

Breaking the bell curve



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It was my first general radiology rotation. “Hey students!” I heard from across the room. “Come and look—we have a celebrity!” Justin Timberlake? I thought. Maybe someone in politics?

The other medical students and I hurried over to the attending station. There we saw a patient’s abdominal CT scan. “Li Fraumeni Syndrome” was written as the

imaging indication. Sounded somewhat familiar. I racked my brain...p53 mutation, SBLA—sarcoma, breast, leukemia, adrenal gland. I didn’t know anything else about the cancer syndrome, only having memorized the pertinent facts and mnemonic in order to answer any related questions on my boards.

The attending pointed to the patient’s chart. “Read this,” he said. The clinical history at first seemed benign. Thirty-nine-year-old female, not a name I recognized. As I continued to read, however, I discovered that she had her first cancer diagnosis—melanoma—at age 16, followed by thyroid cancer, then adenocarcinoma of the lung, later complicated by metastasis to the brain. Her CT scan, performed that day, revealed postoperative changes from a liver mass recently resected, stable metastatic lesions within the axial skeleton, an abdominal scar secondary to previously resected peritoneal metastases,

and bilateral breast implants, the only remaining telltale sign of breast cancer removed years ago. Wow, I thought, this patient has been through the wringer.

“You guys should go and talk to her,” said the attending. “second floor, oncology waiting room.” The other medical student and I looked at each other, then obediently left the reading room and headed in the direction of the Cancer Center.

As we walked, I started to become slightly nervous. I had absolutely no idea what to ask her. This patient’s body had been literally torn apart by one cancer after another. I didn’t want to offend her by seeming to pry into her personal life, and also didn’t want to be the brunt of any misguided anger. I steeled myself for the worst. I wouldn’t have blamed her for being completely disenchanted by the world of medicine, and to take it out on some unsuspecting medical students.

The patient’s oncologist, friendly and jovial, greeted us. He agreed to allow my fellow medical student and me a few minutes alone with the patient before her scheduled appointment.

With his unexpected blessing, we knocked on the patient’s door, paused, and entered. We were greeted by a young woman and her husband. Both, surprisingly, seemed the epitome of health. The patient was slim but not emaciated, dressed in colorful yoga gear—a flowy top and leggings. Her husband was similarly young, pleasant-looking, and well-dressed. We introduced ourselves as fourth-year medical students.

“We’re from radiology, and we were looking at your scans with our overseeing doctor,” the other medical student began. The couple’s faces fell, slightly, and their expressions became wary. “No no, not with any results of scans. We just saw that you were diagnosed with Li Fraumeni Syndrome, and the doctor suggested that we come up here to talk to you a little bit about your experience with the diagnosis.”

With this reassurance that we bore no bad news, the pair brightened. The patient, after looking thoughtfully at her husband, started to speak. Her voice was crisp and clear. She told us that after her initial diagnosis of Li

Fraumeni Syndrome, she was given only one year to live. With this unanticipated news, and at such a young age, she and her husband made the decision to explore places they had never been, places they had always dreamed of seeing. They booked a European cruise, went skiing for the first time in Beaver Creek, Colorado, adventured in Hawaii, and swam in the ocean off of Costa Rica.

The morning after they returned from Costa Rica, however, the patient woke up to her husband standing over her. She laughed—“Hi, I said.” “You just had a seizure,” he replied.

She was diagnosed with metastatic brain cancer. The couple remarked how miraculous it was that she was at home, in her own bed when she had the seizure. Not once did they complain about the actual diagnosis. They were just grateful at the timing of events. “She was completely normal the week before,” said her husband, “jumping off boats into the ocean.”

Despite this unnerving event, the patient’s “one year” prognosis ultimately turned into three years of healthy, vivacious life. “I just keep on going!” she said, happily. She had even declined chemotherapy. The oncologist noted that he did not blame her as “the chemotherapy would have killed her faster than the cancer.”

Still, as with every genetic disorder, the impact of Li Fraumeni Syndrome did not terminate with the patient. It was not until after she had given birth to her third child that she learned that her unusually early cancers were not random events, but were due to a mutation of the p53 gene. With further testing, she found out that both her second and third children—a daughter and son—had also inherited the gene.

The daughter passed away at five-years-old.

Up to this point in the conversation, the patient had discussed her diagnosis and its numerous assaults of her body casually, like they were something that had happened to an acquaintance instead of to her. Her story had unfolded slowly, purposefully, with intermittent well-received details supplied by her husband. But this disclosure was different. The reason was clear—she had



not yet forgiven herself, as it was her own genetic material that led to her daughter's death.

At that moment, she paused. Her husband quietly added, "She was diagnosed with breast cancer at the same time."

Amidst the emotional chaos of these events, however, there was still a beacon of hope. Although proving lethal for the patient's daughter, her son's experience with Li Fraumeni Syndrome had been slightly different. He undergoes routine screening for malignancy, but during our conversation was happy and healthy. Nothing had been detected yet. Still, it is a never-ending waiting game for the patient and her family.

The patient reflected on one incident where a possible mass was seen in her son's colon. At the time, it was indeterminate whether the mass represented malignancy or stool burden. She and her husband chuckled at the memory of chanting, "pray for poop!" Further workup revealed no mass. There wasn't any couple that day more relieved to hear a diagnosis of "poop."

With the possibility of cancer always on the horizon, however, the patient is emphatic about being upfront with her living children, now 11-years-old and 7-years-old, about her condition. "It is never a 'booboo,' she said, "It is cancer."

I was astonished at the remarkable calm and nonchalant manner in which the patient spoke about her terminal diagnosis. She appeared to have completely come to terms with the continuous onslaught of malignancy and the unpredictability of her future. She seemed so resolute, so level-headed, despite what I had seen on her scans, and what I was sure her doctors had relayed to her about her future.

I imagined she was doing it for her family, for her husband and children who still had so much living to do.

In the room, despite their unmistakable unity and mutual resilience, it was clear that she was the central vessel, keeping the pair afloat through stormy waters. During our visit, when his words faltered, her positivity swelled. When his back slumped, she placed a hand on his knee. It was a dynamic I was certain continued into their everyday life, as well as her interactions with her children,

friends, and extended family. Whatever portion of her being had been eroded away following her child's death, appeared to be stabilized, filled in, and overflowing by a drive to live. She had so much to live for.

After our conversation, for some reason, the patient thanked us. We were definitely the only people in that room who should have been doing any thanking. She also gave us some parting advice. It was that she, like many other patients, did not fit into a "bell curve," and to never approach any patient as such.

Neither I nor the other medical student talked much after we left that room, other than to reflect on how remarkable our conversation had been. "I have never met someone like that," he said.

To this day, I am not sure what my attending had hoped my fellow medical student and I would get out of the interaction. Perhaps he, like me, was amazed at the numerous cancers that had defiled her body. That her one-year prognosis had stretched—doubling, tripling—until it became meaningless. That she was only 39-years-old, and yet had experienced more physical hardship than most people twice her age. Maybe, deep down, he had known that there was more to her than her CT scan revealed. Despite what we are led to believe, a radiographic image does not hold all the answers.

Later, reviewing her chart, I saw something on her final CT report that made my heart lurch. New enhancing mass within the mucosal lining of the stomach. Simple yet powerful words, conveying a probable new malignancy. With the image of the woman behind the screen fresh in my mind, this phrase carried new meaning. I felt unworthy to be reading this information, to know ahead of time what may be in store for this young family. Still, regardless of what these new findings meant for the patient, I hoped that she would continue to be the celebrity I had the privilege of meeting.

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