Nontuberculous mycobacteria in people living with cystic fibrosis

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April 6, 2022

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Imagine that ever since you were a child you have difficulty to breath and you have repetitive infections in your lungs. Also, two or three times per year you are admitted to the hospital for treatment, just so you can continue breathing normally. Imagine also, that way before COVID, every time you met someone new or did something outside, you had a risk of catching an infection. This is the reality of cystic fibrosis patients.

In my dissertation, we are studying a group of bacteria called nontuberculous mycobacteria, because they belong to the same family as the one that causes tuberculosis. These bacteria live in the soil and the water and we are all exposed to them. However, they are only dangerous for people with problems in their lungs, like those living with cystic fibrosis.

In them, it can cause two conditions: infection or disease. So, what is infection? Infection is when they have the bacteria in the lungs, but is not causing any damage. One out of five people progress to disease, we don't know why. And disease means that the bacteria is actively damaging their lungs and compromising their ability to breathe.

In my thesis, I approached this problem from two different perspectives. First, I explored how many people with cystic fibrosis worldwide actually have infections or disease by these bacteria.

I performed something called a systematic review, which means that together with my collaborators, I searched all available research studies in this topic. Then, I reviewed, analyzed and extracted information from them to calculate how big is the problem for the cystic fibrosis community. My results show that 4% of the people with cystic fibrosis are affected by these bacteria. A larger risk than the normal population, where only 50 out of 100,000 are affected. My results are really important for authorities and researchers. Authorities can use my results to properly allocate resources to solve this problem. While researchers can use them as reference material to evaluate interventions and plan new research projects.

In the second part of my thesis, we wondered why some people develop disease and what is the role of patient's genetics in this progression. My results show that people who advanced to disease had a stronger activation of their immune response genes than those that stayed at the infection stage. But, as they got worse, this means that their immune response is probably not protecting them but damaging them. These genes we found are really important because they will let us develop a test that predicts the risk of progression to disease by these bacteria in people with cystic fibrosis. This way, clinicians will be able to provide better care by categorizing the risk of a patient and focusing their efforts in those with higher risk of progression, while reassuring those with lower risk.

Thank you very much