Cystic fibrosis: The urgent need to report on mutations among patients of African descent

In this *SARJ* issue, Mphahlele *et al.*[1] have reported on a condition that is being increasingly diagnosed in people of African descent: cystic fibrosis (CF). The article will contribute to rectifying the misconception that this condition is restricted to people of European descent and underscore the need to further investigate the mutations specific to people of African descent. The delayed age of diagnosis, specifically among patients of African descent, further illustrates the need to educate health professionals, to improve their awareness and raise their index of suspicion of CF in patients of African ancestry. The research also emphasises the need to initiate newborn screening for CF, which has been shown to be critical in identifying the condition in non-white patients in the USA.[2]

In the Mphahlele *et al.*[1] study, the absence of any detected mutation in almost all the non-white patients is consistent with the literature, as this is expected with the currently used 30-mutation *CFTR* panel for which the diagnosis rate of CF in non-European populations is very low.[2] However, mutations in *CFTR* in South Africans of black African ancestry were first reported 2 decades ago;[3] a specific mutation found in black South Africans, *3120+1G>A*, which was initially reported in three African-American CF patients, has been shown to account for 9 - 14% of African-American CF chromosomes.[4] This mutation (*3120+1G>A*) is now routinely tested for in selected medical genetic settings in South Africa (SA).[5] The investigation of specific *CFTR* mutations in various populations of non-European ancestry is gaining increasing attention globally, especially in countries that, like SA, have a population of diverse ethnic background, such as Reunion Island,[6] Brazil[7] and the USA.[2] This research is shedding light on ethnic-specific variants that cannot be detected by the routinely used 30-mutation *CFTR* panel. It is, therefore, reasonable to call for urgent further studies using the full sequencing of the *CFTR* gene in patients of non-European ancestry in SA, in Africa and globally, to reveal more specific mutations that will increase the diagnostic validity and yield of the molecular testing of the *CFTR* gene.

A well-designed *CFTR* expanded panel using ethnic-specific variants is desirable to improve CF carrier detection rates within specific populations in SA, specifically in populations of African ancestry.

Ambroise Wonkam
Division of Human Genetics, Department of Medicine, Faculty of Health Sciences, University of Cape Town, South Africa
ambroise.wonkam@uct.ac.za

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