

Wellcome Genome Campus | andrea_cabibbe_tb_genomics_interview (1080p)

My name is Andrea Cabibbe. I'm a research associate at the San Raffaele Institute in Milan. And we are a supranational reference laboratory for tuberculosis and a WHO collaborating centre in laboratory and TB laboratory strengthening.

And our main field of interest is the molecular diagnosis, is the epidemiology, so molecular surveillance of tuberculosis. And not only limited to tuberculosis, but in our unit we also work with other infectious pathogens. And we are very-- pretty much interested in novel technologies. And in this regard, genomics is playing a crucial role for our everyday work in the laboratory.

So the role of genomics in TB surveillance actually is already a reality, because since 2020 we have recommendations from the WHO to use sequencing-- next generation sequencing-- as a tool for drug resistance surveillance in countries. So it's a reliable tool that can replace other technologies, maybe more demanding or more challenging, to predict the drug resistance trends in countries within the surveillance project.

So and the next step for genomics, for TB, let's say, in the field of tuberculosis beyond surveillance will be the translation into the use for clinical management of patients and people with tuberculosis.

The field of genomics even for infectious diseases, like tuberculosis, is evolving quite rapidly. So now, we can choose among several different manufacturers producing sequencing instruments and also different applications, for example, targeted sequencing from patient samples, or whole genome sequencing for bacterial cultures.

So the technology is there. The applications are there. The point now is to implement an appropriate infrastructure to perform sequencing globally-- worldwide. I mean, we know that the burden of TB is particularly high in certain regions of the world. And there is where we need to build appropriate infrastructure, capacity, supply chains for using next generation sequencing.

And in this regard, the COVID-19 pandemic can represent a suitable environment in terms of laboratory capacity to expand the use of sequencing also to other diseases, like tuberculosis.

Yes. Different steps of complexities at the different steps of the next generation sequencing workflow. And let's say that the basic is that we need a good knowledge of the genomes that we are studying in terms of biomarkers for resistance, in terms of investigations to disclose transmission outbreaks, for example.

So the point is that we need year after year collecting data from all over the world. We need to increase our knowledge of the mechanism-- genetic mechanisms-- that are at the basis of, for example, drug resistance in tuberculosis.

And to do that, really, we need to collect data from different countries, different laboratories. And we need to have a huge representativeness of genomes at global level in order to develop good diagnostics or good-- based on, of course, DNA, molecular diagnostics, or good strategies to control the disease, like including NGS, into the diagnostic algorithms, or into the surveillance algorithms.

And as another step that is important as well is to educate and train all the people that are involved, let's say, in the NGS workflow and cascade, including the clinicians, for example, that received the NGS reports to take clinical decision, in understanding properly and in being able to interpret at the best the genetic information that is given by sequencing of *M. tuberculosis* genomes.

And this is a second step-- so education and training. A third step is how to link the information that is provided by genetic sequencing to the existing clinical decision systems, the hospital databases, or to the surveillance systems.

Because the information brought by the genomes, by genetic sequencing, is really huge. Not all the information is now needed for a clinician or for a public health officer to deal with. So we need to understand what is really needed, what is really informative, and to transfer in an appropriate and sustainable way.

It's very important, but sometimes it's not really straightforward to pass the message that including the genomic information into the care and control of the disease can really improve, and remarkably improve, the control of the same disease.

So this is, it's very important to understand that the novel technology will support the control of the disease in a manner that the public health and the clinical use will become personalised, and will help to reach the concept of precision when the right intervention is delivered at the right person or at the right population at the right time.