



GenoSpace, LLC has Developed Technology that Solves the “Last Hundred Yards Problem” in Delivering Genomic Data and Provides a Needed Value-Added Service to Everyone in the New Genomic Health “Information Ecosystem”

**Healthcare
Genomics
(Private)**

GenoSpace

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**John Quackenbush
CEO**

BIO:

John Quackenbush received his PhD in 1990 in theoretical physics from UCLA working on string theory models. Following two years as a postdoctoral fellow in physics, Dr. Quackenbush applied for and received a Special Emphasis Research Career Award from the National Center for Human Genome Research to work on the Human Genome Project. He spent two years at the Salk Institute working on developing physical maps of human chromosome 11 and two years at Stanford University working

on new laboratory and computational strategies for sequencing the Human Genome. In 1997 he joined the faculty of The Institute for Genomic Research (TIGR) where his focus began to shift to post-genomic applications with an emphasis on microarray analysis. Using a combination of laboratory and computational approaches, Dr. Quackenbush and his group developed analytical methods based on integration of data across domains to learn biological meaning from high-dimensional data. Since joining the faculties of the Dana-Farber Cancer Institute and the Harvard School of Public Health in 2005, his work has increasingly focused on the analysis of human cancer and expanded to embrace systems-based approaches to understanding and modeling biological problems. In 2012 he launched GenoSpace to facilitate genomic data sharing and create an “information ecosystem” to enable twenty-first century personalized medicine.

Company Profile:

GenoSpace, LLC, is a Cambridge, Massachusetts-based company that is pioneering a bold and innovative software platform for advancing 21st-century genomic medicine. Founded in 2011 on the principle that individuals should own and control access to their own genomic data, GenoSpace has built a robust software system for securely storing vast amounts of genomic data and making it available to individuals and groups who need to connect with each other and share data.

GenoSpace’s “information ecosystem” consists of secure, reliable, user-friendly portals and computational tools that enable individuals, physi-

cians, health-care systems, research scientists, contract research organizations, and pharmaceutical companies to access genomic and other data and to build relationships with other stakeholders in mutually beneficial ways that can accelerate disease research and drive personalized medicine.

The age of genomic medicine has arrived, brought about by a million-fold drop in the cost of DNA sequencing and a thousand-fold increase in the speed of gene-sequencing instruments. As the cost of sequencing a human genome falls below \$1,000 and genomic sequence data becomes accessible to anyone, the challenge will lie in managing this valuable and highly personal asset. GenoSpace is poised to lead the coming information revolution as the leader and most trusted source for creating technologies that harness—and protect—the most powerful data on earth, our own. To learn more visit www.genospace.com.

**Interview conducted by:
Lynn Fosse, Senior Editor
CEOCFO Magazine**

CEOCFO: Dr. Quackenbush, with a relatively new company, would you give us a background on your vision and how it has developed so far?

Dr. Quackenbush: The interesting thing about working in the field of genomics during the past few years has been the incredible transition in our ability to generate genomic data. We are seeing an explosion of technological advancements that are dramatically reducing the cost and time it takes to sequence a human genome.

The first draft human genome sequence was announced complete in 2000, a project that took about twelve years, and cost upward of three billion dollars. Today, we can sequence a genome for about a thousand dollars in about a day.

This advancement has dramatically changed the way we think about using genomic data, as well as the way we think generally about health care, biomedical research, and the potential for personalized medicine. I always used to say that if I had cancer, I would mortgage my house and sequence my genome to try to find genetic variants that might be relevant for my treatment. The fact is, today I could pay for the sequence using only my credit card.

Data generation is rapidly becoming a commodity, with companies in a “race to the bottom” to drive down cost and further shorten the time required to sequence a genome. However, one interesting challenge remains, and that is how to effectively use the data. No one has yet solved the “last hundred yards” problem, which is how to make the information available and meaningful to the those who have a need to use it today. Health care professionals want to offer better treatments and direct their patients to appropriate therapies. Pharmaceutical companies are increasingly developing drugs that target specific genetic lesions. Contract research organizations are looking to recruit patients to trials that have inclusion criteria based on genetics. Disease foundations increasingly want to fund research to search for the genetic basis for disease and get patients involved in the process. Scientists want to use genomic and clinical data to uncover the mechanisms of disease. Patients themselves want to improve their health care and want to advance research by broadly sharing their own data.

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the various constituencies involved in health care. This allows us to solve the “last hundred yards problem” in delivering genomic data and provides a needed value-added service to everyone in the new genomic health “information ecosystem.” We have developed the technology required to allow GenoSpace to be the repository for people who want to securely store their data and yet share it broadly with others who need to use it today.

CEOFCFO: How has your concept developed so far?

Dr. Quackenbush: A few years ago, my partner, Mick Correll, and I, began looking at this problem and thinking about how people share data. In health care, everyone is concerned

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about safeguarding what is known as protected health information (PHI) that can be linked back to a patient. Consequently many types of data are often “de-identified” before it is shared, reducing its potential value. When we thought about de-identifying genomic data, we realized that everyone’s genome sequence is uniquely identifiable. If I gave your DNA to the FBI, they would have a “fingerprint” that is absolutely unique to you. Even less than one tenth of one percent of your DNA is enough to identify you; and so, in this case, the idea of de-identification flies out the window.

