



Where I work Lucía Spangenberg

In this picture I'm using a MinION Mk1C, a portable, cheap and easy-to-use DNA-sequencing machine, to detect SARS-CoV-2 in a nasal swab. This is part of the work the Pasteur Institute of Montevideo is doing to combat coronavirus in Uruguay. Our team has been analysing DNA samples from all over the country to detect clusters of the virus and its variants.

Doing genetic sequencing this easily is still exciting to me. About 20 years ago, this work was very time-consuming, and the price tag of sequencing the whole human genome was around US\$3 billion. Today, it can be done in a week for around \$1,000. That means the technology is increasingly available, and can help more people worldwide.

In 2002, when I finished secondary school, I was torn between studying biology and informatics. So, I googled 'informatics and biology' and the term 'bioinformatics' came up. It was not something I'd ever heard of, but it combined the two subjects I loved most.

This field has such huge potential to help people with rare diseases, who might

otherwise spend years without a correct diagnosis. This science can help to steer patients to better treatments.

That is why I and two colleagues formed a start-up, called GenLives. In Uruguay, genetic sequencing is still used mainly for research purposes. The goal of GenLives is to make it available to anyone who needs it. Since we started in 2015, we've helped hundreds of patients.

There are still illnesses that remain a mystery – about 50% of our cases remain unsolved, which is pretty standard in this sort of work. That is why, in this field, sharing your data is so important. One patient's data could help with another person's diagnosis. International DNA databases are growing fast, and I hope that, soon, all the remaining cases will be solved.

Lucía Spangenberg is a bioinformatician affiliated with the Pasteur Institute of Montevideo in Uruguay and co-founder of the company GenLives. She is also a teacher at the Catholic University of Uruguay. **Interview by Patricia Maia Noronha.**

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by Pablo Albarenga.