

## The Reluctant Consultant

### (My Story)

**January 2025**

#### **Anniversaries trigger reflection!**

Two years ago, I left my full-time job to re-set. As many in this situation, I started an LLC and became an independent consultant to pay the bills while figuring out next steps. I did not plan on working like this for long, but life had a different plan, and I am now reflecting on the incredible kaleidoscope of experiences I've gathered.

By now a “clinical genomics veteran”, I’ve had the privilege of helping an enormous array of entities in this space, from baby startups incubating their vision to large health systems establishing in house testing and pharmaceutical companies incorporating genomics into their clinical trials roadmap.

Being exposed to such vastly different environments in a short amount of time helped distill some of the central mechanisms that underlie much of the friction that eventually creeps into all organizations (and might I say: sometimes shockingly early in the lifecycle of an entity!). I’m concluding that these dynamics are difficult to change in our insanely complex work environments, but it sure is helpful to be aware, and strive towards the ideal to the degree possible!

- Scope and resources are rarely aligned
- Responsibility is given but matching authority is not
- Foundation building is prematurely abandoned to invest into rapid growth
- Strategic roadmaps and implementation reality are misaligned

What to make of all this? Many enthusiastic entrepreneurs continue to try but few succeed beyond a few years. Academic medical centers struggle to keep genetic testing services in house. Existing companies consolidate. But... medical genomics is still a very young discipline! A perhaps fitting historical example for this type of evolution is cathedral architecture... Many early attempts to build bigger ones failed: working conditions were dangerous, roofs collapsed because they were not supported by the foundation. Building sites were abandoned due to lack of funding, forcing architects and stone masons to pack their tools and look for new building sites. Until visionary architects came up with new methods and innovations that moved that field forward.

For our industry this translates to “Many avenues need to be explored to find sustainable business models”. As frustrating as it often is to be part this evolution, it is vital to keep trying and collectively push us closer to the point where our beautiful field can finally become mainstream!

**March 2024:** Much has been posted about the challenges and consolidation of the clinical genomic testing industry. A wise person in my network summed it up like this: “We are exiting a decade of excessive spending and growth at all costs; laboratories now need to slim down and focus on their core strengths to survive”. The

anniversary of my decision to leave a well-paid job in this environment causes me to reflect on the past and look ahead.

I started 2023 exploring two exciting leadership opportunities on a part-time contract basis. Sadly, both vanished 9 months later for (different) reasons that tie deeply into the systemic issues the healthcare industry has been grappling with. And just like that I became a full-time consultant, constantly on the look for new work!

It is tempting to focus on the disillusionment that is beginning to permeate our field – but the need to make a living as a consultant has also provided me with an unexpectedly positive perspective. Much of my recent work has focused on advising startups that are energized by the precipitous drop in the cost of clinical genome sequencing and push ahead with innovation and creativity. As someone who trained in clinical molecular genetics two decades ago when the community was electrified by the promise of personalized medicine, I have seen the excitement grow stale under chronic resource constraints and lack of reimbursement. It is incredibly rewarding to see that the vision of genomic medicine is still alive! Onwards and upwards – the time is right to learn from the past and explore new approaches.

**February 2023:** Two decades ago I decided to jump... After a very productive postdoc in Human Genetics and several successful years as Junior Faculty at Harvard Medical School I was well on track to start my own research lab. Except it just didn't feel right. Like so many who enter a career in healthcare I was drawn to use my skills to help patients and I was lucky to be awarded a unique opportunity to become a fellow at Harvard Medical School's Training Program for Clinical Molecular Genetics - one of the rare paths for PhDs to become directly involved in clinical care. I took the Board Exam in 2007 and have been practicing this beautiful discipline ever since.

The early 2000s were inspiring - the human genome sequence had just been completed, GWAS studies began to shed light on common, complex disease, and molecular genetic testing was believed to be a firm component of clinical care in the near term future. About a decade ago the advent of NGS removed many barriers, creating tremendous excitement in the medical community. I was the proverbial person at the right place and the right time... As an early adopter of NGS I had the privilege to help define standards and guide our community. I spent many years at Harvard Medical School, then some time on the commercial side of Molecular Diagnostics. Fast forward to 2023. Our field is on shaky grounds - on "both sides of the isle". Commercial players are struggling and consolidating, health systems are scrutinizing finances, and the undeniable promise of genomic medicine continues to be met with obstacles.

After years in academia and then the commercial sector I decided to jump again... I left my full time job at the beginning of 2023 to pause and reflect on how I can best dedicate my experience to advance implementing genetic testing for everyone and move the needle just a little bit towards using genomic information proactively to manage and sometimes treat disease. I was fortunate to have a professional network that provided consulting opportunities to make this step less scary.

I continue to be passionate about the field of genomic medicine but like so many I am struggling with current reality. I never thought that I'd be someone who just "rips the bandaid off" - but here I am. I am committed to dedicate my nearly two decades of experience in cutting edge clinical genomic testing to where it can make an impact. Exactly where is unclear...The comfort of a full time position is a lure for sure - but until such an opportunity comes my way it seems I've entered the army of independent consultants!