Unusual presentation of cystic fibrosis as diffuse dermatitis

Sir,

Cystic fibrosis is one of the most common autosomal recessive disorders among Caucasians. Most of the presentations are related to lung disease and exocrine pancreatic deficiency. Severe dermatitis is a rare initial presentation of the disease.¹

A non-Caucasian 5-month-old boy born at term to unrelated parents (weight 3.900 g) was healthy and exclusively breastfed until 4 months of age when he developed an erythematous eruption on the chest. The child was evaluated and diverse diagnoses were proposed: atopic dermatitis, contact dermatitis and scabies. The lesions were treated with several topical formulations including corticosteroids, antibiotics and antifungals, with no resolution. Following this, the rash became generalized and the patient further developed cough and wheeze; therefore, oral antibiotics were prescribed. However, as his general condition worsened, and he also developed edema on his lower limbs, he was transferred to Teaching Hospital Edgard Santos, Salvador, Bahia, Brazil. Upon admission, the patient was very irritable with generalized edema, wheezing and diffuse erythematous papules with overlying scaling or peeling of skin involving face, chest, limbs and genital area [Figures 1 and 2]. Body weight and length recorded were 7.090 kg and 62.2 cm, and his nutritional evaluation showed a weight/age (W/A) Z-score = +0.99; height/age (H/A) Z-score = −1.75 and weight/height (W/H) Z-score = −0.52. Mucosal examination was normal and no abnormalities were found. Abnormal laboratory values were as follows: hemoglobin 9.7 g/dL, albumin 1.8 g/dL, aspartate aminotransferase 116 IU/L, alanine transaminase 60 IU/L, Na⁺ 133 mEq/L. Blood, urine and faecal cultures were negative. Initial diagnosis was drug eruption with secondary hypoalbuminemia. However, cystic fibrosis was investigated for and he presented two positive sweat tests (chloride 99/98 and 86/85 mEq/L). The steatocrit value was 22% and oropharyngeal cultures isolated *Staphylococcus aureus, Klebsiella pneumoniae* and *Pseudomonas aeruginosa*. A genetic test was performed and the mutations F508del and 3120+1G>A were identified. Fecal elastase-1 level was lower than 15 μE1/g. Pancreatic enzyme replacement was instituted, as well as vitamins, antibiotics and nutritional therapy. The patient exhibited improvement in skin lesions, edema and nutritional status. He was discharged in good clinical condition after 37 days of hospitalization. Currently, he is 7 years old and is being followed-up as an outpatient at the same multidisciplinary cystic fibrosis clinic with good nutritional status: W/A Z-score = 0.6; H/A Z-score = −0.1; body mass index Z-score = +1.38, and stable clinical parameters [Figures 3 and 4].

Severe and generalized dermatitis as an initial presentation is rare and approximately 30 cases have been described.¹² Edema, anemia and malnutrition are serious clinical manifestations of cystic fibrosis in infants. It usually occurs in infants with ages varying from 2 weeks to 15 months old.¹ The rash typically appears as erythematous papules which evolve in weeks to months into extensive plaques with desquamation. The lesions are first noted in perioral or diaper areas and extremities; subsequently, the dermatitis can become generalized with no response to topical formulations, including corticosteroids, antibiotics, antifungals or oral zinc supplementation.¹

The skin lesions could be mistaken for other diseases, delaying the diagnosis and appropriate treatment. Differential diagnosis of this...
kind of rash without associated systemic manifestations includes atopic dermatitis, psoriasis, seborrheic dermatitis, Langerhans cell histiocytosis, immunodeficiency syndromes and acrodermatitis enteropathica. Kwashiorkor and essential fatty acids deficiency must be included in the differential diagnosis, especially if other clinical features are present.\textsuperscript{1,3} This patient was previously diagnosed with atopic dermatitis, contact dermatitis and scabies.

Edema, gastrointestinal and pulmonary symptoms usually occur 1–2 months after the beginning of the rash; these symptoms were also noted in the case described. Laboratory abnormalities include hypoproteinemia, hypoalbuminemia, anemia, low cholesterol, zinc deficiency, undetectable liposoluble vitamins, steatorrhea, elevated transaminases and alkaline phosphatase.\textsuperscript{1,2,4} Most of these abnormalities were observed in our patient.

