



HHS Public Access

Author manuscript

N Engl J Med. Author manuscript; available in PMC 2023 September 19.

Published in final edited form as:

N Engl J Med. 2019 April 04; 380(14): 1382–1383. doi:10.1056/NEJMc1901268.

Case 40-2018: A Woman with Recurrent Sinusitis, Cough, and Bronchiectasis

Lisa Bastarache, M.S.,

Julie A. Bastarache, M.D.,

Joshua C. Denny, M.D.

Vanderbilt University Medical Center, Nashville, TN

TO THE EDITOR:

The Case Record by Mojica et al. (Dec. 27 issue)¹ highlights the importance of considering the diagnosis of cystic fibrosis in adults. We reviewed 842 cases of cystic fibrosis in our electronic health record (EHR) and found that 8.4% were diagnosed in patients older than 30 years of age. Among 368 patients who received a diagnosis of cystic fibrosis at Vanderbilt University Medical Center, the age at diagnosis has increased over time, including one woman who recently received a diagnosis at 80 years of age. Case presentation and genetics varied according to age, a finding consistent with other reports.^{2,3} Pulmonary symptoms predominated in patients older than 30 years of age, of whom 91% did not have the F508 *CFTR* mutation. Meconium ileus and failure to thrive predominated in newborns, of whom 71% were homozygous for the F508 mutation.

Diagnosing cystic fibrosis in adults can be difficult. We have proposed that combining multiple EHR phenotypes into a “phenotype risk score” may facilitate earlier identification of these patients.⁴ Indeed, the 47-year-old woman who was described in the Case Record had a phenotype risk score in the 98th percentile for cystic fibrosis.

References

1. Case Records of the Massachusetts General Hospital (Case 40-2018). *N Engl J Med* 2018;379:2558–65. [PubMed: 30586519]
2. Jain R. Diagnosing cystic fibrosis in adults: better late than never. *Ann Am Thorac Soc* 2018;15:1140–1. [PubMed: 30272501]
3. Rodman DM, Polis JM, Heltsh SL, et al. Late diagnosis defines a unique population of long-term survivors of cystic fibrosis. *Am J Respir Crit Care Med* 2005;171:621–6. [PubMed: 15591474]
4. Bastarache L, Hughey JJ, Hebring S, et al. Phenotype risk scores identify patients with unrecognized Mendelian disease patterns. *Science* 2018;359:1233–9. [PubMed: 29590070]

lisa.bastarache@vumc.org .

No potential conflict of interest relevant to this letter was reported.