As Use of Genomic Data Expands in Cancer Care, Patients Share Their Stories

Including patients and patient advocates in the research enterprise is not only valuable but also expected.

December 3, 2019 By National Cancer Institute

Last spring, at the end of his sophomore year of college, Jace Ward developed double vision in his left eye. He thought the problem might have been related to a recent car accident. But when doctors investigated, they found a tumor in his brain.

After the tumor was discovered, a biopsy showed that Jace had an almost uniformly fatal form of brain cancer called diffuse intrinsic pontine glioma (DIPG), which is usually found in children.

“This was my worst nightmare,” Jace said at a recent NCI-sponsored scientific workshop about personal genomic data, where he was a speaker.

Jace had been invited to the meeting to share the story of how he and his family used genomic information about his tumor to try to find a treatment that might extend his life—and the challenges they faced in the process.

A turning point in Jace’s story was his decision to allow the surgeons doing the biopsy to collect additional samples of his brain tumor for analysis with a genomic test.

The test revealed that Jace’s tumor had a genetic mutation linked to brain cancer, called H3 K27M. And, as Jace soon learned, NCI-supported researchers had been testing a new targeted therapy, ONC201, in patients whose tumors have this mutation.

Jace did not meet the criteria to enroll in one of the clinical trials of the investigational drug. He was, however, accepted into an expanded-access program that allowed him to receive the drug following treatment with radiation therapy.

As part of the trial, Jace takes five pills—a single treatment of ONC201—once a week. Feeling tired the next day has been the only side effect for Jace.

After 9 weeks of treatment, Jace was doing “exceptionally well,” according to his mother, Lisa.

Now a junior studying pre-law and business at Kansas State University, Jace has been invited to speak at a planned congressional hearing on DIPG and research funding next year.

“It was just lucky for Jace that he had the [H3 K27M] mutation—if you could say that a young man who develops DIPG at age 19 can be considered lucky,” said his mother.

Moving Toward Precision Medicine

Jace is one of an increasing number of patients with cancer who are receiving genomic tests. Doctors use the results of these tests to match individuals with the most appropriate treatments—an approach known as precision medicine.

But the expanding use of personal genomic data has raised questions, including: Which patients should undergo genomic testing? Who controls a person’s genomic data? What are the potential benefits and risks of sharing one’s genomic data with researchers or family members?

To explore such questions, NCI hosted the genomic data workshop, held on the NIH campus in Bethesda, Maryland, in September. The meeting included researchers and clinicians as well as patients with cancer, survivors of the disease, and patient advocates.

One aim of the meeting was to give patients a strong voice in the conversation about legal, ethical, and security issues related to personal genomic data, noted Sean Davis, MD, PhD, of NCI’s Center for Cancer Research, who helped to organize the workshop.

“We heard powerful stories from people who had received their own genomic data and then figured out how to share the results with researchers in the hope that this would improve their own care or benefit other people with the same disease,” he continued.

But other speakers, Davis went on, “conveyed a realistic view of the potential risks associated with sharing genomic data.”

The risks of sharing one’s genomic data include the possibility that a person’s identity and disease status could inadvertently be made public, information that might also affect family members, several presenters said.

They noted, however, that certain risks may be acceptable to patients with advanced cancers who are trying to stay alive by sharing their genomic data with clinicians and researchers.

“For me, the urgent question is: How can I avoid death?” Jace said. “And if there’s a chance I might lose some privacy in the process of avoiding death, then I should be able to decide to take that risk.”

His mother added, “The concern for us—and for the parents of most children with rare diseases—is that our children are dying.”

She went on, “Why should we be less worried about kids dying than we are about the potential

risks of sharing genomic data?”

Genomic Data and Clinical Care

One of the main messages from the workshop was that “patients want to be able to make decisions about their personal genomic data for themselves,” said Elizabeth Hsu, PhD, of NCI’s Center for Biomedical Informatics and Information Technology after the meeting.

“Ultimately, the patients are the owners of the data,” Hsu continued. “We in cancer research are merely the stewards of the data.”

The timing of the workshop, she noted, reflected the fact that genomic approaches long used in the laboratory to investigate the biology of cancer are increasingly playing a role in clinical care.

“Many patients right now are starting to feel that they have viable biological or genomic information that they can act on,” said Amanda Haddock, who is president of Dragon Master Foundation, a nonprofit that aims to accelerate cancer research, at the workshop.

It is not yet routine for patients with cancer to have their tumors sequenced, she noted, adding that she hoped this would happen soon.

For decades, most patients with DIPG and other tumors in particularly sensitive areas of the brain have not undergone biopsies, because doctors have thought that the potential risks outweigh the benefits, Haddock explained.

But as Jace’s story illustrates, clinical trials are testing new treatments aimed at specific molecular changes in tumors. And to be eligible for these trials, patients will need to have biopsies so that their tumors can be tested for certain changes.

“You can’t practice precision medicine if you don’t know what you’re treating,” Haddock said.

“DIPG Won’t Wait”

The median survival for patients with DIPG is less than a year. Jace said that when it came to making decisions about his treatment, he did not have the luxury of time.

“DIPG won’t wait,” he said. “It won’t wait to take my voice, and it won’t wait to take my life.”

The urgency in Jace’s voice “is something that I hear from patients with advanced cancers all the time,” said another speaker, Stacy Gray, M.D., of City of Hope, after the meeting.

For these patients, she continued, the time horizon for sharing data and getting meaningful results is “entirely different from how things have traditionally been done in clinical medicine and academic research.”

