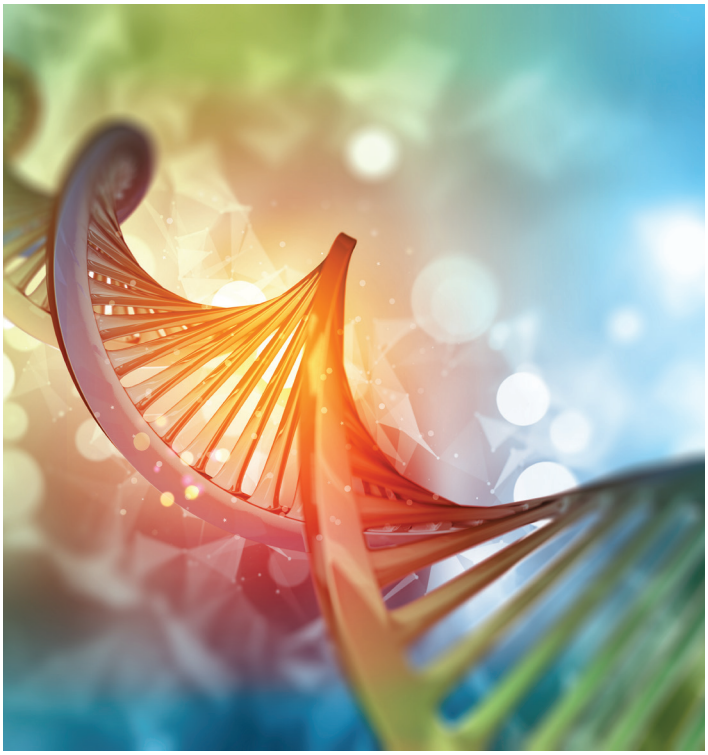
# Customer spotlight

## IVF and genomics: positioning ourselves for the future

### Introduction

Sequence46 is a Los Angeles–based laboratory dedicated to offering fertility clinicians access to the most advanced technologies for reproductive health, as well as services for clinical and research testing. This requires transparency into the process of preimplantation genetic testing, including its challenges and nuances.

We interviewed Catherine Welch, founder and managing partner of Sequence46, for her unique perspective in a start-up laboratory setting, to learn more about how *in vitro* fertilization (IVF) and genomic technologies are working together to advance the fertility field.



**Catherine Welch, MBA, TS (ABB)**

**Thermo Fisher Scientific:** Please tell us about your laboratory and what you are trying to achieve.

**Catherine Welch:** Our lab, Sequence46, offers preimplantation genetic testing (PGT) services, both PGT-A (aneuploidy) and PGT-SR (structural rearrangements). Our goal is to deliver the best genetic testing and, perhaps more importantly, offer the highest level of service and support so that our customers truly understand the benefits of genetic testing and the genomic technologies we utilize.

Reporting for PGT is challenging and confusing, with different ways to call aneuploidy or mosaicism. We want our customers to understand the basis of what we do and how we do it. One of the most important and impactful things we've done is change the way we report. We don't report aneuploidy or euploidy. We report "abnormal cells detected" or "no abnormal cells detected," in an effort to change the mindset of our customers and their understanding of the analysis. What we're trying to achieve is to describe exactly what's in the sample that we receive, rather than the embryo as a whole. Hopefully we help simplify the idea behind genetic testing.

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**Thermo Fisher:** Previously, you used Illumina™ VeriSeq™ technology, but when you set up your new lab you chose the Ion ReproSeq™ PGS solution. Can you tell us why?

**Welch:** We came across Thermo Fisher Scientific and Ion Torrent™ NGS technology toward the end of our search, and we're really glad we did. The service and research collaborations have been amazing. Support for the hardware and equipment is also excellent. We chose Thermo Fisher because we knew the platform would meet our expectations; when we do this type of testing, something is bound to go wrong. If and when it does, we need to have support available to remedy the situation as quickly as possible so we can get the most accurate results out to our end users. That has been key, and it's been great for us. The Thermo Fisher support team is easy to reach and understands the urgency of us needing to get in touch with them right away.

