Scientists hail new cystic fibrosis treatment

October 31 2019

Alexandra Caldas, a 23-year-old woman suffering from cystic fibrosis competes in a nautical marathon, on September 14, 2019 off Bora-Bora in French Polynesia

A new triple-drug therapy that tackles the genetic causes of cystic fibrosis has been shown to be highly effective in treating the rare life-threatening disorder, scientists reported Thursday following landmark

clinical trials.

The finding—30 years after the gene that causes the disease was identified—was called a "cause for celebration" by Francis Collins, the director of the National Institutes of Health who was part of the team that made the original genetic discovery.

Trikafta is a combination of three drugs that target the CFTR gene responsible for the disease, which affects about 30,000 Americans and results in the formation of thick mucus build-up in the lungs, digestive tract and other parts of the body.

That in turn results in respiratory and digestive problems, and exposes patients to complications such as infections and diabetes, with average life expectancy in the 30s and 40s—and historically even lower before advances in drugs that alleviate symptoms.

The new therapy targets the most common mutation of the CFTR gene, the Phe508del mutation, which represents around 90 percent of cases.

"The results of a pair of phase 3 clinical trials in the Journal and in a simultaneous publication in the *Lancet* document impressive benefits," Collins wrote in an editorial for the *New England Journal of Medicine* on Thursday.

The two trials examined how much air patients could expel in a forced breath, an established marker of the disease's progression.

In the first trial, mean levels increased by 13.8 percent compared to a placebo, while in the second trial, mean levels rose 10 percent from baseline compared to an earlier drug combination.

"This should be a cause for major celebration," said Collins, but he

added that more work was needed on patients with other forms of mutations who would not respond to the new therapy.

Beyond that, he added, the best outcome would come when the more than 70,000 people with the disease worldwide do not need drugs because of a permanent cure achieved through gene editing.