Democratising data-driven healthcare

On the news that Sophia Genetics has sequenced 100,000 patient genome data, we spoke with **CEO, Jurgi Camblong**, about how data-driven healthcare solutions could revolutionise patient care and precision medicine. Sophia Genetics AI software is used by hospitals worldwide to better diagnose patients suffering from congenital disorders and cancer.

"Since 2000, there has been a huge race to sequence the first human genome and for a long time we haven't understood the power of such data. In 2010-2011, when sequencing costs decreased, hospitals started to use these technologies, a bit naively, to better diagnose patients for hereditary congenital disorders.

When we started in 2011, our ambition was simply to help the maximum number of patients in the world to move into this new era of medicine – which we called data-driven medicine – leveraging on the first technologies that will be adopted by the hospitals using digitalised patient information in genomics. The opportunity in this area was that hospitals would be properly equipped but would need high support to render this data accurate for clinical diagnostics.

We started with the top university hospitals that were all equipped with these platforms. By doing that, we were being exposed to much more complexity than other companies in this area. We have worked on data being produced by hundreds of hospitals and we've been exposed to a lot of different cases. This is the unique way to build AI, from bottom-up; AI cannot be applied top-down – never believe what you will hear from the 'big guys'.

The way you build it is exactly the way we learned to build self-driving cars, learning from 'us' making different routes every day. What counts is the number of cases you face and the number of different patterns to recognise, so that your algorithm can learn from these patterns and can leverage on that to make data accurate and spot the genetic variants/mutations that are unique to a patient – to explain the cause of a disease.

We started with this approach and this led us to where we are today – managing the largest network in the world: over 250 hospitals that are all connected, all pulling patient data into a unique platform and sharing their knowledge about the clinical relevance of these variants.

This is what Barack Obama spoke about a year ago when he launched Precision Medicine Initiative at the White House. He explained that the problem was not anymore about data production, the problem was an operational problem. The problem was about creating a collective intelligence, where hospitals would be connected to pull data and share knowledge. Once you have done that, when you're in a network, you can leverage on network effects and you can learn from the data that are being put into the system, but also from the human expertise from the clinical experts that are making decisions.

By the end of this quarter, we will have supported the clinical diagnostics of over 100,000 patients with genome profiling that have been analysed by our artificial intelligence. The data are being crunched and a clinician takes a decision – we're talking about an actionable 100,000 genome profiles.

With legacy technologies patients could be misdiagnosed, whereas with this technology it is much more precise. We can look at things we were not previously able to look at. For example, if you were suffering from a melanoma, you would be given many different drugs. We were treating cancer according to the tissue type – big mistake. This was because we didn't understand cancer; we've been able to understand cancer only since we've been able to sequence DNA. Even in lung cancer, from one patient to another it will be very different, according to the molecular events. Thanks to our approach and thanks to the adoption of genome sequencing, we can understand the molecular events behind a disease. So, we can give an appropriate treatment that fits this molecular event and not the disease tissue."