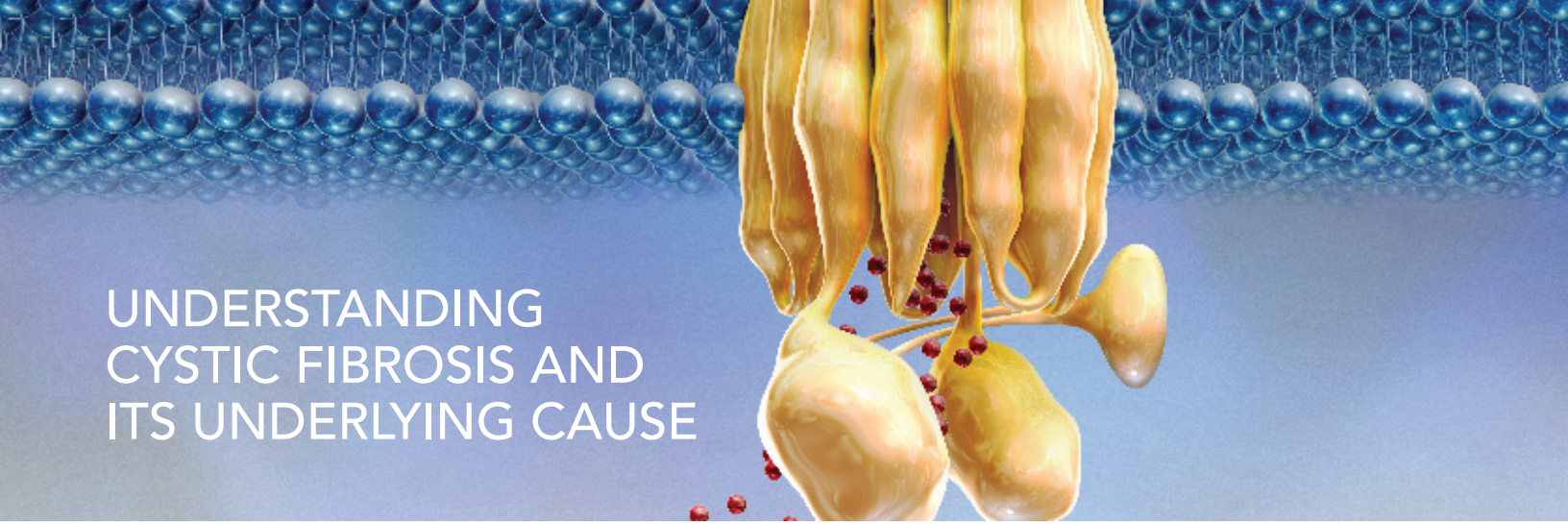

UNDERSTANDING CYSTIC FIBROSIS AND ITS UNDERLYING CAUSE

>> FAST FACTS

- Cystic fibrosis (CF) is a life-threatening, genetic disease for which there is no cure. It is caused by defective cystic fibrosis transmembrane conductance regulator (CFTR) proteins that result from mutations in the *CFTR* gene.[1,2]
- Approximately 70,000 people worldwide have CF, including 30,000 in the United States,[3] nearly 3,800 in Canada,[4] 35,000 in Europe,[5] and 2,500 in Australia.[5]
- Approximately one in 3,500 children in the United States is born with CF.[6]

CAUSE:

- CF is a life-threatening genetic disease caused by defective or missing CFTR proteins, which result from mutations in the *CFTR* gene. CFTR proteins are essential to regulating the flow of salt and water into and out of cells in a number of organs, including the lungs.[7]
- When the proteins don't work properly or don't exist, abnormally thick, sticky mucus accumulates and can cause chronic lung infections and progressive lung damage.[7]
- Children must inherit a defective *CFTR* gene from each parent to have CF. One out of four children born of two parents with the defective *CFTR* gene inherit CF.[8]
- People who inherit a defective *CFTR* gene from one parent and a normal copy from the other are CF carriers. Although they have no symptoms, two carriers can pass the disease on to their children.[8]

DIAGNOSIS AND PROGNOSIS:

- Most people with CF are diagnosed by six months old through routine newborn screenings. The results are usually confirmed through a sweat test and a genetic test.[7]
- Every week 12 young people with CF in the United States either die or get a lung transplant.
- The median predicted survival for a person with CF born today is approximately 38 years.[6]
- The average age of death for people with CF has not changed in the last 25 years and remains in a person's mid-20s.[6]
- Lung disease is the leading cause of death among people with CF. It is caused by recurring infections and chronic lung inflammation.[9]

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SYMPTOMS AND MANIFESTATIONS:

- In people with CF, abnormally thick, sticky mucus blocks the passages in a number of organs and leads to a variety of symptoms. In particular, mucus builds up and clogs airways in the lungs creating an environment for bacteria to grow.[7]

UNDERSTANDING CFTR

The CFTR protein works like a channel, regulating the flow of salt and water into and out of cells in a number of organs. A healthy balance helps protect the lungs by creating a thin, watery layer of mucus. When the proteins don't work properly or don't exist, abnormally thick, sticky mucus accumulates and can cause chronic lung infections and progressive lung damage.[7]

CFTR MUTATIONS

- There are more than 1,800 known *CFTR* mutations.[6]
- Approximately 90 percent of people with CF have least one copy of the F508del mutation.[6] In these people, the CFTR protein is defective and does not generally reach the cell surface.[10]
- G551D is a mutation that is found in approximately 4 percent of people with CF.[6] G551D is one of a number of "gating mutations," so called because although the CFTR protein reaches the cell surface, it does not open (gate) properly to allow for a healthy exchange of salt and water into and out of the cell.[1]

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