



MAILBAG

Q: Three years ago while undergoing routine prenatal blood tests I found out that I had the genetic mutation of CF. This came as a total shock, but my mother, father and two sisters had a genetic test done on them and they were all carriers. I'm just now trying to do some research on it and find out what I can do to minimize the chances of symptoms because I haven't had any yet. I would like to have more children and I did not know if something like that would put me at greater risk. I absolutely need information about what to watch for and anything that might put me at greater risk. I was diagnosed at 21 and now I'm 24. Some information would be great, and if there is something I can do to help with researching this I would be more than happy to try. Thanks and I look forward to getting some good information from you.

Taylor Suske, Georgia, USA

A: A diagnosis of the illness Cystic Fibrosis (CF) can only be done by sweat test and/or by genetic detection of the paired mutation necessary to establish the diagnosis of CF. This mutation must be identified on both chromosomes of the pair n°9 which reside in the nuclei of the cells of the body. Per definition the illness CF is present only if the mutation is present in both of the chromosome pair.

You mention that you have one mutation for CF, found in a routine prenatal blood test. It is not clear, however, if you carry it in only one of your chromosomes n°9, or if is present in both of these chromosomes. You might even have another one of the more than 1500 different CF mutations known in the chromosomes n° 9. In the case that you have one mutation in one of the chromosomes n° 9, you would only be a carrier for CF and your condition is called "heterozygoty". If the mutations are present in both chromosomes n°9, the condition is called "homozygoty" and the characteristic for CF illness would be present. A carrier of one CF mutation is a healthy person, like your parents and sisters, and will never have any CF symptoms. On the other hand, if you have the CF mutation in both your chromosomes n° 9 you would, in fact, have the illness. In your case this must be clarified.

If you really have CF, on the basis of your description you have an extremely mild form of the illness, since despite a pregnancy until now no CF symptoms have appeared. In other words your form of CF would be so mild that no symptoms have been expressed until this day.

This means that your present condition does not pose any problem for a second or a third child. It is an advantage to know your genetic diagnosis of the CF condition in the case that CF symptoms do appear before, during or after future pregnancies, in order to treat the CF symptoms in the proper way. The diagnosis of CF is like a label, but it does not mean that every case is necessarily a severe case of CF. From your letter it is not clear whether you have the CF illness or not, but you seem to be a CF carrier, who is in effect a healthy person.

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