Oman Medical Journal [2022], Vol. 37, No. 1: e345

Letter in Reply: Can We Justify Cystic Fibrosis Mutational Analysis among Omani Neonates?

Said Al Balushi[1]* and Younis Al Balushi[2]

[1]Pediatric Department, Sohar Hospital, Sohar, Oman
[2]Pediatric Department, Royal Hospital, Muscat, Oman

ARTICLE INFO
Article history:
Received: 18 August 2021
Accepted: 18 August 2021

ONLINE:
DOI 10.5001/omj.2022.45

Dear Editor,

We appreciate the author's positive comments about our article published in the March 2021 issue of the *Oman Medical Journal*.[1]

We agree with your suggestion of the justification question on the need for genetic sequencing testing of the cystic fibrosis transmembrane conductance regulator (CFTR) gene for cystic fibrosis (CF) diagnosis during the neonatal period. This is especially true for those highly suspected neonates, even with a negative family history of CF and our patient is a good example.

As we know, CF is a serious disease that can cause severe respiratory and gastrointestinal complications, which can be prevented or minimized by early detection and treatment, ultimately improving the overall prognosis later in life. Also, there has been a significant rise in the number of people with CF in Oman, similar to many gulf countries with high consanguinity rates.[2] However, due to socioeconomic status, the disease in Oman is still diagnosed based on clinical features of the patient and a sweat chloride test.

The main barriers to genetic testing of CFTR mutation in Oman, similar to other countries, are the cost and the undetermined mutations,[3] as > 2000 different mutations have been detected worldwide.[4]

The consideration on the need for extensive genetic testing for CF diagnosis during the neonatal period should be considered according to disease prevalence and the cost versus benefit of early diagnosis and treatment.

We believe our article can provide information on areas where we can work with the Ministry of Health in Oman. With the recent establishment of a Genetic Centre at the Ministry of Health and the availability of services, such a task would not be difficult.

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-e329.
3. Kerem B, Rommens JM, Buchanan JA, Markiewicz D, Cox TK, Chakravarti A, et al. Identification of the cystic fibrosis gene: genetic analysis. Science 1989 Sep;245(4922):1073-1080.
4. De Boeck K. Cystic fibrosis in the year 2020: a disease with a new face. Acta Paediatr 2020 May;109(5):893-899.

*Corresponding author: ✉dr.saeed86@hotmail.com