Letter to the Editor: Novel Cystic Fibrosis Gene Mutation C.4242+1G>C in an Omani

Patient: A Case Report

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Received: 14 April 2021
Accepted: 18 August 2021

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DOI 10.5001/omj.2022.44

To the Editor:

In March 2021 issue of the Oman Medical Journal, Al Balushi et al.\(^1\) reported a novel cystic fibrosis (CF) gene mutation C.4242+1G>C in an Omani neonate. The reported mutation truly expands the spectrum of CF mutations in Oman already addressed in the literature. I do 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 to the predicted CF prevalence of 1 in 8,264 and the estimated carrier frequency of CF of 1 in 94.\(^2\) Second, consanguinity, a major risk factor for breeding of CF cases, is a noticeable phenomenon in Oman. The published data revealed that consanguineous marriage is the culturally preferred type of marriage 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 still enormous and more than 2000 different mutations have been detected worldwide.\(^4\) Second, the larger the number of tested mutations among neonates 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.
References:


