



Genetics and Lung Disease

What should I know about genetics and lung disease?

If you compare the genes of a group of people, you will see that there are many different kinds of genes. No-one else has exactly the same genetic make-up as you, unless you have an identical twin. These variations in genes between people (called *genetic variants*) influence things such as your height and your blood type. Some variations can also make you more or less likely to develop certain diseases such as asthma. Usually the effect of a variation in a gene is very small, so even if you have a gene that can cause asthma, you may never actually get asthma. So, a change in a gene (a *genetic variant*) does not predict for sure if you will get asthma, but it may increase your risk of getting asthma. For a few lung diseases, genes play a much bigger role. These are mainly rarer diseases that can run in families, such as cystic fibrosis, pulmonary hypertension (high blood pressure in your lungs) and alpha-1 antitrypsin deficiency that can cause liver and lung disease. The environment can also interact with genetic variants and contribute to lung diseases (called *environmental triggers*), but we do not yet understand how this happens.

Should I be tested to find out if I have problems with my genes?

Genetic testing for most *common lung diseases* is not recommended at this time. We still do not fully understand the role of genes in these diseases and their interactions with environmental triggers. Gene tests cannot *accurately* predict if you will get diseases such as asthma or COPD. However, research in this field is making good progress and in the future, gene testing might help predict who is most likely to get the disease and which drugs might be most effective in treating the

disease. The exception is alpha-1 antitrypsin deficiency, a lung disease where the genetics is well understood.

If you have a lung disease where genes play a big role, or if you have a family history of this type of disease and you are worried that you might have inherited the condition, then genetic testing might be helpful for you. Talk to your lung physician (pulmonologist) or health care provider. They can put you in contact with a genetic counselor who can explain what tests are available and what information the test will give you. Sometimes genetic testing can be helpful to confirm the diagnosis of a disease, especially for cystic fibrosis and alpha-1 antitrypsin deficiency. A gene test might therefore be recommended alongside other clinical diagnostic tests for these conditions.

What kinds of information will I learn if I have genetic testing?

The information you will learn will depend on what disease is being tested and your own personal situation. It is important that you talk to a genetics specialist who is trained to explain this information. For example, if you have pulmonary hypertension (PH), some forms are caused by gene changes but others are not. You might choose to take a gene test to find out if you have the genetic form. If you are a healthy person but you have a family history of PH, you might choose to have a gene test to find out if you inherited the gene and are at risk of getting PH in the future. The results of these tests can have a big impact on other members of your family, so it is important to think things through carefully before taking a genetic test. A genetic counselor can explain what the test involves and how the results might affect you and your family. This will help you decide if genetic testing is right for you.

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