Correspondence

Insights and considerations on CFTR variant reporting in a study of cystic fibrosis patients in Saudi Arabia

CFTR 1548del G and 1549del G. *Navigating the discovery of novel mutations*

To the Editor

I read with interest the article titled, "Geographic distribution of common cystic fibrosis variants in Western and Southern Regions of Saudi Arabia," by Almaghamsi et al,[1] which was recently published in the Saudi Medical Journal. The study was carried out at King Faisal Specialist Hospital and Research Center, Jeddah. It offers valuable insights into the prevalence and distribution of cystic fibrosis (CF) variants in Saudi Arabia. However, I would like to address a point regarding the novel variant mentioned in the study.

The study reports the identification of a novel cystic fibrosis transmembrane conductance regulator (CFTR) variant, 1549del G, in 2 patients with CF, which had not been previously reported or published in Saudi Arabia. Upon closer examination, it appears that this variant may not be entirely novel, as it was previously documented as 1549del G in 2001.[2] Furthermore, reading the study reference, it was found that this may be a known polymorphism in the Saudi population as 1548del G rs397508205.[3,4] Further sequence clarification is warranted to accurately characterize the variant and its significance within the context of CF genetics.

Despite this discrepancy, the study remains instrumental in highlighting the genetic landscape of CF in the Western and Southern regions of Saudi Arabia. The identification of common CFTR variants and their geographic distribution provides valuable information for genetic counseling, diagnostic testing, and therapeutic interventions tailored to the Saudi Arabian population.

I commend the authors for their efforts in advancing our understanding of CF genetics in Saudi Arabia and encourage further research to elucidate the genetic factors contributing to CF in diverse populations. By fostering collaboration and knowledge-sharing, we can continue to improve the care and outcomes of individuals affected by CF worldwide.

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Reply from the Author

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