**Sometimes it**

***is***

**All in the Genes**

**Part I—"The Genetic Test"**

by

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"Your pregnancy seems to be progressing just fine, but we'll do some blood work to be on the safe side. As long as we're drawing blood from you today, Nancy, would you consent to participate in one of the genetics studies we're conducting?” Dr. Kwin prided herself on putting her patients at ease, but her question caught Nancy a little off guard.

"What would that entail, Dr. Kwin?" While Nancy believed Dr. Kwin wouldn't ask her to do anything that wasn't a good idea, she never said yes to anything until she had all the information.

Dr. Kwin began to explain, "Well, as you know, PBH is a research and teaching hospital. One research team is trying to determine the frequency of the gene for the genetic disorder cystic fibrosis in the U.S. population. And since you are having blood drawn anyway as you enter your second trimester...."

Genetic disorder? Cystic fibrosis? The phrases made Nancy feel panicky. She interrupted Dr. Kwin mid-sentence. "You don't think I have that disease, do you? Isn't cystic fibrosis serious? How could I have caught it?" The questions came out in a rush.

Hearing the anxiety in Nancy's voice, Dr. Kwin quickly reassured her. "No, no. I'm quite sure that you do not have cystic fibrosis. You can't 'catch' it; you can only inherit it. If you did have cystic fibrosis, you would have been diagnosed when you were a toddler. You have absolutely nothing to worry about. However, let me emphasize that if you feel uncomfortable about participating in this study in any way, just say 'No.' My feelings won't be hurt. There is no pressure to participate."

"If it's inherited and I don't have it, why would they want to test me?" The situation still didn't make sense to Nancy.

"First, let me explain some basics. Cystic fibrosis, or CF, is a disease that is caused by defects in a particular gene. Actually, because CF is genetic, 'disorder' is a better term than 'disease.' The word disease should really be used for illnesses caused by bacterial or viral infection. However, people use both words for CF.

"The reason that they need to include people who don't have CF in the study is that cystic fibrosis is a recessive disorder. With a recessive disorder, a person has to have two defective versions of the CF gene to have CF. Therefore, you can be a carrier of one defective version but not have CF at all. It is precisely that which has inspired the study. They want to know what percentage of people in the United States carries one defective gene for cystic fibrosis. That percentage is called the gene frequency and can provide a lot of information for people working on CF."

Dr. Kwin went on to explain, "CF is one of the most common genetic disorders among Americans of European descent. Although it has many symptoms, the worst are severe respiratory problems that typically lead to death around the age of 30. One of the first reliable tests for any genetic disorder was the one developed for CF in 1990. Because CF is one of the first inherited illnesses that researchers have been able to get a handle on, these sorts of genetic screenings are pioneering work not only for CF research but also for understanding other genetic ailments."

Nancy considered the flood of information for a few seconds. "I can't see what harm it would do. You run so many tests on me anyway. What's one more?"

"I'll have one of the staff bring you the consent forms and explain the procedure to you further. *If* you do consent, your biggest decision will be whether or not you want to have the results reported to you. You need to think carefully about that question before signing the consent form."

As Dr. Kwin left the room, Nancy thought to herself, *what are the pros and cons of knowing the results?*

**Questions:**

1. Discuss why Nancy might or might not want to know the results of her blood test for CF.
2. Dr. Kwin told Nancy that she has "absolutely nothing to worry about." Although Nancy cannot get CF, is Dr. Kwin's statement entirely correct?
3. Did Dr. Kwin provide Nancy with enough information about cystic fibrosis and the test to make a good decision?
4. Should Nancy consent to the test? Provide the reasoning for your answer.
5. Name five steps Nancy should take to ensure the health of her baby moving forward

Nancy had found it hard to concentrate at work that afternoon and it was almost impossible to think about what she was doing as she prepared dinner that evening with her husband Jake. Suddenly Jake's voice snapped her back to the kitchen, "Nancy, you're about to put the carrots and cucumbers in the garbage." Jake surveyed the salad bowl where she had been working and smiled at the rare opportunity of catching his wife in a mistake. "And is vegetable peel salad your latest craving?"

"Oh, honey, I need to talk to you about something." The stress in Nancy's voice quickly wiped the smile off of Jake's face. "Remember when I went for my pre-natal checkup a couple of weeks ago, and I told you I was participating in a genetic study?" "Yeah, something about MS or something, right?"

"No, CF, cystic fibrosis." Nancy corrected him. "Well, it turns out that I tested positive."

"What!? How did this happen? You don't seem sick at all."

In response to his panic, Nancy calmed down and explained, "No, no, of course *I'm* not sick.

Remember, I told you that they were testing to see if I was a genetic carrier. How it happened was that I inherited one bad version of the gene, a bad 'allele' they call it, for cystic fibrosis from one of my parents."

He felt he should know why, but slowly Jake asked, "If you don't have CF, what makes you so worried that you're throwing sliced cucumbers into the garbage?"

"Because there is a chance that I could pass my bad CF allele to our child. And if you are a carrier and pass on your CF allele, then she or he *will* have CF."

"CF is pretty serious, right?"

"Yes, pretty serious."

Trying to be optimistic, Jake asked, "Yeah, but what's the chance that I'm a carrier too?" "Dr. Kwin says the latest research indicates 1 in 29," Nancy answered soberly.

Jake's jaw dropped. "Can I get tested? What is the likelihood that you'll pass on the CF gene? What is the chance that I'll pass on the gene? What is the chance our baby will have CF?"

"Yes you can, and *will* get tested. I don't care if you are afraid of needles." Nancy left no room for further questions there. "I talked to Dr. Kwin at length today and I have a pretty good handle on this genetics thing. So, turn down the stove, sit down with me, and I'll answer your questions."

"Let's start with the basics,"Nancy began. "We all have tens of thousands of different genes in each of the cells in our body. These genes provide the instructions on how to make all the components of our body. For almost all of those genes, we have two copies of each different gene. I inherited one copy of the gene that can cause CF from my mother and one from my father. One of them, I don't know which, was a defective copy, what they call a 'mutant' allele. One of the alleles I got from my parents was a normal, working copy. Because CF is what they call 'recessive,' I'm fine because I have that one good copy. It takes two bad copies of the gene to give you cystic fibrosis." **Questions:**

Help Nancy finish answering Jake's questions.

1. The normal or good copy of the CF gene can be written shorthand as "F", and the mutant or bad copy of the CF gene can be represented as "f". Using this shorthand style, write out Nancy's genetic make-up for this gene.
2. What is the chance that Nancy passed on the CF allele to her baby?
3. What is the chance that Jake passed on the allele if he is a carrier?
4. What is the chance that their baby will have CF if they are both carriers? If their first child has CF, what is the chance that their second child will have CF? What is the chance that the baby will inherit CF if only Nancy is a carrier?