Katherine Stueland â?? CEO, GeneDx

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Katherine Stueland, CEO of GeneDx, describes transforming the 26-year-old NIH spinout into a scalable, patient-focused business. Leveraging the Infinity database, strategic biopharma partnerships, AI-driven interpretation, and cost-efficient exome and genome testing, GeneDx accelerates rare disease diagnosis. The company is aiming to assist in the expansion of newborn screening programs as well as shaping global precision medicine while supporting long-term therapeutic development.

What organisation did you inherit when joining as CEO in 2021, and what strategic levers did you identify to transform GeneDx into a scalable growth company?

Stepping into a 26-year-old organisation spun out of the NIH, there were no surprises in terms of incredible science, really strong technology, and an academic mind-set. When I joined GeneDx five years ago, I already knew the company well because we had tried to compete with them at my prior company. We had launched an exome product, and every geneticist with whom we had strong relationships told us the same thing: do not bother trying to compete with GeneDx. You will never catch up. Their accuracy stems from all the data and richness of their experience.

When I stepped in, I found exactly what their reputation promised. There was, and still is, an incredible scientific rigour and integrity, deep commitment to helping patients, dedication to R&D and new discoveries. What was missing was the pathway to scale. How do we grow this business sustainably? The other missing piece was driving utilisation from multi-gene panels to exomes and

genomes.

That became my mission. Over the past five years, we have brought down cost of goods and turnaround times to make exomes and genomes price-competitive with multi-gene panels. The journey has focused on diagnosing as many families as early as possible using our industry-leading technology while continuously reducing costs and improving accessibility.

You have described a â??pay it forwardâ?• model in rare disease. How does this philosophy translate into a scalable business strategy that aligns patient impact with long-term profitability?

The rare disease ecosystem is unlike any other, with a remarkable intersection of families, researchers, biopharma companies, and patient advocacy groups. I have worked on the therapeutic side, including two FDA approvals, and collaborated with many patient advocacy groups. A consistent theme is always how do we diagnose patients earlier? That is what GeneDx does exceptionally well. Our data strategy makes us better than anyone at diagnosing patients with rare disease.

Our internal strategy mirrors the external ecosystem. You have all these stakeholders wanting to help one another. When a family receives a new diagnosis, an incredible support system immediately mobilises to connect them with researchers, help them fundraise, and guide next steps. There is an incredible community of parent advocates trying to help their own children whilst supporting other families. As a company, we want to embody that pay it forward approach.

Our secret sauce is our database, Infinity. We have tested more patients with rare diseases than anyone in the world, having tested over 2.5 million patients, run one million exomes and genomes, and generated more than eight million phenotypic data points. The combined genotypic and phenotypic data in Infinity allows us to more rapidly upgrade or downgrade variants of unknown significance. Every time we test a new patient, we learn more and enrich Infinity, which helps the next patient. Each patient informs the next, which is the pay it forward strategy in action.

Testing more also reveals higher disease prevalence than previously thought. In some cases, we are finding three, four, or five times more patients than expected. This creates a virtuous cycle where more data gives investors better reasons to fund the companies supporting rare disease, which ultimately helps more patients.

Balancing patient-centred mission with healthy business fundamentals is critical, and we have seen companies fail to strike that balance. We view running a healthy business as essential to continue reinvesting so we can help more patients.

Today we focus on segments with high unmet need. We have concentrated predominantly on pediatrics, where good guidelines and reimbursement pathways exist. Our tests are reimbursed about 55 percent of the time, which means we are constantly fighting denials on behalf of patients and our company. We want to reach 70 to 80 percent.

As new therapeutics emerge for different patient populations like cardiovascular genetic conditions for example, we generate data to bring to payers in partnership with biopharma companies. Once good reimbursement exists, we can focus on diagnosing adults with adult-onset conditions. We balance unmet needs, clinical guidelines, and reimbursement to keep our business healthy whilst expanding into new markets.

In a landscape that includes major sequencing players, what fundamentally differentiates GeneDx?

The major sequencing companies are doing an excellent job advancing the field, particularly in driving down the cost of generating raw genomic data. That progress has been critical for the industry. Where GeneDx differentiates itself is not in producing the sequence alone, but in interpreting it at scale. Generating a genome is one step. Accurately and consistently extracting clinically meaningful insight from that volume of data in a way that informs patient care is the more complex challenge.

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How do you see the progress of transitioning from traditional screening to genomic newborn screening at scale within the US healthcare system?