The etiopathogenesis of the skin changes in cystic fibrosis still remain elusive, but it is apparently related to concomitant deficiencies of protein, zinc, essential fatty acids and possibly copper.\textsuperscript{3,5} The essential fatty acids deficiency occurs even in patients who have pancreatic sufficiency.\textsuperscript{4} Furthermore, children are more susceptible to essential fatty acids deficiency due to high metabolic demands.\textsuperscript{2}

This atypical clinical presentation of cystic fibrosis may be mistaken for dermatitis of different etiologies, and the presence of edema and protein deficiency, which can cause false negativity in the sweat test, can contribute to diagnostic delay.\textsuperscript{2,3} Moreover, there are no pathognomonic histopathologic findings of this rash, and the microscopic aspect of the lesion is common to various diseases such as eczematous dermatitis, seborrheic dermatitis and drug reactions.\textsuperscript{1}

Early diagnosis and proper cystic fibrosis treatment are crucial for this clinical presentation since it is associated with poor prognosis and the lesions may evolve with secondary infection and septicemia, a life-threatening condition whose sequelae can be severe and progress to death.\textsuperscript{1,3} Nevertheless, the rash usually improves after 2 weeks of nutritional therapy and pancreatic enzyme replacement, as in this case.\textsuperscript{1}

**Declaration of patient consent**

The authors certify that they have obtained all appropriate patient consent forms. In the form, the legal guardian has given his consent for images and other clinical information to be reported in the journal. The guardian understands that names and initials will not be published and due efforts will be made to conceal patient identity, but anonymity cannot be guaranteed.

**Financial support and sponsorship**

This work was partially supported by the Research Support Foundation of the State of Bahia (FAPESB) PPSUS 020/2013.

**Conflicts of interest**

There are no conflicts of interest.

Bianca Sampaio Bonfim, Lais Ribeiro Mota\textsuperscript{1}, Carolina de Godoy Almeida\textsuperscript{2}, Ana Paula de Brito Aguiar\textsuperscript{1}, Renata Lúcia Leite Ferreira de Lima\textsuperscript{4}, Ângela Peixoto de Mattos\textsuperscript{5}, Edna Lúcia Souza\textsuperscript{5}

School of Medicine of Bahia, Federal University of Bahia, \textsuperscript{1}Post-Graduation Program in Interactive Processes of Systems and Organs, Institute of Health Sciences, Federal University of Bahia, \textsuperscript{2}State University of Bahia, \textsuperscript{3}Climério de Oliveira Maternity Hospital, Federal University of Bahia, \textsuperscript{4}Department of Biology, Institute of Biology, Federal University of Bahia, \textsuperscript{5}Department of Pediatrics, School of Medicine of Bahia and Teaching Hospital Professor Edgard Santos, Federal University of Bahia, Bahia, Brazil

**Correspondence:** Prof. Edna Lúcia Souza, Santa Luzia Avenue, 379/902, Horto Florestal, Salvador, Bahia, Brazil.

E-mail: souza.ednalucia@gmail.com

**References**

1. Lovett A, Kokta V, Maari C. Diffuse dermatitis: An unexpected initial presentation of cystic fibrosis. J Am Acad Dermatol 2008;58:S1-4.
2. O’Regan GM, Canny G, Irvine AD. “Peeling paint” dermatitis as a...
Letters to the Editor

4. Darmstadt GL, McGuire J, Ziboh VA. Malnutrition-associated rash of cystic fibrosis. Pediatr Dermatol 2000;17:337-47.
5. Muñiz AE, Bartle S, Foster R. Edema, anemia, hypoproteinemia, and acrodermatitis enteropathica: An uncommon initial presentation of cystic fibrosis. Pediatr Emerg Care 2004;20:112-4.

How to cite this article: Bonfim BS, Mota LR, de Godoy Almeida C, de Brito Aguiar AP, de Lima RL, de Mattos ÂP, et al. Unusual presentation of cystic fibrosis as diffuse dermatitis. Indian J Dermatol Venereol Leprol 2018;84:461-3.

Received: September, 2017. Accepted: February, 2018.

This is an open access journal, and articles are distributed under the terms of the Creative Commons Attribution-NonCommercial-ShareAlike 4.0 License, which allows others to remix, tweak, and build upon the work non-commercially, as long as appropriate credit is given and the new creations are licensed under the identical terms.