

In the Lab with Alba Timon-Gomez, PhD

Q&A with an MDA Development Grant-winning Researcher

Growing up in Spain, Alba Timon-Gomez, PhD, was encouraged by her biology teacher to see how far she could take her aptitude for the subject. This experience led to her current position as a Postdoctoral Associate at the University of Miami's Miller School of Medicine, where she focuses on mitochondrial myopathy research. In fact, she and her team have been awarded an MDA Development Grant of \$210,000 for three years to study mitochondrial respiratory chain plasticity regarding how it is regulated in healthy and stress-induced cells.

We checked in with Dr. Timon-Gomez to learn more about her and her work.

How did you get into research on mitochondrial myopathies?

It was by chance. I was always interested in how cells adapt to different stresses, and mitochondria have a central role in this process. My thesis on this topic used yeast as a model organism. For my postdoctoral studies, I decided to come to Miami to Dr. Antoni Barrientos' lab to work on human cells to address questions on disease mechanisms. He offered me the chance to participate in this project, and I thought it was interesting to try to understand the pathogenesis of mitochondrial myopathies.

What research is the MDA Development Grant funding?

Many of these mitochondrial myopathies are affecting the machinery that produces energy inside the cell. This machinery is inside the mitochondria, and it's called the mitochondrial respiratory chain.

It's dynamic, and this dynamism explains how some patients with the same mutation have different clinical effects. Our goal in this grant is to understand the plasticity of this machinery that produces energy to understand the molecular biology of these diseases.

Specifically, we study the proteins HIGD1A and HIGD2A. These two proteins have a role in the formation of this respiratory chain that will produce the energy, but they are only activated in low-oxygen conditions. For example, during high intensity exercise, or at high altitude, or if you are diving, situations like these. Our hypothesis is that these proteins regulate the formation of the respiratory chain and its dynamism to allow the cell to adapt the energy production to different energetic situations. It's important because high levels of HIGD1A have been shown to decrease the clinical symptoms in animal models of mitochondrial myopathies. We want to understand how they are doing that.

How will this grant advance your research?

We were studying these proteins in normal conditions but with this grant we are also going to study them in low-oxygen conditions and under different types of stress. We hope our results will allow us to understand new mechanisms of the formation and dynamics of the machinery that produces energy in the cell.

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How will this research advance understanding of mitochondrial myopathies?

I think results will improve our understanding about the molecular factors underlying mitochondrial myopathies. Low oxygen levels have been shown to improve the clinical symptoms and increase the lifespan in mouse models of mitochondrial myopathies. Also, increasing levels of HIGD1A in those models was decreasing their symptoms. We hope our studies will impact our understanding of these molecular pathways that lead to these diseases.

How might this lead to treatments and therapies?

It's not possible to put every patient at low oxygen levels because they would have to be sent to high altitude places, for example. They also tried to do intermittent hypoxia treatment and it didn't work. So, we are trying to increase the levels of these proteins, HIGD1A and HIGD2A, in some patients to try to understand how it increases the cell survival. This could also lead to the discovery of new therapeutic targets. Also, maybe doing gene therapy to increase the level of these proteins will help the patients to reduce their symptoms. Of course, it's a long path ahead.

What do you like to do outside the lab?

I love traveling around the world and playing sports. In my free time, I play tennis, volleyball, and go to the gym.

What are the some of the places you've enjoyed visiting the most?

I would say Hawaii because of the amazing landscapes and nature. Also, Thailand — I really enjoyed learning about the culture.