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Biomedical Research on Cystic Fibrosis and My Life

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Approximately 32,000 people in the US today have Cystic Fibrosis. Right now, most of them cannot expect to see age 40. My mom works to help discover a cure for this deadly disease. She analyzes data from studies on mice and people, working to save thousands of lives. I am researching this because I may be interested in a biomedical research career myself.

Cystic Fibrosis (CF) is a genetic disorder that happens when a child inherits defective CF genes from both parents. The defective genes result in the body making abnormally thick, sticky mucus. This mucus clogs the lungs and hinders the digestive system from doing its job. CF patients have trouble breathing and are very susceptible to lung infections. In the 1950s, most people with CF died in elementary school. Now, thanks to research, the life expectancy is 37 years.

One of the most important events in CF research was the discovery of the CF gene in 1988. Using this discovery, people have made a mouse model for CF. This is a strain of mice that has had the CFTR genes (the CF genes) mutated by scientists, giving the mice a form of CF. This model has greatly helped research on CF, because it enables us to test new treatments on mice to make sure they are safe and effective before giving them to people. Although this is not a perfect imitation of human CF, it is still extremely helpful in predicting how the treatment will work in people.

Recently there has been a new drug invented for CF using the mouse model. This drug causes a certain mutation in the CFTR genes to be ignored, so the CFTR genes can function normally and make CFTR protein. Scientists have given this drug to the CF-model mice, and the mice were able to make normal CFTR protein, which seemed to

function normally, too. There were no bad side effects observed in the mice. Because of this mouse model result, scientists are able to quickly move forward with a study of this drug in people.

My mom is a biostatistician who works for the Cystic Fibrosis Foundation. She plans studies and analyzes data from studies on animals and people. This includes using probability to make sure enough people or mice are studied to get a reasonably trustworthy result. For example, if a new is given drug to only two people and they both improve, one can't tell if it was just chance that they both improved due to natural ups and downs in symptoms or if it was the drug doing its job. She uses mathematics to figure out new study procedures that use fewer people but still get the same amount of information. This is useful because the fewer people need for a study, the less the study costs and the faster the results can be obtained. Additionally, she tries to figure out if there are any flaws in experimental plans. Some flaws are hard to find and require knowledge of the disease, experiment procedures and mathematics to figure out.

We have not yet found a cure for CF, but there are many treatments to make the lives of people who have it better. For example, it was recently discovered that breathing in vaporized saltwater helps clear the lungs of CF patients. Also, there is a vibrating vest that CF patients can put on to help clear their lungs. Even though there is no cure for CF yet, studies on animals, especially mice, have greatly helped to advance our knowledge and to bring us closer to a cure. In fact, the new drug talked about earlier that makes the body ignore a certain CFTR mutation might become a cure for the 10% of CF people that have this mutation! This drug is the most promising drug yet for CF, and it was discovered thanks to research with mice.

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Mary J. Emond PhD, Associate Professor of Biostatistics, University of Washington. Personal interview; assistance with medical terminology. 17 Feb. 2009.

Reflection

I chose to write about Cystic Fibrosis (CF) and research on it using mice for many reasons. One was because my mom studies CF, not just to make money but because she wants to help save peoples' lives. I might want to do a job that helps other people be healthier, too, so I decided to find out more about CF research and what my mom does. I wanted to understand more of what she was saying. Also, I feel really badly for the kids who have to live with it. I wanted to know if there was anything I or we could do and see what progress is being made on a cure for it. Another thing ~~is~~ I wanted to know how mice and other animals are being used for research. I learned a lot. I learned that there is actually a special kind of mouse made for research on CF and that because of this research we might have found a cure for about 10% of the people with CF! This is really cool, and I will stay tuned to see what happens with this. I also found out there are animal models used for research on many other diseases. I never realized just how important these animals are for keeping people healthy. From my reading for this project, I also know more now about what biomedical researchers do, including people besides my mom. I would like to help in making biomedical breakthroughs, and I will definitely be considering a job in biomedical research for my future.