



Bringing Liquid Biopsy Home: Scaling Precision Oncology with AÍMA

Imagenome Laboratory (INOVIE Group) evaluates methylation-based circulating tumoral DNA detection via Nanopore sequencing, offering a high-performance, local alternative to centralised testing models.

"The results obtained with the AÍMA platform are extremely encouraging. The ability to detect circulating tumoral DNA on our own Oxford Nanopore sequencer, supported by such a fluid software workflow, opens real perspectives for the future of oncology diagnostics at the local level. This represents a significant step forward for laboratory autonomy. This approach is perfectly aligned with the way diagnostics are implemented across Europe and in many countries worldwide, where decentralized laboratory testing is the standard."

Dr Pierre-Jean Lamy, with Dr Gregory Egea

*Dr Pierre-Jean Lamy is Head of Imagenome Laboratory & Scientific Director INOVIE Group
Dr Grégory Egea is Medical Biologist at Imagenome Laboratory, INOVIE Group.*

The Challenge: Accessible ctDNA detection

In the field of liquid biopsy, identifying circulating tumour DNA (ctDNA) traditionally relies on detecting somatic mutations (Variant Allele Frequency). This approach often faces sensitivity limits when dealing with wild-type tumors or cancer subtypes without any known mutation. Sensitivity is also reduced when the tumour fraction in plasma is extremely low, as is frequently the case in early-stage disease or after treatment. In addition, the presence of clonal hematopoiesis (CHIP) can generate mutations originating from blood cells rather than the tumour, which may lead to false positives and often requires sequencing matched constitutional DNA to disambiguate the signal. Furthermore, many high-performance analysis solutions currently require shipping samples to the United States, which increases turnaround times and complicates logistics.

Dr Pierre-Jean Lamy and Dr Gregory Egea at Imagenome partnered with AÍMA to evaluate an alternative: a detection algorithm based on DNA methylation patterns that can be executed locally on their existing Oxford Nanopore Technologies equipment.

A Seamless Diagnostic Workflow

The pilot study involved the analysis of 11 samples—a mix of healthy controls and confirmed cancer cases. The laboratory utilized an ID-Solution extraction protocol, followed by a three-day sequencing run. The analysis was performed blinded for Aíma, meaning that the sample status (healthy vs. cancer) was not disclosed during the analysis phase.

- **User Experience:** The Aíma web interface allows for both individual and bulk uploads of BAM and BAI files generated throughout the sequencing process.
- **Efficiency:** The team noted the platform's ease of use, particularly for large datasets. "The bulk upload feature is highly efficient, taking approximately two hours to process three massive samples, which is very satisfactory for a clinical laboratory workflow."
- **Responsiveness:** The platform was described as clear and highly responsive, transforming complex raw data into actionable reports without requiring advanced in-house bioinformatics expertise. Reports are generated and available within minutes after the upload of the sequencing data is completed.

Biologically Consistent Results

The study demonstrated remarkable scientific robustness. The Aíma platform achieved **100% specificity** across the 5 healthy control samples and **100% sensitivity** on the 4 samples with known mutations (previously validated via a 50-gene panel on AGENA mass spectrometry).

A key highlight involved two samples where no mutations had been initially detected by standard methods:

1. One was correctly flagged by Aíma as cancer. This result was consistent with the clinical context, suggesting the algorithm could detect epigenetic signals even in the absence of detectable circulating tumour DNA mutations.
2. The other one was assessed as healthy by Aíma. As it happens, this case was tested several times without any mutation identified, therefore with suspicion that indeed no ctDNA was to be found in the sample.

"It is clear that this approach worked very well," the technical team noted, confirming that the results were in perfect alignment with the known clinical status of the patients.

Empowering Local Laboratories

The strategic advantage of the Aíma approach lies in its compatibility with *in situ* sequencing. Unlike externalised solutions, the data and analysis remain within the laboratory. This combination of speed, potential cost reduction, and increased precision through multiomic analysis (DNA fragmentation and native methylation) positions the technology as a transformative tool for modern diagnostics.

Key Performance Metrics:

- **100% Specificity** on control samples.
- **100% Sensitivity** on samples with known pathogenic variants.
- **Local Analysis** via Oxford Nanopore, eliminating the need for international sample export.