Oman Medical Journal [2022], Vol. 37, No. 1: e344

Can We Justify Cystic Fibrosis Mutational Analysis among Omani Neonates?

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ARTICLE INFO

Article history:
Received: 14 April 2021
Accepted: 18 August 2021

ONLINE:
DOI 10.5001/omj.2022.44

Dear Editor,

In the March 2021 issue of the *Oman Medical Journal* by Al Balushi et al,[1] reported a novel cystic fibrosis (CF) gene mutation C.4242+1G>C in an Omani neonate. The reported mutation expands the spectrum of CF mutations in Oman already addressed in the literature. I agree with the authors' call on the need for extensive genetic testing for CF diagnosis during the neonatal period. This is based on the following dual points. First, CF is a cumbersome disease in Oman. The available data pointed out the predicted CF prevalence of 1 in 8264 and the estimated carrier frequency of CF of 1 in 94.[2] Second, consanguinity, a major risk factor for CF cases, is a common phenomenon in Oman. The published data revealed that consanguineous marriage is culturally preferred in Oman, accounting for 49% of marriages.[3] Although genetic testing for CF mutations improves the diagnosis of symptomatic patients and helps identify asymptomatic carriers and at-risk couples, two factors might hamper its implementation among Omani neonates. First, the undetermined mutations are sizable, and > 2000 different mutations have been detected worldwide.[4] Second, the large number of tested mutations among neonates, which is essential for the better efficiency of testing, implies more financial cost.[5] Nevertheless, mutational analysis remains a justifiable option to contain a further rise in CF prevalence in Oman and minimize its long-term detrimental effects.

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