Hereditary mucoepithelial dysplasia and severe respiratory distress

Mahmoud Halawa
University of Florida

Mutasim N. Abu-Hasan
University of Florida

Mai K. Elmallah
University of Massachusetts Medical School

Follow this and additional works at: https://escholarship.umassmed.edu/peds_pp

Part of the Congenital, Hereditary, and Neonatal Diseases and Abnormalities Commons, Pediatrics Commons, Pulmonology Commons, and the Respiratory Tract Diseases Commons

This work is licensed under a Creative Commons Attribution-Noncommercial-No Derivative Works 4.0 License.

Repository Citation
https://escholarship.umassmed.edu/peds_pp/58

This material is brought to you by eScholarship@UMMS. It has been accepted for inclusion in Pediatric Publications and Presentations by an authorized administrator of eScholarship@UMMS. For more information, please contact Lisa.Palmer@umassmed.edu.
Case report

Hereditary mucoepithelial dysplasia and severe respiratory distress

Mahmoud Halawa a, Mutasim N. Abu-Hasan a, Mai K. ElMallah b, *

a Division of Pulmonary Medicine, Department of Pediatrics, College of Medicine, University of Florida, Gainesville, FL 32610, USA
b Division of Pulmonary Medicine and Gene Therapy Center, Department of Pediatrics, College of Medicine, University of Massachusetts Medical School, Worcester, MA 01655, USA

Abstract

Hereditary mucoepithelial dysplasia (HMD) is a rare autosomal dominant disorder characterized by mucoepithelial disruption of the skin, hair and mucous membranes. It results from defective gap junction formation and leads to non-scarring alopecia, mucosal erythema, perineal erythematous intertrigo, involvement of the conjunctival mucosa, and pulmonary disease. We present a case of severe respiratory distress in an initially healthy full term infant born to a mother with HMD. This infant later developed signs and symptoms of HMD. A high index of suspicion for pulmonary infection with atypical organism is essential in infants with a family history of HMD who present with respiratory distress.

© 2015 The Authors. Published by Elsevier Ltd. This is an open access article under the CC BY-NC-ND license (http://creativecommons.org/licenses/by-nc-nd/4.0/).
infiltrates (Fig. 1A). A bedside flexible bronchoscopy with BAL was repeated and revealed diffuse inflammation with mild bronchomalacia, but the BAL fluid was no longer “milky”. BAL cell count showed 51% polymorphonuclear cells, 1% lymphocytes and 48% monocytes. She was continued on Piperacillin/Tazobactam, and restarted on Vancomycin. In addition Tobramycin nebulizer therapy, Fluconazole and IV corticosteroids were initiated. She gradually improved and 5 days after her transfer to our institution, she was extubated to CPAP for a day, then to nasal cannula for a few days, then to room air. She completed 21 days of IV antibiotics, and was discharged home on room air. During her outpatient follow up visits at 3 months and 6 months, she had no cough or respiratory symptoms and her oxygen saturations were 100% on room air with a negative respiratory examination, and normal CXR (Fig. 1B).

However, she developed severe photophobia and was diagnosed with corneal keratitis. She currently sees genetics, ophthalmology and dermatology for a suspected diagnosis of HMD.

Of note, HMD is prevalent in the family of the patient’s mother, affecting at least 3 generations. The patient’s mother was diagnosed with HMD when she was 17 months old after presenting with recurrent lung infections. She also had a history of recurrent sinusitis, skin infections and visual impairment. Three young adults have died in the extended family from pulmonary complications and pneumonia (Fig. 2).

Discussion

To our knowledge, this is the first report of severe respiratory distress due to bacterial pneumonia in an infant born to a mother with HMD. Although there are no specific genetic tests for HMD, the HMD is suspected in this infant given the development of photophobia and corneal keratitis and the strong family history of HMD. HMD is a rare, possibly under diagnosed [3,4], autosomal dominant disease [5]. Pathology results from defective gap junction formation, and thus a paucity of these gap junctions. This compromise in the gap junctions affects many epithelial surfaces including the skin, eyes, mouth, and respiratory airways [2–4].

Pulmonary involvement normally occurs later in life and can be fatal. In fact, pulmonary fibrosis and pulmonary failure have been recently reported in a family with HMD [3]. Moreover, affected individuals with HMD have spontaneous pneumothoraces and “fibrocystic changes” [2]. No reports of chest imaging using computerized tomography (CT) scan have been reported to date. Although, the pathophysiology of pulmonary disease in HMD needs to be more specifically defined and examined, we speculate that disruption of the integrity of the airway epithelium increases the susceptibility to recurrent and/or chronic bacterial infections thereby affecting airway repair mechanism which potentially leads to structural damage and fibrotic changes.

The infectious organisms reported in both pulmonary and systemic infections in patients with HMD include P. aeruginosa, coagulase positive Staphylococci, Neisseria, alpha Streptococci, Pneumococci, Hemophilus influenzae, and Candida albicans [2]. Our patient presented with overwhelming respiratory and systemic infections with P. aeruginosa, MRSA, Staphylococcus, Acinetobacter and Enterobacter. At five days of age, these organisms are more likely to be acquired during vaginal delivery and less likely to be community acquired. Since the patient’s mother was diagnosed with HMD which typically has mucosal compromise and a higher rate of bacterial colonization in the perineal region, this infant was at an increased risk of infection.

In conclusion, our case illustrates the need to better characterize pulmonary involvement in HMD. In addition, there needs to be a
high index of suspicion when presented with an infant with respiratory symptoms with a known family history of HMD, or family members with the triad of non-scarring alopecia, well-demarcated mucosal erythema and erythematous intertriginous plaques [2–4].

Funding source

Parker B Francis Fellowship and NIH 1 K08 HD077040-01A1 (MKE).

Acknowledgment

Dr. Mai K. ElMallah is funded by the Parker B Francis Fellowship and by 1 K08 HD077040-01A1 (NIH).

References