The Story Within
Personal Essays on Genetics and Identity

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Introduction
Amy Boesky

The body is never a single physical thing so much as a series of attitudes towards it.

LENNARD J. DAVIS,
"The End of Identity Politics"

This collection grew out of the conviction that personal narrative reveals a great deal about the connections between genetics and identity. As contributors, we share challenges and complexities associated with genetic conditions, as well as the hope that ongoing research may provide better clinical options in the future. Further, we share a commitment to disclosing our experiences, improving ways that genetic identity gets talked about and understood by the wider public.¹

Rapid expansion in the field of genetics has deepened misunderstandings about the extent to which genetic information determines us. As these essays reveal, no aspect of genetic identity is simple. Risk of predicted disease may be perceived as unbearable by one individual while remaining manageable for another. One person chooses to learn all he can about his genetic make-up, while another defers that information or actively chooses not to discover it. Whether, when, and how to intervene in a predicted genetic condition are all highly individual decisions, as are determinations about whether and how best to involve family members, immediate and extended. Our responses to these issues remain as individual as we are.

Yet the complexities surrounding these questions are often misunderstood. As a writer and professor of literature, I believe narrative helps to
**Undiagnosed**  
Kelly Cupo

*In April 2010, the Icelandic volcano Eyjafjallajökull erupted for the second time in less than a month, pouring floodwaters from its glacier and releasing a cloud of ash that moved across Europe. There are probably lots of volcano stories out there. Here’s mine.*

Once upon a time, sophomore year of college, I was sitting on my bed talking to a friend—we’ll call her The Friend—about a former roommate—we’ll call her Rachel—who wasn’t delivering on the friendship part of our friendship. Run of the undergrad mill. The Friend was sitting on my roommate’s roll-y chair. Me, under my covers in bed.

We’d just gotten back from breakfast in the dining hall. Pancakes like paper. Butter, unwhipped.

“I feel like Rachel’s being really selfish, and it’s ticking me off. She comes in here and is friends with me when it’s convenient, but then she turns it off in a second when it isn’t,” says yours truly: heroine, princess. Me.


“I get that we’re not totally compatible, but I really care about her! We lived together a whole year, and I just don’t think I should be treated that way.”

Her turn. “You know what, Kelly, I feel like you get extra-sensitive when it comes to thinking people are selfish. It’s probably because of your mom and everything.”

Oh what.

We were not talking about my mom. Why are we talking about my mom? She goes on. She’s diagnosing me.
Driving North
Charlie Pierce

Almost thirty years ago, my father drove to a flower store to buy geraniums to plant on the family plots for Memorial Day. They were tearing up the streets of the small Massachusetts town in which he had lived since 1951. After they detoured him around the town square, he drove north and didn’t stop until he got to Montpelier in Vermont. He was missing for three days.

For the next four years, he battled through the worst of the Alzheimer’s disease that had been afflicting him for more than a decade, the disease that my mother had kept in their small house like a malignant secret. It nearly tore my family apart.

My father died twenty-three years ago last June. Twelve years ago, I wrote a book about it all, about the disease and my family and about the race among scientists to solve the riddle of Alzheimer’s so that more families will not have to go through what mine went through. Eventually, all four of my father’s siblings developed the symptoms of the disease and then died. At one point, each of them was convinced they saw their dead mother standing in front of them. One of them thought a college football team was living in his attic. The last one, my aunt, took to sending checks in odd, pin-money amounts to the Vatican. (Even more remarkably, someone there was cashing them.) Therapies had improved considerably by then. My father stopped speaking a year before he died. By 2003, my aunt was still capable of telling the same stories, over and over again. She told about the time we all walked to Mass on Christmas morning in the snow, right up until nearly the end.
Of Helices, HIPAA, Hairballs . . .
and Humans

Misha Angrist

In early 2006, Scientific American ran a cover story entitled "Genomes for All," written by George Church, a prominent geneticist at Harvard. Church envisioned a day in the near future when complete genome sequences would be affordable for most people. So, he wondered, how exactly were we going to go about this?

For me, the question itself was a revelation. Church was saying what I had felt for years but could never articulate: the time for genetic exceptionalism—the idea that genetic information is special and should be treated differently than, say, cholesterol levels or cancer diagnoses—was over, and the time to include large numbers of ordinary volunteers in the act of genome sequencing had arrived.

To that end, Church launched the Personal Genome Project, a visionary experiment in which informed citizens could have their genomes sequenced for free if they consented to make the results—and their medical information—available for public use. I badgered Church until he let me into the pilot group of ten participants, which included, among others, Church himself, Steven Pinker, and Esther Dyson.

