

## High-quality, cost-effective genomic testing and tumor profiling using NGS and OncoPrint assays

Hospital Italiano de Buenos Aires (HIBA), founded in 1853 in Argentina, is an institution committed to health care, teaching, and research. It has more than 750 inpatient beds, a university, and a research institute for translational medicine and biomedical engineering.

### Laboratory mission

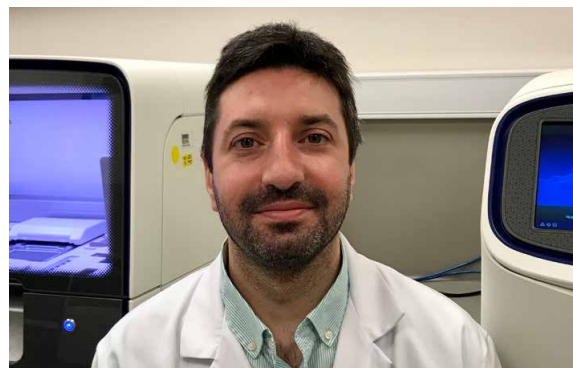
We are a nonprofit health care system with an integrated tertiary-care university hospital. We are devoted to providing safe and high-quality health care to our community. We endeavor to reach the highest levels of care, provide undergraduate and graduate training, and undertake basic, clinical, and population-based research.

### Laboratory vision

We aspire to be a hospital of excellence, a leader in the Argentine health care system integrated into the international scientific community.

### Values

- **Competence**—Professionalism at HIBA results from years of experience and competence in health care.
- **Transparency**—Information is shared clearly and effectively, respecting every individual's right to be informed.
- **Trust**—Trust-based relationships and high-quality medical services are basic tenets at HIBA.
- **Respect**—Relationships are built upon recognizing value in others and respecting individual identity and human dignity.
- **Integrity**—Ethics, rectitude, and honesty are the cornerstones of all our actions.
- **Commitment**—As a community-oriented institution, HIBA is dedicated to generating social value through health care, teaching, and research.



**Federico Jauk is a pathologist specializing in molecular pathology. Since 2019, he has been serving as the head of the Sequencing Laboratory at the Hospital Italiano de Buenos Aires in Argentina.**

Jauk has experience in cancer genomics and clinical biomarker testing, including techniques such as real-time PCR, fragment analysis, Sanger sequencing, and next-generation sequencing (NGS). Over the past few years, he along with his team have set up and analytically validated many genomic tests for solid tumors and hematologic malignancies.

### Introduction

- **What are the mission and goals of your lab at the Hospital Italiano?**

The Sequencing Lab at the Hospital Italiano is a core facility specializing in Sanger sequencing and NGS. It provides sequencing services to several departments within the hospital and performs its own in-lab tests.
- **How do you choose the type and scope of what you want to provide?**

The genomic tests we intend to provide are in high demand. As our resources are limited, tests should be adapted to the workflow and platforms we currently use. If possible, we choose supplies/kits of globally proven effectiveness, but sometimes we design, set up, and analytically validate tests based on cost-effectiveness.

- **What challenges are you trying to address?**

Latin America is a developing region, and the implementation of NGS techniques is difficult in that context. However, access to genomic tests has been improving over the past few years.

Here, supplies are more expensive than in the US or Europe, lab personnel are limited in number, and the test is not always reimbursed. So, it is harder to implement these tools and techniques.

The genomic testing market has many providers, both locally and abroad. However, they have varying quality and performance standards, which poses a challenge to the users.

We ourselves opt for local development whenever possible, following high-quality standards.

- **What made you look for help outside of your lab?**

Analytical validation (AV) of an NGS panel is a difficult and time-consuming task. We had the experience of validating the Ion Torrent™ Oncomine™ Focus Assay panel, which took several months to be launched.

There are some limitations at HIBA. Even as the demand for genomic tests is growing every day, our personnel are very busy and mostly focused on everyday tasks, with little time to analytically validate the latest large NGS panels. We are able to perform AVs on our own, but sometimes it is not as cost-effective as partnering with an AV team especially when you intend to analytically validate the panel within a short period of time.

- **What made you want to work with Thermo Fisher Scientific on this project?**

We have had the Ion Torrent™ PGM™ system for 6 years and the Ion GeneStudio™ S5 Plus System for 2 years now, and we are quite used to the workflow and to getting libraries sequenced relatively fast. We also have been using Ion AmpliSeq™ and Ion Torrent™ Oncomine™ panels for a couple of years with satisfactory results.

