

Two Case Reports of Netherton Syndrome: Hair Shaft Examination Is Known As a Diagnostic Test

Armen Malekiantaghi¹, Ahmad Khodadad², Vajiheh Modaresisaryazdi³, *Kambiz Eftekhari⁴

¹Assistant Professor of Pediatric Gastroenterology, Pediatric Gastroenterology and Hepatology Research Center, Tehran University of Medical Sciences, Department of Gastroenterology, Bahrami Children's Hospital, Tehran, Iran. ²Professor of Pediatric Gastroenterology, Tehran University of Medical Sciences, Department of Gastroenterology, Children's Medical Center, Tehran, Iran. ³Associate Professor of Pediatric Gastroenterology, Islamic Azad University, Yazd Branch, Department of Pediatric, Yazd, Iran. ⁴Associate Professor Of Pediatric Gastroenterology, Pediatric Gastroenterology and Hepatology Research Center, Tehran University of Medical Sciences, Department of Gastroenterology, Bahrami Children's Hospital, Tehran, Iran.

Abstract

Background

Netherton syndrome is a rare autosomal recessive disorder consisting of ichthyosis form dermatosis, hair shaft abnormalities and an atopic diathesis that presents as widespread erythematous skin. The aim of these reports is emphasis on the importance of the examination of hair as a diagnose route.

Case presentation

Case 1: A 6 months old boy with respiratory distress and severe erythematous itchy scaling lesions. He had been under treatment of topical steroid without improvement. Case 2: A 28 days old boy admitted for failure to weight gain with presentation of extensive dermatologic involvement, severe dehydration and respiratory distress.

Results

Examination of hair under light microscopy revealed trichorrhexis invaginata, highly suggestive for Netherton syndrome.

Conclusion

In countries where access to genetic diagnostic tests is difficult, hair examination is the best and inexpensive definitive diagnostic method compared to the expensive genetic tests for diagnose of Netherton syndrome.

Key Words: Hair, Ichthyosiform dermatosis, Netherton syndrome, Trichorrhexis.

*Please cite this article as: Malekiantaghi A, Khodadad A, Modaresisaryazdi V, Eftekhari K. Two Case Reports of Netherton Syndrome: Hair Shaft Examination Is Known As a Diagnostic Test. Int J Pediatr 2018; 6(4): 7495-99. DOI: **10.22038/ijp.2018.29913.2634**

*Corresponding Author:

Kambiz Eftekhari (MD), Mailing Address: Pediatric Department, Bahrami Children's Hospital, Kiaee Street, Tehran Iran, Fax: +982177568809, Postal Code: 16417-44991

Email: : dr_k_eftekhary@yahoo.com

Received date: Jan.13, 2017; Accepted date: Feb.22, 2018

1- INTRODUCTION

Differentiation of disorders presenting with scaling skin feature in infancy are difficult, since many of them have similar presentation (1). Many inflammatory skin disease (psoriasis), immunodeficiency (Wiskott-Aldrich syndrome, hyper Immunoglobulin E syndrome), malignancies, genetic disease, infections and infestations (scabies), nutritional deficiencies (Acrodermatitis enteropathica), and allergic disease, and Netherton syndrome share in symptoms and signs (2).

The infant with Netherton syndrome typically displays a generalized scaling erythematous skin covered by fine, translucent scales, which can be difficult to distinguish clinically from above mentioned disorders. Netherton syndrome is a rare inherited, autosomal recessive disorder consisting of ichthyosiform dermatosis, hair shaft abnormalities (trichorrhexis invaginata), and an atopic diathesis that presents as widespread erythematous skin (3). In the first months of life it may present with poor weight gain, edema, recurrent infections and fluid and electrolytes disturbances as predominant features of disease. Recently Spink 5 is found as an abnormal responsible gene of the disease locating on chromosome 5 (2, 4).

Laboratory abnormalities that may be observed include elevated serum Immunoglobulin E (IgE), peripheral eosinophilia, hypoalbuminemia and aminoaciduria. Diagnosis is based on the clinical presentation and proved by genetic study. Finding hair shaft anomaly (bamboo shape) is very suggestive sign for diagnosis of this syndrome, since gene detection is expensive and sometimes unavailable. Here we report two cases of Netherton syndrome that both diagnosed by examination of their hair under light microscope with specific feature of the disease; named bamboo like shape. Hair

abnormality has been considered as a pathognomonic finding by some authors (5, 6).

2- CASE REPORTS

Case.1: A 6 months old boy hospitalized for respiratory distress and generalized severe erythematous itchy scaling lesions. His parents were consanguineous and one of his siblings died at the age of one with a similar pattern. Physical examination revealed cachectic appearance with extensive dermal erythematous scaling lesions, extremities edema and respiratory distress. He evaluated for causes of hypoalbuminemia. Because of normal liver, kidney and cardiac function, it seemed hypoproteinemia was due to a losing process either entropathy or dermal lesions. Liver enzymes and floctometry and immunoglobulins except IgE were in normal range. Peripheral eosinophilia was seen. Dermal biopsy suggested Acrodermatitis enteropathica and atopic dermatitis as a diagnosis.

The lesions didn't improve with administration of zinc supplement. Examination his hair under light microscopy revealed trichorrhexis invaginata, highly suggestive for Netherton syndrome (**Figure.1**).

Despite of appropriate antibiotics, electrolyte and nutritional support, his condition didn't improve properly. The severity of disease and protein losing status resulted to life threatening condition. A trial of systemic corticosteroid with doses 2mg/kg/day led to dramatic improvement of disease. Prednisolone tapered to 5mg every other day after two weeks, and discontinued within two weeks later. A low-dose of topical steroid for limited time was considered. In follow up for three years; he gained weight without systemic problems.

