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

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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:

1. Al Balushi S, Al Balushi Y, Al Busaidi M, Al Mutawa L. A Novel Cystic Fibrosis Gene Mutation C.4242+1G>C in an Omani Patient: A Case Report. Oman Med J 2021 Mar;36(2):e243.
2. Fass UW, Al-Salmani M, Bendahhou S, Shivalingam G, Norrish C, Hebal K, et al. Defining a mutational panel and predicting the prevalence of cystic fibrosis in oman. Sultan Qaboos Univ Med J 2014 Aug;14(3):e323-9.
3. Mazharul Islam M. Consanguineous marriage in Oman: understanding the community awareness about congenital effects of and attitude towards consanguineous marriage. Ann Hum Biol 2017 May;44(3):273-86.
4. De Boeck K. Cystic fibrosis in the year 2020: A disease with a new face. Acta Paediatr 2020 May;109(5):893-9.
5. Rosenberg MA, Farrell PM. Assessing the cost of cystic fibrosis diagnosis and treatment. J Pediatr 2005 Sep;147(3 Suppl):S101-5.