The AV of the Oncomine Focus Assay was a nice yet demanding experience, and its further implementation and results were good as well, so we wanted to try other Oncomine assays, such as the Ion Torrent™ Myeloid Research Assay and Oncomine™ Childhood Cancer Research Assay.

- **How was Thermo Fisher able to address any challenges?**

The Oncomine and Ion AmpliSeq panels are robust, end-to-end solutions that are easily adapted to our lab workflow, and they help us to set cost-effective approaches to NGS research.

- **What value did the AV team provide to your lab?**

The AV team has experienced professionals, and they are acquainted with regulations, local and international. They have a practical view that helps save time and money when analytically validating tests. The interaction between our group and the AV team was mutually beneficial—we could understand each other and focus on the AV process considering the shortcomings of the region.

### **NGS testing questions**

- **What was the driver, or what were you looking to accomplish or solve, when choosing a technology to provide a test?**

Since we are a large referral hospital, we try to choose the most cost-effective technologies.

- **Describe why you chose NGS technology for your lab.**

NGS technology provides comprehensive approaches to precision oncology research.

Turnaround time is a critical factor. With Oncomine assays, the turnaround time is relatively short, and the amount of nucleic acid required is quite low, sparing tissue or sample.

- **How were you getting your answers before bringing NGS technology into the lab (reference labs, platforms other than NGS, turnaround time delays, outsourcing, etc.)?**

We used single-gene testing by other techniques (e.g., qPCR, Sanger sequencing, fragment analysis); however, in some cases single-gene testing is not comprehensive enough.

Testing several genes in parallel using different techniques is generally more time-consuming than the NGS approach. Sequential single-gene testing shows delays in turnaround time compared with NGS; moreover, the sample can degrade over time.

- **Which new NGS tests have you brought on board?**

We started with the Ion Torrent™ OncoPrint™ *BRCA* Research Assay and a custom Ion AmpliSeq panel for *CFTR*, then used the analytically validated OncoPrint Focus Assay and OncoPrint Myeloid Assay. The OncoPrint Childhood Assay is under AV process right now. As you will notice, most panels are related to precision oncology research.

- **What roadblocks, barriers, and challenges did you encounter when bringing NGS testing capabilities in house?**

Our region is not doing well right now—it is vulnerable to external market conditions and uncertainty. There are supply issues, too.

- **What impact are you having with testing since bringing NGS precision oncology research assays on board?**

There has been an improvement in the way we provide genomic data that could lead to specific clinical research decisions. It has also become more affordable than outsourcing. Besides, we have achieved multidisciplinary teamwork, as well as generated local data.

- **What impact does bringing this test in-house have on the Hospital Italiano as a whole (economics, cost/revenue to hospital, higher reliable results, ease of use, tissue requirements, etc.)?**

Revenue has increased in a cost-effective way. As we already had the sequencers, there were no additional equipment expenses. This placed us in a good position in Argentina—we are now a bigger player in the field of NGS testing.

- **Please elaborate on the experience you had with your Thermo Fisher AV project manager. How did they help you through the AV process?**

They were highly supportive in every aspect, clarifying all that was needed. We had a very productive experience with them. The explanations and feedback they provided were very important to every part of the process.

- **What lab situation would ideally benefit from using the Thermo Fisher NGS platform and AV consulting services?**

It would benefit labs that want to analytically validate tests in a cost-effective and rapid way.

## Closing questions

- **How will you expand your testing portfolio in the future?**

We would like to expand with the OncoPrint Childhood Assay and Ion Torrent™ ReproSeq™ kits from Thermo Fisher, and a hereditary cancer panel from another supplier.

- **Do you expect to use the AV consulting service again? Who would you recommend to use the AV services in the future?**

We will probably use the service again, whenever possible. We would recommend it to other labs intending to offer high-quality and analytically validated NGS solutions.

- **Do you have any parting advice for other labs looking to bring NGS testing to their institutions?**

Performing NGS tests at a clinical research level is a difficult task. To do this in a sustainable way, it is important to be acquainted with the context you are working in, taking into account local regulations and quality assurance standards.

Find out more at [thermofisher.com/av](https://thermofisher.com/av)

**ThermoFisher**  
SCIENTIFIC