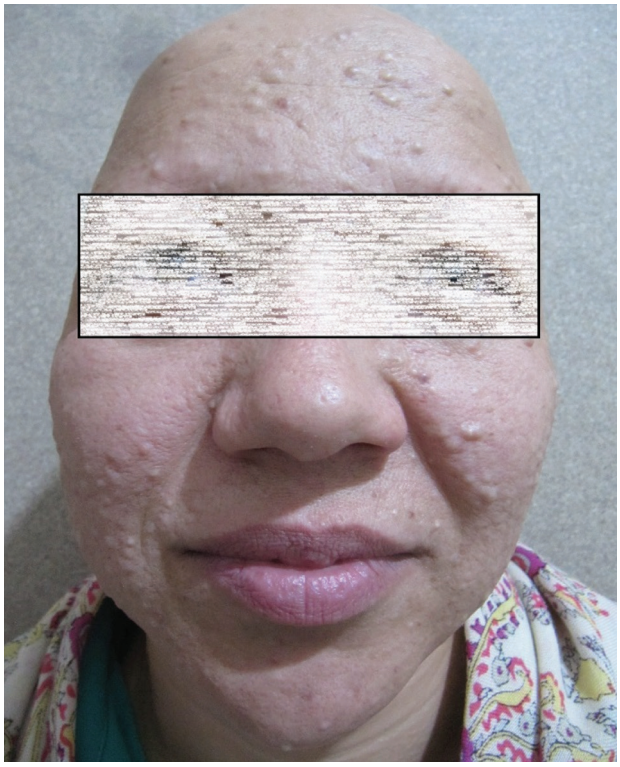

Figure 1. Total hair loss with papular lesions.

exhibited normal physical development relative to her age, healthy eyesight, hearing, bone and teeth development, and normal sweating.

The patient from the age of 2 years and her brother from the age of one underwent all available therapeutic methods until turning 20 without any response to medications. Although we conducted no biopsy and histology tests on the patient, the patient's brother was suffering from the same symptoms and had experienced biopsies and multiple experiments showing that hair follicles were substituted by follicular cysts. Unfortunately, the documents were not available to us at the time of this study.

DISCUSSION

Atrichia with papular lesions (APL) is a rare genetic disease and a type of alopecia with keratin cysts. Its diagnostic criteria were first suggested by Zlotogorski *et al.* before later being revised by Yip *et al.* (Table 1) [1].

The genetic test is the golden standard for APL diagnosis. However, as genetic test conditions were unavailable, the patient and her brother were diagnosed based on the clinical findings, including total permanent hair loss from the first months of life and milia-like papules and follicular cysts spread all over the body since infancy (Figure 1). Similar symptoms in a family member, no hair growth in the genital and axillary areas, lack of eyebrows and eyelashes, and hypopigmented suture-like lines on the scalp (Figure 2) were also used for making the

Table 1. Diagnostic criteria for atrichia with papular lesions
Atrichia with papular lesions (APL)[1]

Major criteria (4 out of 5 required for diagnosis)
1. Permanent and total loss hair in the first months after birth
2. White and milia-like papules on face, scalp, arms, elbows, thighs, or knees from infancy or childhood
3. Mature hair follicle structures replaced by follicular cysts in scalp histology; filled with cornified material
4. Mutation(s) in the human hairless gene through genetic testing
5. Clinical and/or molecular exclusion of rickets due to lack of vitamin D
Minor criteria (supplementary criteria)
1. Cousin marriage
2. Absence of secondary axillary, pubic, or body hair growth, and/or sparse eyebrows and eyelashes
3. Healthy development, including normal bones, teeth, nails, and normal sweating
4. Whitish-hypopigmented streaks on the scalp
5. Lack of response to any treatment

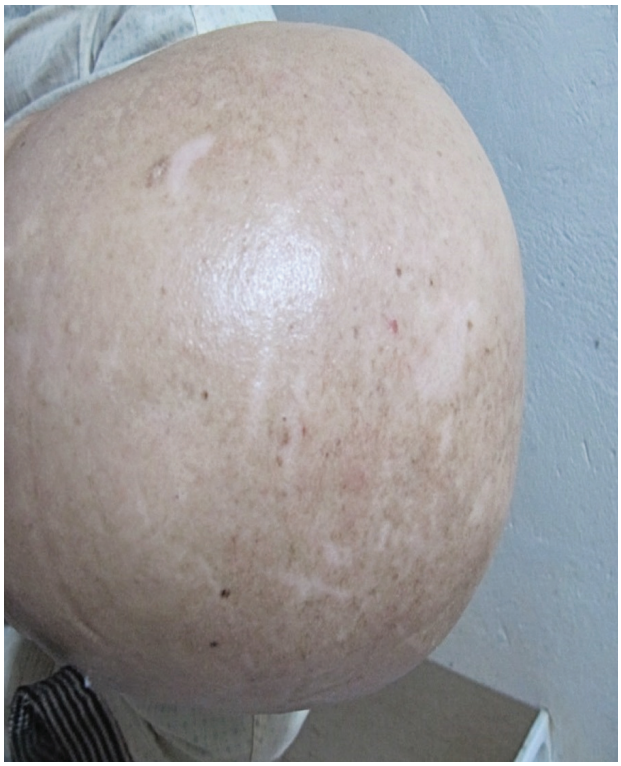

Figure 2. Whitish hypopigmented streaks on the scalp.

APL diagnosis. Healthy physical growth, laboratory results such as a normal vitamin D serum level, therapeutic experiences like a lack of response to available treatments, and the biopsy results of the patient's brother were other diagnostic factors. Furthermore, once the patient history was examined and the clinical picture was documented, some other dermatologists also verified the diagnosis.

CONCLUSION

In this study, the diagnosis of APL was made based on clinical findings and available test results considering the lack of equipment and new tests in deprived areas. As a secondary conclusion, it can be stated that in conditions where using modern equipment and sophisticated tests are unavailable for any reason, APL diagnosis relies upon epidemiological data, clinical findings, and the available tests. Such a diagnosis can prevent imposing unnecessary medical expenses while reducing suffering from ineffective therapeutic processes.

Acknowledgment

First and most, we would like to express our gratitude to Dr. Mohammad Ibrahimi Rad and Dr. Babak Soltani, dermatologists in Gorgan City, for their support and valuable remarks during this process. We would also like to thank Samad Gharavi, disease prevention and treatment expert of healthcare in Maraveh Tappeh, for his engagement.

Conflict of Interest: None declared.

REFERENCES

1. Thomas M, Daniel S. Atrichia congenita with papular lesions. Indian J Dermatol Venereol Leprol. 2011;77(1):70-2.
2. Sprecher E, Bergman R, Szargel R, et al. Identification of a genetic defect in the hairless gene in atrichia with papular lesions: evidence for phenotypic heterogeneity among inherited atrichias. Am J Hum Genet. 1999;64(5):1323-9.
3. Yip L, Horev L, Sinclair R, et al. Atrichia with papular lesions: a report of three novel human hairless gene mutations and a revision of diagnostic criteria. Acta Derm Venereol. 2008;88(4):346-9.
4. Sprecher E, Lestringant GG, Szargel R, et al. Atrichia with papular lesions resulting from a nonsense mutation within the human hairless gene. J Investi Dermatol. 1999;113(4):687-90.
5. Wang S, Tu C, Feng Y, et al. Atrichia with papular lesions in a chinese family caused by novel compound heterozygous mutations and literature review. Dermatology. 2013;226(1):68-74.