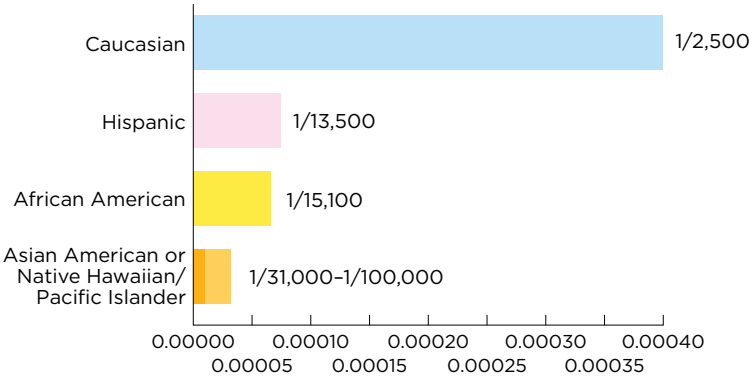
Cystic Fibrosis (CF)

Cystic Fibrosis (CF) is a lifelong, hereditary disease that causes thick, sticky mucus to form in the lungs, pancreas, and other organs. In the lungs, this mucus blocks the airways, causing lung damage, making it hard to breathe, and leading to serious lung infections. In the pancreas, it clogs the pathways leading to the digestive system, interfering with proper digestion.[1] In 90 percent of cystic fibrosis cases, the airways are affected.[2]

Who Has It

CF is the second most common life-shortening, inherited disorder occurring in childhood in the United States, after sickle cell anemia.[3] Approximately 30,000 Americans have CF, and there are an estimated 1,000 new cases diagnosed each year.[4] The overall birth prevalence of CF in the United States is 1 in 3,700 (Figure 1).[5] It occurs equally in male and female babies and affects nearly every race.[6] However, cystic fibrosis occurs most commonly among Caucasians of Northern European descent; an estimated 1 in 2,500 Caucasian births are affected.[7]

Figure 1: Estimated Cystic Fibrosis Prevalence Rates by Race and Ethnicity



Source: CDC. Cystic Fibrosis Clinical Validity. September 10, 2007

More than 10 million Americans are unknowing, symptomless carriers of the defective cystic fibrosis gene. In order to develop CF, an individual must inherit a defective gene from each parent. Each time two carriers of the defective gene conceive, there is a 25 percent chance that the child will have CF. There is a 50 percent chance that the child will be a carrier of the gene, and 25 percent chance that the child will not have the gene at all. The odds remain the same with each child. The severity and symptoms of the disease vary considerably due to different mutations of the gene.[8]

Deaths

Between 1999 and 2006, 3,708 people in the U.S. died from cystic fibrosis. Most of these deaths were among Caucasians (3,355). The age-adjusted death rate among Caucasians (0.22 per 100,000) is much higher than that among other racial and ethnic groups (Figure 2).[9]

Survival

In the 1950s, few people with CF lived to go to elementary school. In 1985, the median survival age was about 25 years. In 2007, the predicted

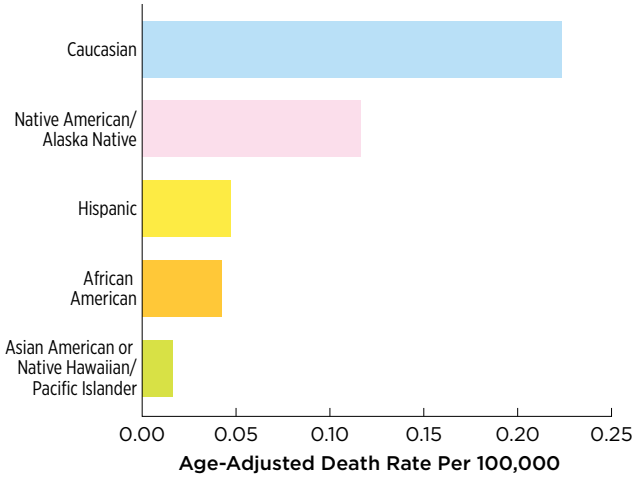
survival age was 37.4 years. Thanks to dedicated researchers and earlier diagnosis, the age of survival for patients with CF continues to increase.[10]

Symptoms and Diagnosis

As a genetic disease, CF begins at conception, though symptoms may not appear at first. Diagnosis is sometimes delayed for decades because of mildness of the symptoms or failure to recognize them. Typical symptoms include:[11,12,13]