Newborn screening in the US is in a moment of transition and is advancing faster than I expected. For years, parents could access only limited screening through heel prick testing, which screens for very few conditions. This began to change with the Human Genome Project, which its real promise was to ensure that everyone could benefit from all the information residing within each of us. While there were good reasons for more limited techniques like heel prick screening, like the availability of technology and cost, we at GeneDx have set out to solve these challenges.

In 2024, we published initial results from the ground-breaking GUARDIAN study, the largest newborn screening research study in New York State which today has screened over 22,000 healthy babies using a whole genome sequencing. We examine more than 450 clinically actionable conditions and have found that 3.2 percent of these otherwise healthy newborns actually have a disease with clinically actionable treatment available. 92 percent of these conditions would not have been discovered using traditional limited testing. Without our Guardian study screening, the average age of diagnosis for these diseases would have been seven to eleven years. We are now eliminating about a decade of unnecessary disease progression and healthcare system costs.

This data is helping build an important case for expanding newborn genomic screening to more states. For example, in Florida the Sunshine Genetics Act which established a genetic sequencing pilot program for newborns is now in place because of the data generated through the GUARDIAN study. Another initiative, BEACONS (Building Evidence and Collaboration for GenOmics in Nationwide Newborn Screening), is the first NIH-funded national research partnership with state newborn screening programs and is now a multi-state study. GeneDx will provide sequencing and interpretation for the BEACONS program which is exploring the feasibility of genomic newborn screening on a national scale. We recently announced seven participating states examining more than 700 genes.

As you can see there is real momentum here, and what we want to see is more states adopting GUARDIAN-like protocols. Given this administrationâ??s mandate to support prevention, genomic newborn screening perfectly demonstrates how we can prevent symptom manifestation and unnecessary disease progression. The US system should ramp up its genetic screening capabilities as we have the technology and a solid protocol as a starting point. Our aim as GeneDx is to be a driving force alongside the many incredible researchers sharing our commitment to moving the needle.

From a reimbursement perspective, what are the primary economic case points for expanding access to genetics testing?

Approximately 80 percent of Americans have access to genomic testing today through commercial insurers and state Medicaid programmes, which is encouraging.

Payers reimburse testing because they recognise the clinical value and are persuaded by the health economic case. Without an accurate diagnosis, they are still bearing the cost of disease through a greater number of hospitalisations, unnecessary procedures, and treatments that may not address the underlying condition. In many cases, they are paying for years of symptoms without clarity. In that context, they are already absorbing the financial burden of not having a diagnosis. Deploying our testing earlier in the patient journey reduces downstream costs. The economic argument has therefore been compelling.

The fact that we are often only recuperating 70 to 80 percent of costs reflects structural realities within the diagnostics sector. We process every test that enters our system, and therefore have to complete prior authorisations, comply fully with payer contracts, and frequently engage in appeals processes that require additional clinical documentation. While that administrative friction is inherent to the industry, we have continued to lower our cost base and reduce pricing for whole exome and whole genome testing. Payers have responded positively to that discipline. As we bring down costs while demonstrating improved outcomes, the value proposition strengthens for both patients and insurers.

What is the strategic rationale behind your partnership with Komodo Health, and how does it extend GeneDxâ??s role from diagnosis to influencing long-term patient outcomes and therapeutic development?

When we consider the journey of a rare disease patient, our core commitment at GeneDx is to diagnose as many individuals as early as possible. An accurate diagnosis, however, is only the starting point. From there, we want to ensure patients and clinicians understand the full range of options available to them, whether that involves enrolment in a clinical trial, access to an FDA-approved therapy, or connection to a patient or parent advocacy group. Diagnosis should lead to a clear, actionable care pathway.

To support that objective, we are building a broad network of biopharma partners. The more aligned partners we have, the more options we can present to each patient and clinician as they determine the most appropriate next step.

Our partnership with Komodo Health strengthens this effort by adding longitudinal, real-world data to our genomic insights. It allows us to examine patient cohorts at scale and understand what individuals have experienced prior to diagnosis across hospitalisations, interventions, unsuccessful treatments, or participation in clinical trials. In many cases, patients endure a multi-year diagnostic odyssey, and that journey carries both clinical and economic consequences.

By integrating Komodoâ??s data, we can step back and contextualise the full patient journey, independent of any specific therapeutic intervention. This enables us to help biopharma companies better understand disease burden, identify the optimal timing for therapeutic intervention, and design more informed clinical trials. It also supports the development of stronger evidence packages that can be presented to regulators such as the FDA by grounding therapeutic development in real-world

patient experience.

Regarding international capabilities, we are just beginning to focus on expanding our reach. For any Ministries of Health or regions buying sequencers from other diagnostic players, we want to put our database to work for patients worldwide. We now provide interpretation as a service. Any region buying sequencers can tap into GeneDxâ??s Infinity database for interpretation. Whether newborn screening or population-scale screening, we are eager to put our data to work for more patients globally.