We found the Ion ReproSeq PGS solution was superior in many of the areas we felt were important in a next-generation sequencing platform. We chose Ion Torrent technology for a whole host of reasons, including service, support, research collaborations, and advances in the hardware itself. Software developments are also important and, of course, price. Across the board, we were happy with all that Thermo Fisher had to offer.

> "Automation greatly reduces the potential for error and helps reduce our liability."

We love the Ion Chef™ System and the Ion GeneStudio™ S5 sequencer because they make things easy and simple. The Ion Chef System simplifies the workflow by providing automated template prep. You put everything in and then at the push of a button it begins the prep. It gives us peace of mind knowing that when we put something in, what comes out is going to be accurate. Automation greatly reduces the potential for error and helps reduce our liability.

This also ties back to what I mentioned before about our reporting: it's a bit different than what is typically seen in the industry. We use Ion Reporter™ Software, and we essentially copy and paste into our report rather than retyping a result, so we don't have transcription errors. We feel it's critically important to show customers exactly what the software is reporting, rather than interpreting it in a way that may be more confusing.

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Another aspect that was important to us is the capability of running 96 samples on the Ion 530™ Chip. You can run 96 different samples, and for this protocol, there are 96 different indices, 96 different barcodes. This definitely gives us peace of mind that every single embryo is individualized and there is no chance of mix-up. What comes out is what we put in: the sample ID barcoding keeps all the samples straight. This was definitely another reason why we chose Thermo Fisher. Liability in what we do is a major factor, so we appreciate extra details such as this to add a layer of protection in the workflow.

From the perspective of a start-up lab, we strongly believe that PGT-A is at a point where it's a standardized product, and if we can run the kit based on the manufacturer's specifications without making modifications we would see excellent results. This was the best avenue for us to take and we've been able to accomplish this with the Ion ReproSeq PGS kit protocol: it works well right off the shelf.

We've also seen the commitment of Thermo Fisher to IVF research grow through the expansion of their reproductive genetic health team. That commitment is important, and we really appreciate it because it makes us feel like what we're doing is important and making a difference in the greater genomics field.

**Thermo Fisher:** What research work are you most excited about and why?

**Welch:** We're investigating the potential to do both PGT-M (monogenic) and PGT-A on a single platform, and we're thrilled about that opportunity. We're in regular communication with the Thermo Fisher team regarding progress and what that would look like on our end. I know that many of our partners are anxiously awaiting another alternative to the solution that's currently out there in the field. It's important for them to streamline their operations, and in this way they would be able to send [samples] to one lab. Knowing that PGT-M and PGT-A have traditionally been run on separate assays, we're excited about the possibility to run them on a single platform that may provide better results and a more streamlined process here in our lab. That's definitely something we're looking forward to.

Right now, research is extremely important for us and what we've done with the Thermo Fisher team has been great. All of our partners really enjoy and are excited about what is to come in our field, so that has been fantastic.

> "We're excited about the potential to run them [PGT-M and PGT-A] on a single platform that may provide better results and a more streamlined process here in our lab."

**Thermo Fisher:** What are the major trends that you monitor, and how do you position your lab for the future?

**Welch:** Our goal at Sequence46 is to shift the mindset of PGT-A testing in general. At industry meetings, we're still debating whether or not PGT-A is useful. I don't believe that's the conversation we should be having. In certain situations, it's very clear (and research supports) that PGT-A helps achieve a successful pregnancy, and in others it helps reduce time to pregnancy.

There are so many benefits of using PGT-A. I think what's more important is to really understand and agree on best practices, so we can offer this more in the future. In order to do that, we as reference labs need to collaborate with the rest of the industry in educating them on how we make calls on certain samples and lay all the information out there, so we become an extra tool in the toolbox.

