A letter in Reply :
A Novel Cystic Fibrosis Gene Mutation C.4242+1G>C in an Omani Patient.

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Received: 18 August 2021
Accepted: 18 August 2021

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DOI 10.5001/omj.2022.45

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 option on the need of genetic sequencing testing of CFTR gene for CF diagnosis during the neonatal period especially for those highly suspected neonate, even with negative family history of CF as our patient gave us a good example.

As we know,CF is a serious disease, which can cause severe respiratory and gastrointestinal complications that can be prevented or minimized by early detection and treatment ,ultimately to improve the overall prognosis later in life..Also,,There has been a significant rise in the number of people with cystic fibrosis in Oman similar to many gulf Countries with high consagunity rates. [2] However,due to socioeconomic status ,still the disease in oman is 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 as there are more than 2000 different mutations have been detected worldwide. [3]

Basically, the consideration on the need of extensive genetic testing for CF diagnosis during the neonatal period should be given to disease prevalence and the cost vs. benefit of early diagnosis and treatment of such disease. .

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

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