Do you envision GeneDx evolving into a more formal R&D partner to biopharma companies?

Today, we are collaborating with dozens of biopharma companies. We connect patients to them for clinical trials and provide deeper insights from the genotypic and phenotypic data contained in our Infinity database. We also run targeted testing programmes with some of these companies. For example, Jaguar Gene Therapy has a treatment for autism that we support through an autism testing programme.

Strategically, we work with multiple pharmaceutical companies to explore various ways of connecting them with patients and delivering valuable insights from our data, whether to support investment decisions or regulatory submissions.

How has the acquisition of Fabric Genomics strengthened your artificial intelligence capabilities, and how do you see artificial intelligence reshaping diagnostic interpretation and precision medicine more broadly?

The Fabric acquisition has been an awesome addition to the way that we are thinking about helping more patients. Fabric gives us the ability to provide interpretation as a service and position Infinity to help more patients in a decentralised way. It taps into our central intelligence in Infinity for any health system, in US or beyond, that wants to buy their own sequencers.

The vast majority of the US market today relies on a central lab, and Fabric is helping us reach more patients in a decentralised setting. We are able to executive on testing better, faster, and cheaper than any other model in the US. But in areas where labs might want to do their own sequencing, we are able to now put our data on top of existing systems through Fabric. That has been incredible.

More broadly, we view artificial intelligence across three layers. First, operationally, where it supports functions ranging from revenue cycle management to regulatory reporting. Second being within interpretation itself. There is a limited number of trained geneticists globally, and we must scale their expertise. Tools such as MultiScore use artificial intelligence to interrogate our database and prioritise the variants most likely to be disease-causing. Rather than manually reviewing vast datasets, our experts are presented with the most relevant signals, improving speed, consistency, and cost efficiency.

The third layer is therapeutic enablement. A vast majority of rare disease patients still do not have an FDA-approved therapy. Our objective is to expand and enrich our dataset and collaborate with biopharma partners to deploy artificial intelligence in target discovery and clinical development. The long-term ambition is to invert that statistic over time and move toward a future in which the majority of patients have a defined therapeutic option.

We will continue to deepen our data assets, broaden our biopharma partnerships, and apply artificial intelligence not only to improve diagnosis but to advance the broader promise of precision medicine.

In a funding environment increasingly focused on revenue discipline and profitability, what lessons can emerging companies draw from GeneDxâ??s approach to execution and sustainable growth?

I joined the company at a moment when capital markets were contracting sharply, particularly in diagnostics. In hindsight, that environment served us well as it compelled us to be clear about what we are distinctly good at.

There was a period when many diagnostics companies attempted to be all things to all patients as a one-stop shop across major areas like oncology, reproductive health, rare disease, pharmacogenetics. The market correction forced us to take a more disciplined approach and find out where do we truly have differentiation, and where can we build a durable, profitable business?

Profitability became a priority, not as an end in itself, but as a foundation for sustainability. The idea that a patient-centric business cannot also be profitable is outdated. In fact, profitability strengthens patient impact. It enables reinvestment, supports long-term infrastructure, and positions the organisation to expand from a place of strength.

For us, that meant starting in pediatrics, ensuring appropriate reimbursement, and building a profitable core. From there, we can extend into adult populations and, ultimately, broader newborn screening. By concentrating on areas where we can deliver the greatest clinical impact and demonstrate financial discipline, we are able to scale with confidence.

In my view, focus and discipline accelerate progress. Attempting to do everything at once often leads to a riskier model and a delayed path to profitability. By contrast, a methodical but decisive strategy allows you to move quickly and sustainably. Our 1,400 employees would tell you we operate really fast every day. The difference is that we move fast with clarity, rather than chasing breadth at the expense of long-term strength.

As we wrap up, do you have any final reflections or key messages you would like to share about GeneDx?

The rare disease space is incredibly special. It is a very tight-knit ecosystem that we are proud to be a part of. I know that there are moments where it feels like investors turn their attention to GLP-1s and to cancer and leave rare disease patients behind. But we are incredibly grateful to all of the parent advocates in particular, as well as our partners in biopharma, who have the resilience that we share.

Despite the evolution of focus from investors, it will continue to come back to rare disease. We are committed to making sure that it continues to be a priority in the US and globally. It is part of our responsibility to make sure the advances and improvements in cancer care, from diagnostics to cancer immunotherapy and healthy people living longer, gain the same momentum in rare diseases. We just have to continue to work together as one big cross-functional team to really change the game for more of these families.

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