- salty-tasting skin (which parents often notice when they kiss their child)
- wheezing or shortness of breath
- persistent cough and excessive mucus
- frequent lung infections, such as pneumonia and bronchitis
- frequent sinus infections (sinusitis)
- growths in the nose (nasal polyps)
- poor weight gain and growth
- foul-smelling, greasy stools
- swollen belly, accompanied by abdominal gas and discomfort
- broadening of the fingertips and toes

Figure 2: Cystic Fibrosis Age-Adjusted Death Rates by Race and Ethnicity, 1999-2006



Source: NCHS 2005

Early identification of CF through newborn screening programs has led to improved survival, better lung function and growth with less intensive therapy, and reduced cost of therapy. So far, 37 states have adopted newborn screening programs.[14] The sweat test remains the standard diagnostic test for cystic fibrosis. It measures the amount of salt in a child's sweat, with a high salt level indicating that a person has cystic fibrosis. Genetic testing is available for cystic fibrosis, but it does not detect all of the mutations that can cause the disease.[15]

Treatment

Though a cure for cystic fibrosis has yet to be developed, patients have a variety of options to treat their symptoms. Common treatments for those with CF include airway clearance techniques and medications to clear mucus from the lungs, prevention and management of infections, and proper nutrition.[16]

Racial/ Ethnic Differences

African Americans

Cystic fibrosis is much more common among Caucasians than among African Americans. It is estimated that 1 in 15,100 African Americans are diagnosed with cystic fibrosis, compared to 1 in 2,500 Caucasians (Figure 1, above).[17] In 2007, of the 24,511 patients in the Cystic Fibrosis Foundation's Patient Registry,

only 4.2 percent were African American;[18] about 12 percent of the U.S. population is African American.[19]

Between 1999 and 2006, there were 131 deaths due to cystic fibrosis among African Americans. The age-adjusted death rate of 0.04 per 100,000 for this population was lower than most other racial and ethnic groups (Figure 2, above).[20]

The Delta F508 genetic mutation, the most common CF gene mutation, has been found in approximately 44 percent of African American individuals with clinically diagnosed cystic fibrosis. In comparison, it has been found in over 72 percent of Caucasians with CF.[21]

Hispanics/Latinos

Cystic fibrosis occurs in about 1 in 13,500 Hispanics, compared to 1 in 2,500 Caucasians (Figure 1, above).[22] Only 6.8 percent of people in the Cystic Fibrosis Foundation's 2007 Patient Registry were Hispanic,[23] even though Hispanics represent 15.4 percent of the total population.[24]

There were 178 Hispanic deaths due to cystic fibrosis between 1999 and 2006, and age-adjusted rate of 0.05 per 100,000. This rate falls towards the lower end of rates for all racial and ethnic groups (Figure 2, above).[25]

The Delta F508 genetic mutation, the most common CF gene mutation, has been found in 54 percent of Hispanic individuals with clinically diagnosed cystic fibrosis, compared to over 72 percent among Caucasians with CF.[26]

Asian Americans and Native Hawaiians/Pacific Islanders

There are limited data available on cystic fibrosis for Asian Americans and Hawaiians/Pacific Islanders. One national study found that 1 in 31,000 to more than 1 in 100,000 Asian American births are affected by cystic fibrosis. This is a lower birth prevalence than Caucasians (1 in 2,500), African Americans (1 in 15,100), and Hispanics (1 in 13,500; Figure 1, above). Research in Asian countries find much lower rates and it is likely that most Asian American cases result from having one Asian and one Caucasian parent.[27]

There were only 16 deaths among Asian Americans and Pacific Islanders due to cystic fibrosis between 1999 and 2006. The age-adjusted death rate for this group is unreliable due to so few deaths, but was around 0.01 to 0.02 per 100,000 for this period, making it the lowest among all racial and ethnic groups (Figure 2, above).[28]

American Indians/Alaska Natives

Due to their small numbers in terms of the U.S. population, available data on cystic fibrosis among American Indians/Alaska Natives are limited. Small sample sizes mean that estimates are not considered statistically accurate and are not published or released. However, research indicates that CF may be common among American Indians and Alaska Natives, especially the Pueblo and Zuni[29]

Between 1999 and 2006, there were only 26 deaths due to cystic fibrosis among Native Americans and Alaska Natives. However, the age-adjusted rate for this population was second only to Caucasians at 0.12 per 100,000 (Figure 2, above).[30]

Resources

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