Interview: Jurgi Camblong â?? CEO, Sophia Genetics, Switzerland

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30.03.2017

Tags:

Switzerland, Sophia Genetics, AI, Genetics, Digitalization, Epidemiology

As Artificial Intelligence and Next Generation Sequencing started to converge, Sophia Genetics was created to bring the world towards an era of real-time epidemiology. CEO Jurgi Camblong discusses the companyâ??s worldwide impact thus far, and the next steps towards making Data-Driven Medicine a reality for patients around the world.

What transformation in medicine led to the creation of Sophia Genetics?

The world is rapidly approaching an era of Data-Driven Medicine as two big technological advances are currently colliding in the field of genomic medicine. The first is big data analytics or artificial intelligence. The other transformative advance has been the arrival of Next Generation DNA Sequencing (NGS).

With the arrival of NGS, producing genomic data is no longer a challenge, and the possibilities to learn from the analysis of large sets of patient data, including complete genomic profiles, are immense. Patients can now be clustered based on a wide range of attributes, and outcomes data can be studied to see how previously analyzed or diagnosed patients from specific clusters responded to different treatments, thus providing physicians with precious information about which medicine to prescribe moving forward.

However, seizing those opportunities requires the creation of large patient data sets, and that experts and hospitals move away from silo modes and connect to one another to start sharing their knowledge for further use by Artificial Intelligence (AI) technology. As usual with AI, the more data is pooled, the more AI systems will be able to learn and create value for patients.

What is the role that Sophia Genetics is playing in pushing the world toward the era of real time epidemiology?

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Sophia Genetics has developed SOPHiA, an artificial intelligence which is connecting hospitals and experts, and analyzing the genomic data they produce. Our technology has been well received by healthcare stakeholders because it addresses their main concerns.

A first concern is standardization. In fact, NGS data can be incredibly â??noisyâ?• or â??dirtyâ?•, and as such difficult to analyze and compare. This noise results from the various technologies used to produce the genomic data. Because of this noise, clinicians were initially reluctant to shift to NGS and continued to rely on legacy technologies. Standardization was an important step to make NGS mainstream. This is the first challenge Sophia Genetics took on, and we developed SOPHiA to clean up NGS data and take out this noise, effectively standardizing data produced using different NGS technologies. This was an incredibly complex task, and is probably the most complex that anyone in the field of big data analytics has taken to date. Our customers use our Software as Service (SAAS) platform to enter raw NGS data into our system and get back clinical grade genomic data within two hours.

Another concern is privacy, as genomic data can be considered as the most precious personal information. Robust data security measures are a must to get partnersâ?? buy-in. Thus, from the start we have invested a lot in data encryption and security to handle patient data in a secure and private manner.

How do you assess Sophia Geneticsâ?? impact thus far, and your progress in terms of building up this network of healthcare institutions sharing knowledge?

In terms of impact, the main KPI from our perspective is the number of patients being analyzed using SOPHiA. Back in 2014 we supported the diagnosis of 5000 patients, in 2015 that increased to 20 000, and last year we helped diagnose 60 000 patients. For 2017, we expect to surpass 100 000 by Q1, and by 2020 we expect to hit the 1 million patients per year mark.

With respect to building up the network, we are proud to have now more than 250 hospitals and over 600 experts who are geneticists and biologists using our platform. We are also very proud to have extended our network not only to a number of hospitals in Europe or Latin America, but even to institutions in Africa; we very recently signed a deal with an institution in Cameroon.

In these more remote regions hospitals canâ??t necessarily afford their own sequencers, but with the participation of some of the university hospitals we work with we have created a network whereby second and third tier hospitals can get access to our platform and capabilities by sending samples to our clients who effectively become sub-contractors. This more than anything else demonstrates how our platform is helping to democratize genomic analysis capabilities and helping more and more people get access to this level of diagnosis and care.

What will be some of the next steps for Sophia Genetics in terms of enhancing the capabilities of your Artificial Intelligence systems?

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Since the launch of SOPHiA we have been engaging in a constant stream of partnerships with other companies from our field so that our clients can easily get access to their capabilities.

To truly continue enhance the power of SOPHiA, we must start collecting data that are not yet available to us, and adding more information to build a larger collective intelligence. In the case of oncology, we are also considering information related to tumor type and stage. We do not yet have indication on therapy outcomes, or even other common patient data like their medical history. Being able to feed this information to SOPHiA will really bring us to the era of real time epidemiology, where we can study the factors explaining why treatments are more or less effective for different patients, and the genomic and other causes behind that.

There are two barriers here that we are currently working to overcome. One is to get more physicians handling this data by taking advantage of SOPHiA.

The second issue is that many researchers and physicians who do use SOPHiA are not particularly willing to share their knowledge. This I find difficult to understand, because as a physician you should work to help as many patients as possible, including patients being treated by other physicians.

On both fronts the key is once again ensuring that our users have an incentive to use SOPHiA, and that we provide actionable insights to them. For example, for some complex disease areas, users are now adding symptom information to allow us to filter the important genes quickly and deliver results faster; the users get the benefit of faster results, while we get more data that SOPHiA can learn from.

Another example is that for germline disorders, we now have a feature where users can flag new genetic variants as either pathogenic or benign; here, the user gets to claim finding a new or rare variant amongst his peers, other physicians who see the same variant later get more information about how other experts have interpreted the data with which to make a better decision, and once again SOPHiA gets more data to learn from.

In fact, thanks to the expertsâ?? variant flagging, SOPHiA is now able to classify the variants associated to certain germline disorders as a clinician would do in 98% of cases.

How might your Artificial Intelligence system and the data you are collecting eventually be able to be used to develop new therapies?

Much of the potential on this front comes from learning more about the genomic factors behind why different patients respond to the same medication differently. This can guide treatment selection, but also the development of new therapies, new treatment cocktails. For example, if we consider that today patients are receiving a drug A because they have mutation X, through the study of a large number of patient cases we might be able to find that patients with mutation Y as well as X actually do even better on this treatment. With such a finding, it would then be possible to look for an existing drug or develop a new one which replicates the effect of gene Y.

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