Case.2: A 28 days old boy admitted for not weight gain with extensive dermatologic involvement, extremities edema, severe dehydration and respiratory distress. He was born full term with 2,500gr birth weight, and hospitalized for 10 days because of pneumonia in his first days of life. His parents were consanguineous and family history for similar case was negative. Liver function tests, renal function were in normal range. Immunoglobulins except IgE were in

normal range. Examination his hair under light microscopy revealed trichorrhexis invaginata, highly suggestive for Netherton syndrome (**Figure.2**). Starting corticosteroid with doses 2mg/kg/day resulted to improvement of disease. After two weeks prednisolone, tapered to 0.2mg/kg every other day and continued for three months, and then discontinued. The response to the treatment was excellent.



Fig.1: The bamboo shape Hair shaft anomaly in Netherton syndrome with light microscopy examination (case.1).



Fig.2: The bamboo shape Hair shaft anomaly in Netherton syndrome with light microscopy examination (case.2).

3- DISCUSSION

Netherton syndrome should be considered on the top of differential diagnosis list in a newborn with erythroderma and abnormal looking scalp hair, or in an older child with ichthyosis linearis circumflexa and spare lusterless hair (2). It usually presents after two years of age but also can manifests within the first 10 days of life with unspecific dermal feature including itchy eczematous rash, and failure to thrive and other unusual presentation (1, 4). The skin defect causes massive loss of heat and water and proteins which all are essential for normal growth and development. In addition, patients are susceptible to life threatening infection because of such leaky membrane. There are no specific laboratory findings and skin biopsy specimen are generally nonspecific and unhelpful in diagnosis of disease (8). The hair shaft abnormality is often difficult to find a hair with pathognomonic features on light microscopic examination (5). There is no specific treatment and the goal is management the symptoms and prevents skin infection and other complications. Symptoms of Netherton syndrome tend to improve with age, and life span for most patients is normal. Increased risk of skin cancer is reported (11, 12).

1. Kutsal E, Gücüyener K, Bakirtaş A, Eldeş N, Oztaş M, Dursun A. Youngest netherton patient with infantile asthma. *Tuberk Toraks*. 2008;56(1):104-8.
2. Cork M, Robinson D, Vasilopoulos Y, Ferguson A, Moustafa MacGowan A, Duff G, et al. New perspectives on epidermal barrier dysfunction in atopic dermatitis: gene-environment interactions. *J Allergy Clin Immunol*. 2006; 118(1):3-22.
3. Joannie D, Sun M, Linden K. Netherton syndrome: a case report and review of the literature. *Int J Dermatol*. 2006; 45(6):693-7.
4. Macknet C, Morkos A, Job L, Garberoglio M, Clark R, Macknet KD J, et al.

Comparing to other published literatures (3, 7-9), we found our cases interesting to their early presentation, prominent systemic problems including hypoalbuminemia, malnutrition and failure to weight gain, and early diagnosis with a simple test. Both had hyper IgE that is often seen in these patients and one of them had peripheral eosinophilia that commonly seen in Netherton syndrome. Another finding was experience of excellent response to systemic corticosteroid with improving both systemic and dermal problems in our patients. Our patients were the first example, to our knowledge, that respond dramatically to corticosteroid in acute and sever phase of exacerbation. In follow up, both of our patients are in well condition.

4- CONCLUSION

We recommend examination of the hair as a simple and useful tool whenever we are faced to an infant with failure to weight gain and extensive scaling dermal lesions, an index of suspicion for Netherton syndrome, although it is a very rare condition.

5- CONFLICT OF INTEREST: None.

6- REFERENCES

- An infant with netherton syndrome and persistent pulmonary hypertension requiring extracorporeal membrane oxygenation. *Pediatr Dermatol*. 2008; 25(3):368-72.
5. Rakowska A, Kowalska-Oledzka E, Slowinska M, Rosinska D, Rudnicka L. Hair shaft videodermoscopy in netherton syndrome. *Pediatr Dermatol*. 2009; 26(3):320-2.
6. Goujon E, Beer F, Fraitag S, Hovnanian A, Vabres P. 'Matchstick' eyebrow hairs: a dermoscopic clue to the diagnosis of Netherton syndrome. *J Eur Acad Dermatol Venereol*. 2010;24(6):740-1.
7. Leclerc-Mercier S, Bodemer C, Bourdon-Lanoy E, Larousserie F, Hovnanian A, Brousse N, et al. Early skin biopsy is

helpful for the diagnosis and management of neonatal and infantile erythrodermas. *J Cutan Pathol.* 2010;37(2):249-55.

8. Krasagakis K, Ioannidou D, Stephanidou M, Manios A, Panayiotides I, Tosca A. Early development of multiple epithelial neoplasms in netgerton syndrome. *Dermatol.* 2003;207(2):182-4.

9. Katugampola R, Finaly A, Harper J, Dojcinov S, Maughan T. Primary cutaneous CD30+ T cell lymphoproliferative disorder

following Cardiac transplantation in a 15-year-old boy with Netherton syndrome. *Br J Dermatol.* 2005;153(5):1041-6.

10. Hosomi N, Fukai K, Nakanishi T, Funaki S, Ishii M. Caspase-1 activity of stratum corneum and Serum interleukin-18 level are increased in patients with Netherton syndrome. *Br J Dermatol.* 2008; 159(2):744-5.

11. Oji V, Traupe H. Ichthyosis: clinical manifestations and practical treatment options. *Am J Clin Dermatol* 2009;10(6):351-64.