

Marcel Nelen, UMC Utrecht – Early Experience with Illumina TruPath™ Genome

Introduction

Marcel Nelen leads the Genome Diagnostics section at the University Medical Center Utrecht (UMC Utrecht) in the Netherlands, one of eight academic centers supporting rare disease diagnostics nationwide. His team recently became one of the first to evaluate Illumina's TruPath technology. In this interview, Marcel shares his experience, impressions, and what this technology could mean for clinical genomics. This interview summarizes the first impressions of the laboratory using this product for the first time. No result was delivered to patients or used in a clinical testing.



Marcel Nelen

University Medical Center Utrecht
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Interview

Marcel, could you introduce yourself and your work?

I'm the head of the Genome Diagnostics section at UMC Utrecht, part of the Department of Genetics. We handle a wide range of rare disease diagnostics and work closely with all academic centers in the Netherlands. Our mission is simple: give patients answers as quickly and reliably as possible.

Why did you choose to partner with Illumina to test TruPath?

I've worked with Illumina for many years on various innovation projects. When I took on this new role in Utrecht, we were discussing upcoming technologies, and TruPath came up. It looked innovative and promising, and we were interested in exploring what it could enable.

What has the collaboration been like so far?

It has been great to be among the first to try the technology. The results have been strong from the start. One area where it's already showing real potential is non invasive prenatal testing – an excellent fit for this type of phased data.

What aspects of TruPath feel the most novel?

The biggest innovations are the library prep directly on the flow cell, which simplifies the workflow significantly, and the ability to use native DNA for proximity based phasing. The long phased regions are extremely useful in clinical genetics.

What surprised you most when using the system?

How easy it was. A technician ran the first experiment, and the results were strong straight away. Removing the library prep step breaks down a major barrier.

Can you share a practical example from your evaluation?

One of the first samples we ran was a spinal muscular atrophy (SMA) family. We had previously spent more than six months analyzing this locus using long read technologies. With TruPath, the first run produced a clean, accurate result.

How does the workflow compare to standard short read and long read approaches?

Short read workflows require robotics and separate library prep; TruPath doesn't. Long read methods require DNA shearing and size optimization. TruPath uses native DNA, and the phased blocks are much larger – sometimes megabases. Having that level of phasing is something I haven't seen before.

What do you see as the main benefits of TruPath data?

Phasing is the standout benefit. With short reads, phasing is difficult, and we often need parental samples. TruPath significantly reduces that need. It also offers advantages over both PacBio and ONT in terms of phasing size and ease of workflow.

How would adopting TruPath impact your lab?

We could potentially retire several of our current workflows. We have 15–20 different workflows today, many of them requiring specialized skills or older technologies. TruPath could consolidate many of these into a simpler, more generic workflow. And because it runs on the same instruments and uses the DRAGEN environment, the transition would be relatively seamless.

What does your team think of the workflow?

They're enthusiastic. It's easy to use, requires no new equipment, and integrates naturally into the lab's existing setup. Early experiments for our non invasive prenatal diagnostics project have been very encouraging.

How could TruPath affect turnaround time?

Removing library prep saves at least a day. In routine whole genome testing that's helpful, but in neonatal intensive care (NICU) settings—where our turnaround time is about seven days – saving a day can have meaningful clinical and economic impact.

What problems could TruPath help you solve that are challenging today?

Reducing or eliminating the need for parental samples in recessive disease cases, improving structural variant detection, and enabling more reliable phasing – especially for applications like NIPT. We currently run separate assays, like inversion tests for genes such as Factor VIII; TruPath could replace some of these.

How do you envision the technology impacting the future of your lab?

It allows us to move toward a "one test fits most" model. That reduces training burden, cost, and workflow complexity. It also shortens the diagnostic odyssey for many patients and families.

What motivates your focus on innovation?

The goal is always to provide answers faster and more completely. Rare disease diagnostics can take years. If new technology brings us closer to speeding that up – or making it more accessible – we want to test it.

How do you see your work contributing beyond the Netherlands?

For technologies like NIPT, once we validate the workflow, we plan to publish and make our tools available so others can adopt them. Sharing knowledge with the global community is important to us.

What changes have you noticed in working with Illumina over the years?

Illumina has become much more open – sharing early access to technologies, collaborating across teams, and supporting clinical labs more closely. And the instruments and sequencing quality remain very reliable.

Learn more

[Illumina TruPath Genome](#)

[Illumina NovaSeq™ X Series Innovation Roadmap](#)