Jace had the additional challenge of being a young adult with cancer—someone who was not exactly a pediatric patient but also not quite an adult patient.

“It’s a hard road to navigate for a 19-year-old,” said his mother. “We often feel pretty lost.”

Jace also said that he felt “lost” at times, and his words seemed to resonate at the workshop.

“Jace Ward amazed, touched, and inspired every one of us in the audience,” another speaker, Adam Resnick, PhD, of the Children’s Hospital of Philadelphia, wrote on Twitter.

Educating Patients about Genomic Data

Over the course of the workshop, a vision of the future seemed to emerge in which patients could use their genomic data to try to enhance their own medical care while also contributing their data to cancer research in the ways they see fit.

Several patients, including Jace, expressed the hope that sharing their genomic data with researchers would benefit other people with the same disease, even if they themselves did not benefit.

There may not be a one-size-fits-all approach to how best to address issues related to the sharing of personal genomic data, noted John Wilbanks of Sage Bionetworks, a nonprofit research and technology development organization.

Janet Freeman-Daily, a patient with lung cancer, noted that there is likely to be a range of attitudes about data sharing among patients. She is a cofounder of the ROS1ders, a patient group that has partnered with researchers studying tumors driven by acquired alterations in the ROS1 gene.

“People with advanced cancers are often motivated to share their data with researchers to try to fuel discoveries,” Freeman-Daily said. But, she added, they should be aware of the possibility that sharing personal genetic information could have consequences for family members.

Cancers caused by genetic alterations acquired during one’s lifetime may have different implications for privacy than those caused by inherited genetic alterations, Freeman-Daily noted.

She and other participants at the workshop stressed the need to educate patients about how genomic information about cancer can be used in clinical decisions.

“Researchers need to engage with patients and to empower them so that they can make informed decisions about their genomic data,” said Leah Mechanic, PhD, of NCI’s Division of Cancer Control and Population Sciences, at the workshop.

The same message applies to the general public, she continued, noting that companies are selling DNA sequencing services directly to consumers.

“People need to understand what direct-to-consumer genomics companies do and do not offer, and the limitations of what they provide,” Mechanic said.

New Technologies Needed

Jace’s experience highlighted some practical challenges related to genomic data. To share his data with doctors at different hospitals, Jace had to pick up the computer disk containing his results and either send it in the mail himself or physically take it where it needed to go.

A pressing need for the research community is “to find ways to make it easier for genomic data to be shared, particularly when it comes to rare conditions, because scientists need to pull together data from many patients to gain insights into these conditions,” said Mechanic.

The workshop featured presentations by researchers who are developing technologies, or platforms, that would enable genomic data to be collected, stored, and shared securely by patients and investigators.

For example, Corrie Painter, PhD, of the Broad Institute described Count Me In, a nonprofit organization that partners with patients to advance cancer research.

The organization has collected tumor samples and medical information from about 8,000 patients with different types of cancer. After removing identifying information from the patient data, Count Me In makes the results available to researchers through open-access databases, such as those on cBioPortal.

In addition to collecting and sharing patient data, a goal of Count Me In is to learn from patients who participate in cancer research about ways to improve the experience. Researchers connect with patients through online groups, surveys, and one-on-one communication, Painter noted.

“Throughout the entire process, we work hand in hand with the patients who have the diseases we are trying to help,” said Painter, who is also a cancer survivor. “We have been learning as much from them as they have learned from us.”

As patients, she added, “we all want to feel that what we are contributing is meaningful and useful.”

Making Sense of Genomic Test Results

The meeting ended with a discussion of ways to think about developing a sharable genomic health record that an individual could control and leverage to his or her benefit. Part of this effort would involve studying the optimal ways for patients to receive their genomic information.

In Jace’s case, he received an automated email saying that his online medical information had been updated on the portal where he communicated with his doctors. After following a link to his genomic data, he felt “overwhelmed and alone.”

“I didn’t have the slightest clue what I was looking at,” he added.

To learn more about what the results of the genomic tests meant, Jace’s parents sought help from pediatric and adult brain tumor experts who were not part of Jace’s medical team, including one in Europe.

Once Jace and his family understood the genomic test results, they still needed help finding a clinical trial, because his health care team was not able to suggest possible options, Jace explained.

Help came from the mothers of other children with DIPG who were familiar with research on ONC201—which was funded in part by NCI’s Small Business Innovation Research program—and pointed Jace to possible clinical trials. The Wards had connected with these mothers through a group on Facebook.

Patients as Partners in Research

For decades, noted Painter, patients have essentially been “passive participants” in cancer research.

“But we’re at a turning point, with the research community embracing the value of what patients can bring to the table,” she continued, noting that patients can provide input on a range of questions relevant to their experiences, such as side effects and treatment regimens.

Davis agreed and said the workshop supported this idea of patients as experts. He was struck by stories about patients’ reliance on one another and on support systems that were outside of the medical team, such as online communities of patients.

In general, Davis noted, researchers need to develop stronger connections with patients and patient advocates. “Patients have an incredibly important perspective that we need to include at all stages of research, from the planning of studies to the delivery of results,” he said.

“Including patients and patient advocates in the research enterprise is not only valuable but also expected,” Davis added. “In 2019, they expect to be at the table talking about things that affect them.”

This post was originally published by the National Cancer Institute. It is republished with permission.

© 2026 Smart + Strong All Rights Reserved.