Correspondence

Clinical profile of cystic fibrosis. *Atypical presentation*

To the Editor

I read the interesting study by Najada and Dahabreh[1] on the clinical profile of cystic fibrosis. I have 2 comments on the aforementioned study.

First, cystic fibrosis (CF) is common in Jordan with a reported prevalence of 39 per 100 000 live births.[2] Its prevalence is anticipated to grow in near future. Pediatricians must be alarmed with the high index of suspicion to diagnose CF, particularly in typical CF. However, the diagnosis is cumbersome in atypical cases. Hence, the study by Najada and Dahabreh[1] significantly augments and broaden the clinical alertness and knowledge of pediatrician on CF.

Second, the increasingly reported cases of atypical CF worldwide have made the diagnosis of CF less straightforward for many pediatricians. The clinical profile of CF is protean as CF phenotype is determined by mutations in the CF gene, genetic background, and environment.[3] The study by Najada and Dahabreh[1] addressed 2 interesting points: 1) Atypical CF constituted 8% of the studied CF children. This seems higher than 2% reported worldwide.[4] This might reinforces the suspicion that the prevalence of CF in Jordan is currently more than what was previously reported.[2] In addition, it might reflect the prevailing of undisclosed numerous mutations involving CF transmembrane conductance regulator (CFTR) genes. 2) Najada and Dahabreh[1] rely solely upon the clinical profiles that involved single preponderant feature to address them as atypical CF. However, atypical CF was addressed to include cases with pancreatic exocrine sufficiency, normal or borderline sweat chloride concentrations, or with a single predominant clinical feature.[4] In these atypical cases, the confirmation of the diagnosis of CF requires the detection of one disease causing mutation on each CFTR gene or direct quantification of CFTR dysfunction by nasal potential difference measurement.[5] Considering the aforementioned criteria might expand further the scope of atypical CF in Jordanian children.

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Reply from the Author

Regarding the comment of Prof Al-Mendalawi, our study included only those children with classic (typical) CF, and our aim was to report the unusual (atypical) clinical manifestation of classic CF in children with classic CF in Jordan. These manifestation were early pulmonary hypertension in infancy, hydronephrosis, ichthyosis, severe iron deficiency with short stature, and one single episode of Pseudo-Bartter syndrome in infancy in the absence of other features of CF. These presentations constituted 8% of the initial presentations of classic CF. We did not discuss atypical CF and it was stated in our study[1] that patients with borderline sweat chloride were excluded (atypical CF is borderline sweat chloride and organ involvement specific for CF). We meant by classic (typical) CF sweat chloride >60 and organ involvement specific for CF. I agree with Prof Al-Mendalawi that worldwide atypical CF is approximately 2%, but from the practice I think it could be more than that in Jordan. We are not aware on the common mutations we have and we think the full DNA study on the whole genome at a national study should be carried out.

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