Helping People Breathe

Cystic Fibrosis and Gene Manipulation

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Cystic fibrosis is a genetic disease that affects different parts of the body, primarily the lungs. The symptoms of cystic fibrosis can include persistent lung infections, an inability to breathe, constant coughing, phlegm accumulation, poor growth/weight gain, and bowel movement problems. Patients with cystic fibrosis have an average lifespan of 30 to 40 years. However, due to some recent discoveries, this lifespan might be increased.

One of the reasons why scientists and medical professionals have struggled to address cystic fibrosis is because it is a genetic disease that can't be prevented, unlike bacterial or viral infections. Cystic fibrosis is caused by the presence of two copies of a recessive allele. Therefore, two parents, both of whom carry one allele for the gene, have a 25 percent chance of having offspring who have cystic fibrosis and a 50 percent chance of having offspring who are carriers. That specific recessive allele exists due to a mutation in the gene, and there are more than 1,000 known mutations which result in cystic fibrosis. The resulting effects also vary in the patients due to the different mutations and different genetic makeups of the patients.

Several treatments—such as pancreatic enzyme supplements, inhaled medication, and airway clearance—have helped add many years to the lives of cystic fibrosis patients. Furthermore, investment from different initiatives is currently being devoted to finding more cures and treatments for the disease, while also furthering understanding of its foundations.

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One of the most recent new discoveries about cystic fibrosis considers the genetic makeup of patients who are less severely affected by the disease. Carried out by a team of researchers at the Boston Children's Cystic Fibrosis Clinic, an analysis of the coding sections of the genome—which can contain the most disease-causing mutations—in these patients genetic variants that might be function in cystic fibrosis pa

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The Boston Clinic t the genetics of the other pati disease. If the team's ongoin cystic fibrosis all over the worl easily.

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Easier

found a set of rare and previously unknown
responsible for life longevity and better lung
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hannels (ENaCs), which are semi-permeable
for reabsorbing sodium in the kidney, colon,
result, the scientists hypothesized that these
ydrate the airways of cystic fibrosis patients,
e to build-up of detrimental bacteria. This
could help serve as the foundation for a new,
treatment. If the ENaCs can be effectively
on against the progression of cystic fibrosis

eam is now doing further research to analyze
ients who are affected more severely by the
g research proves successful, people with
d might be on their way to breathing more

out cystic fibrosis, check out "About
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