

Cambridge firm unlocks fatal lung disease

By PATRICIA MANGAN

In a startling discovery that will provide a new way to treat cystic fibrosis, scientists have discovered that the fatal disease is caused by a protein deficiency disorder.

"We now have an understanding of what goes wrong in the cells of patients with cystic fibrosis," said researcher Alan E. Smith.

The finding supports the idea that cystic fibrosis may be treated by having patients inhale an aerosol to deliver the protein into its proper place.

Cystic fibrosis is a genetic disorder that affects about 30,000 children and young adults in the United States.

Their lungs produce a thick,

sticky mucus that can cause fatal lung infections. The abnormal mucus appears because cells cannot expel charged particles called chloride ions, while they absorb other particles called sodium ions too rapidly.

That results from defects in the gene that tells cells how to make a protein called cystic fibrosis transmembrane conductance regulator, or CFTR.

The genetic defects lead to faulty versions of CFTR.

The new study suggests that most cases of cystic fibrosis occur because these versions never reach their proper place on the cell surface.

Results of the studies show

that the critical protein remains stuck inside the cell, where it begins to degrade, and never makes to the cell's surface, where it belongs.

"It's absolutely surprising and not what we expected at all," Smith said. "We expected the protein to be present, but not working."

The study, to be reported in today's issue of the journal *Cell*, is the second major advance in cystic fibrosis research announced by the Cambridge-based Genzyme Corp. in two months.

In September, researchers announced they had repaired defective cells that cause the disease.

Smith said the latest discov-

ery will allow scientists to think differently about treating the disease.

"A treatment for a protein deficiency disease is to add back the protein," he said. "We've done that with other

genetic diseases."

Smith said a patient would need regular treatments in protein replacements, the way treatments are given to hemophiliac patients who receive purified blood proteins.

Take A Friend Along.

\$ 5



City of Cambridge

45.

IN CITY COUNCIL

November 19, 1990

COUNCILLOR SULLIVAN

- WHEREAS: It has come to the attention of this City Council that Genzyme Corporation, a Cambridge based research firm, has just announced they have discovered that cystic fibrosis is caused by a protein deficiency disorder; a fact which will provide a new way to treat this illness; and
- WHEREAS: This finding supports the idea that cystic fibrosis may be treated by having patients inhale an aerosol to deliver the protein into its proper place; and
- WHEREAS: Cystic fibrosis is a genetic disorder that affects approximately 30,000 children and young adults throughout the United States; now therefore be it
- RESOLVED: That this City Council go on record extending its sincere congratulations to Alan E. Smith and his colleagues at Genzyme Corporation for this terrific break through and to further encourage them to continue this most important research on behalf of the people of America who suffer from this illness.

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In City Council November 19, 1990.

Adopted by the affirmative vote of nine members.

Attest:- Joseph E. Connarton, City Clerk.

A true copy;

ATTEST:-

A handwritten signature in black ink, reading "Joseph E. Connarton".

Joseph E. Connarton
City Clerk

Order # 45

S-1109

Councillor Sullivan discovery of Genzyme
Corp. relative to cystic fibrosis.

In City Council,

November 19, 1990

Order adopted