Mick and I understood that dealing with such private data, which we can now regularly generate, requires thinking differently about security. Moreover, the real value in having data lies in the ability to link it back to an individual, to recruit the individual

through a clinical trial, or direct the person to the appropriate personalized therapy. Once we realized this, we knew we needed a technology that was fundamentally focused on data security. Since genomic data volumes are so large, we built the core GenoSpace system using scalable Internet technologies. Basically, we rethought the entire process of data storage, data management, and data access, and then built a secure technology stack that would allow us to store, but still readily access, genomic data and other health information in an integrated fashion.

Around our core, we built a data exchange protocol and tools that allow people secure access to their data. The system solves the problem with

most data access today, which is that it is all or nothing. For example, if I wanted to find out whether a colon cancer patient had a mutation in a gene called KRAS, which might be therapeutically relevant, I don’t need to know their APOE gene mutation status, which is associated with their risk for developing early onset Alzheimer’s. Most of the tools that exist today cannot give you that granularity of access; yet we know that it will be essential. A physician

searching for genetic mutations that are relevant to treating his patients will not want their whole genome sequence but only information relevant to the disease. A pharmaceutical company conducting a trial will want to know how many patients have a particular genetic lesion, but will not need to know everything. By limiting access appropriately, we can make the proper relevant information available in a way that enlightens decisions without overwhelming users of the system.

In the past few months since publicly announcing our company, we have built a number of portals for the various constituencies that require access to data. Although we have built a system that our clients have found useable and useful, we know that there are more ways to add value to the

data. Earlier this week, we announced our first major partnership, with Thomson Reuters. We plan to take the information they have amassed on genomic variants, filter it so that it is relevant in different circumstances, and then redistribute it through our system to physicians, scientists, and others who need to use those data. I think we are in a good position right now to deliver tangible tools and useful information to people increasingly looking for access.

CEOCFO: What is happening with the business today? Are you looking to recruit more people to participate and are you perfecting ways to provide the information?

Dr. Quackenbush: If you look at companies like 23andMe, you will find that they are essentially attacking the retail genomic market. For example, 23andMe is trying to recruit a large number of normal healthy patients. Unfortunately, what you can say about a “naïve” genome — your genome, my genome, or that of other relatively healthy people — is not all that much. We can find out where our ancestors came from, but we probably already have a pretty good idea about that. We can determine whether we have sticky or dry earwax, but there is already a fingertip test for that. Anything truly of interest is relatively limited, and so the retail approach fails to provide much value to the end user. On the other hand, if you think about taking a wholesale approach and recruiting a large number of patients who have particular diseases, you can return some real value back to these people in the form of information that is relevant to their treatment. Moreover, if you build a large database of patients with a single disease— ten to thirty thousand patients, for example — then your ability to make important discoveries becomes exponentially greater than if you had twenty or thirty thousand healthy people. And you can also give patients information that they or their physicians can potentially use to improve care.

Aware of the potential for large-scale recruitment of patients, Mick and I decided to forge partnerships with disease foundations that were pro-

gressive in their approaches to doing research and involving patients. We just completed a pilot project with a major disease foundation that is producing genomic data on a large group of patients, but which needs a place to store the information. We solved their problem by providing them with a research portal and the tools to access the data in a way that allows them to move seamlessly from clinical data to genomic data. We beat out an established software company by creating an innovative, secure, and extremely user-friendly system. Within the next few weeks, we hope to formally announce our partnership with this foundation and describe in more detail how we have solved a fundamental problem for them.

We then plan to move on to the next phase of our work with the foundation, which entails creating a community portal for their patients. From the beginning, our vision has been to make patients the centerpiece of our infrastructure and ecosystem, and this vision meshes well with the foundation’s goals. Mick and I want to help our client make genomic data and personalized genomic medicine available to patients throughout the United States and worldwide. We want to enable everyday people to enter their data into the system so we can return information back to their physicians to help drive their therapies. We also want patients to connect with each other and to groups that are interested in working with them.

We are building toward that next phase by creating a physician report and portal that integrate genomic variants and clinical data for each patient with information from Thomson Reuters, to deliver a world-leading personalized medicine report. We are also building a network of service providers who can generate data, as well as putting the pieces in place to offer a complete set of tools and technologies to service a broad group of potential users. We are excited about accelerating the work we have already done in offering genomic data solutions.

CEOCFO: Are the communities that

should know about you aware and how do you get people to know you exist?

Dr. Quackenbush: We are increasing our word-of-mouth advertising, as well as seeking media opportunities, like this one, which I think will help raise awareness for our company. We were initially conservative about making ourselves widely known before it was time. Bringing a product to market is like catching a wave while surfing: If you start too early people will wonder what you are about and why you have not done anything yet. If you start too late, you will miss the opportunity to be the leader. This year, the time was right, especially since the cost of genome sequencing has taken a dramatic turn by passing the thousand-dollar mark and prompting people to think very differently about how sequencing data might be used.