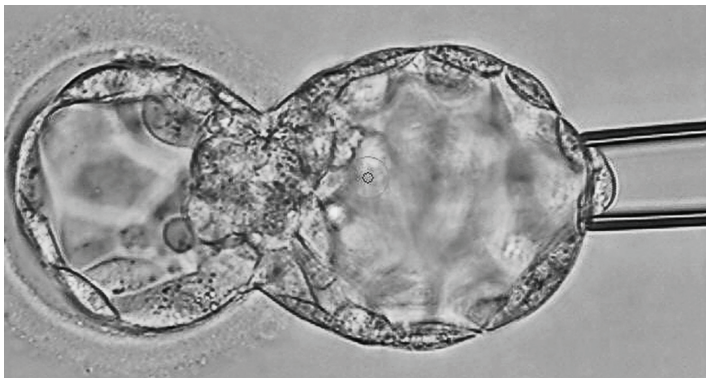

**Photo courtesy of ORM Fertility.**

As a research tool, if it's possible to evaluate embryos based on the biopsy results, embryologists would certainly select the one with the lowest amount of aneuploid cells. Prioritizing embryos with "no abnormal cells detected" over those with some percentage detected remains important. However, knowing that mosaic embryos can make healthy babies, those remain in the ranking and offer a chance at success. The verbiage takes the mystery out of results, hopefully allowing a greater understanding of what it is we are looking at. Having all the information is key.

In the field, one of the next big trends is noninvasive chromosome screening (to screen embryos through the culture media that the embryo grows in as it becomes a blastocyst). One of our projects right now has been looking at that media in collaboration with the Thermo Fisher research team.

We've had some very promising results, and the basic science is what keeps us enthusiastic about what we do every day. We get in the grind of running so many samples per day; it's fun to have something exciting on the horizon for the industry to possibly offer this noninvasive technology in the future. With these advances, IVF clinics that might not otherwise have the ability to biopsy would be able to use this technology and offer PGT-A to their patients in the future. This may ultimately result in a greater population of patients having success. At least that's what we're hoping for.

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**Thermo Fisher:** What advice do you have for other labs who want to stay relevant and embrace new technologies in the IVF space?

**Welch:** Collaboration is huge; our lab is a bit different in that we're not necessarily trying to get everybody to use us. We're looking for partners who are committed to offering PGT-A technology in a certain way; reporting mosaicism, for example. That's not for everybody, and we understand that. Does that mean we don't want to work with you, see what you're doing, and how maybe that will help us to improve? Absolutely not.

> "Look for partners whose vision really aligns with what you're trying to do (for us, that was Thermo Fisher), and the rest will come because you'll be offering not just innovative science, but excellent service."

We have a commitment to science, and ultimately to clinicians and patients. Across the board, our team will show you they are passionate about what they do. We understand the entire process, and are not just techs pushing buttons or pipetting samples. We care about getting the results out accurately and on time, and making our services affordable. We're dedicated to the big picture of moving IVF forward with research; we're not just another service lab in the field.

My advice to other labs would be to work together. Finding partners who are committed to the same initiatives and goals is extremely important to us. For laboratories, whether they're IVF laboratories or reference laboratories, I would say the exact same thing. Look for partners whose vision really aligns with what you're trying to do (for us, it was Thermo Fisher), and the rest will come because you'll be offering not just innovative science, but excellent service.

### Ordering information

| Product | Quantity | Cat. No. |
|---|---|---|
| Ion ReproSeq PGS Kit with Ion 510 Chips | 16 samples/run | A34899 |
| Ion ReproSeq PGS Kit with Ion 520 Chips | 24 samples/run | A34900 |
| Ion ReproSeq PGS Kit with Ion 530 Chips | 96 samples/run | A34901 |
| Ion ReproSeq PGS Training (automated or manual) | 2 or 2.5 days | A31276 |
| Priority Tech Support, Ion ReproSeq PGS Kit* | — | ZGLPSCIONS5 |
| Ion GeneStudio S5 System | 1 system | A38194 |
| Ion Chef System | 1 system | 4484177 |

* Priority tech support is only available in Europe and North America at this time.

Find out more at **thermofisher.com/ionreproseq**

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