Subsequently, I had my complete genome sequenced—most of the six billion DNA letters in my cells—but for all my excitement about the project, my results turned out to be something of a nonevent, as they have for most otherwise healthy people in the PGP and elsewhere. As a card-carrying genetics geek, I was captivated by both the enormity of my sequence and the minutiae. I browsed my favorite genes and was awestruck
The Power of Two

Two Sisters, Two Genes, and Two New Chances at Life

Anabel Stenzel and Isabel Stenzel Byrnes

Anabel

When I contemplate my genetic identity, an image comes to mind: a magnificent blooming tree with three intertwined branches. The tree has been watered by luck, compassion, and resilience.

The first branch is made of the genes of my biracial heritage. My Japanese mother, Hatsuko, and German father, Reiner, both raised in war-torn countries, came to America for higher education. They met in Southern California and fell in love in the mid-1960s. In their genes, they carried core values from their cultures. From my mother’s Japanese heritage came humility, discipline, stoicism, and most of all, *gaman*, the Japanese word for “perseverance.” My father, a German physicist, passed down to his children traits of curiosity, skepticism, the love of nature, and pursuit of the facts.

My parents shared compatible childhood stories of the Allied Occupation and their adult pursuits of the American Dream. Complementing each other’s personalities in the most unusual ways, they wed in 1967. In 1970, our mother gave birth to a healthy boy named Ryuta, meaning “Big Dragon” in Japanese. Nine months later, my mother found herself pregnant again, uncertain about her ability to raise another baby so soon. After all, her own family was so far away and she had limited support.

My mother found herself in labor six weeks early. Just two hours prior to delivery, my mother was told that she was carrying twins. A mysterious miracle transpired when the zygote split and formed two
What If
Laurie Strongin

If you knew Henry would suffer and then die, would you still have had him?”

It was 6:20 one morning in the spring of 2012. I’d come downstairs to make coffee, when I saw my 10-year-old son, Joe, sitting on the couch staring at a picture of his brother Henry on his computer screen. An old picture: Henry in his ever-present Batman costume.

“Yes. Of course,” I said.

Joe was only 1 year old when Henry died, at age 7, of a rare, deadly genetic disease called Fanconi anemia. In an effort to stay connected to this brother who he doesn’t remember, Joe asks a lot of questions.

“Did Henry like to play with me?”

“He loved playing with you. He called you Little JoJo and loved to hold you in his lap and play peekaboo. You made him really happy.”

“Did Henry know he was going to die?”

“No. Up until the end of his life, he never thought of himself as sick. He was always getting better.”

“Was he scared?”

“No. He was really brave. He’d look at the nurse who was about to stick him with a needle and he’d say, ‘Bring it on!’ with a grin. He did a lot of hard things, but he nearly always had a smile on his face. He had big dimples, just like you and Jack.”

“How did he get sick?”

“That’s a really complicated question.”

I explained that Henry got his disease from his dad (my husband, Allen) and me. We each had inherited a defective gene for Fanconi anemia from our mothers, which together we had passed on to Henry.
The Long Arm
Clare Dunsford

As mutations go, the one my family carries is particularly perverse. Part of a group of mutations described as “expanding” or “dynamic,” because the gene changes in size over generations, fragile X—our family curse—cannot be explained by Mendelian inheritance, for the gene is neither completely dominant nor fully recessive. Initially, the pattern of inheritance of fragile X was so contrary to what scientists had known until its discovery that, when it was understood, it was dubbed the “Sherman paradox,” after the scientist who explained its baffling characteristics in 1985, the year my son was born.

What puzzled doctors was that if a condition is X-linked, it usually affects only males, but fragile X affects females, too. Moreover, the males who passed on the fragile X mutation to their daughters seemed unaffected by it. What Dr. Stephanie Sherman figured out was that the seemingly unaffected mothers and grandfathers of boys with fragile X were actually carrying a premutation of the gene. In other words, they were not genetically normal, although they were phenotypically so.¹

The FMR1 gene is located on the X chromosome, on what is called the long arm (all chromosomes have a short arm and a long arm, pinched in the middle by the centromere). Fragile X syndrome is one of a group of conditions called trinucleotide repeat disorders, in which a section of the gene increases in size, duplicating itself in a disastrous stutter to the point that the gene becomes unstable. A typical individual has fewer than 45 repeats of a phrase that reads CGG on the FMR1 gene, but someone, like me, with the premutation of the FMR1 gene has between 55 and 200 repeats. (There is a gray zone of 45 to 55 repeats in which the gene...