Patients themselves will likely become motivated to have their genome sequenced for a wide variety of applications. Health care providers are moving toward personalized medicine, but their information systems are not designed to handle genomic data. We see a growing interest among disease foundations to use genomics both to advance their research mission and to engage patients.

Foundations have particular leverage today as research funding from the National Institutes of Health continues to decline. But they also represent patients and can use the power of numbers to help advance the adoption of genomic medicine and the sharing of data.

We also see the rise of personalized medicine as the perfect opportunity for us to expand awareness of GenoSpace. We issued a press release announcing our formation and then a second describing our relationship with Thomson Reuters. These have informed people that we have a viable and workable solution that we can offer today. Already a number of interested parties have had substantive meetings with us, including a network of oncology practices, major

contract research organizations, disease foundations, independent hospitals, and others.

The CIO of a company that does contract pathology work spoke to us about their plans for personalized medicine, and described a “hole” in their data management capabilities. Everything we told them about our technology convinced him that we have a square peg that is perfectly sized to fit into their square hole. For us, it validated our ability to identify the boundaries of a significant gap in providing access to genomic data and to build a technology that spanned those boundaries.

While we have taken a targeted approach at the beginning, we also recognize that, at some point, it will make sense to undertake a broader outreach effort. But when we do, it will reflect a strategy that designed to reach the right audience at the right time. There is no point in wasting money and effort yelling from the top of a mountain if nobody can hear you.

CEOCFO: Are you able to accommodate all the interest that seems to be coming your way; will you need to add to staff?

Dr. Quackenbush: We are definitely in a growth phase. We went from being basically two guys and a dog to the point where we have a rapidly growing team who are now doing most of the development and implementation, and we plan to ramp up fairly rapidly. One good thing about our business model is that because we use a cloud-based storage system we do not have major capital costs; our costs are really just personnel. In addition, throughout the years, Mick and I have built up a network of highly talented people who are very interested in joining us, which allows us to bring people onboard strategically; first on a part-time basis, then as full-time as our needs grow and as their individual talents become relevant for the work required.

CEOCFO: Are you aware of any competition to your idea?

Dr. Quackenbush: There are many people out there who are pretenders to the throne. I think everyone has come to recognize that genomic medicine is an important space. Many companies have tried to package, analyze, and add value to genomic data, but from everything I have seen, I think we have a unique approach and a unique solution. Companies like DNAnexus, Navigenics, and even 23andMe and even Oracle might see us as competitors; however, I think we are very strategic in how we developed our business model, how we think about our technology offering, and how we build our partnerships, and as result, we occupy a unique market position. There will always be competitors. At the end of the day, however, what we hope to be is not a “MySpace,” but a “Facebook” — a group with the right idea at the right time and right place and one which can really make a difference.

CEOCFO: Is GenoSpace funded to get through your next phases?

Dr. Quackenbush: We are doing quite well for a relatively young company. We are completing a Series A round through a network of high net-worth individuals. The funding we’ve secured can easily carry us forward, particularly since we are already realizing revenue through a number of projects. We are well positioned to build a strong, viable company. Like I said earlier, we have put together a very capital-efficient model for our business.

CEOCFO: Why should investors pay attention to GenoSpace today?

Dr. Quackenbush: Health care and health care costs are becoming one of the single greatest public and private sector expenses. There is an opportunity to make a big dent in those costs by personalizing medical care. More and more drugs are being developed that target specific genetic lesions. The single biggest cost in clinical trials and the single biggest risk factor is recruiting the appropriate patients with the right genetic backgrounds. GenoSpace sits at a nexus for a large number of market forces and we offer a unique solution that

bridges barriers between different groups. Our technology, which facilitates use of genomic data is at the heart of our financial model. While providing software can generate short-term revenue, in the long run, the real value lies in facilitating interactions and transactions. If you want to add data, connect to someone, or share data, there is a cost our customers because there is a real cost to us to make it happen. But we can make money on these interactions and we see an opportunity to also share in revenue with our partners. We think we have arrived at a win-win solution that is scalable and, given market forces, offers the opportunity for exponential growth.

CEOCFO: What should people remember most about GenoSpace?

Dr. Quackenbush: This has been an incredibly interesting ride. My partner, Mick Correll, and I have really enjoyed the process of building a company from the ground up. Like everyone else, we have made mistakes and have learned from them. Prior to starting GenoSpace, we worked together for more than at the Dana-Farber Cancer Institute to build software and database solutions for biomedical research. In that time, we learned a lot about managing health care and health care information were able to identify critical needs and shortcomings in the system. We have also learned a great deal about the various groups who want to use genomic data and their unique requirements. And we’ve developed good strategies for solving them.

For us, building a business has been fun, but it is also extraordinarily rewarding to be doing something we think has great potential to improve the quality of peoples’ lives and the quality of health care and medical care. In some ways, we see ourselves as social entrepreneurs in addition to financial entrepreneurs, and it has been an honor to work with an extremely talented group of people and partners.