



Facing Life With a Lethal Gene

By AMY HARMON
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The test, the counselor said, had come back positive.

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Suzanne DeChillo/The New York Times

Katharine Moser at a nursing home in Manhattan with her cousin James Dowd, who has Huntington's disease. [More Photos](#)

Katharine Moser inhaled sharply. She thought she was as ready as anyone could be to face her genetic destiny. She had attended a genetic counseling session and visited a psychiatrist, as required by the clinic. She had undergone the recommended neurological exam. And yet, she realized in that moment, she had never expected to hear those words.

"What do I do now?" Ms. Moser asked.

"What do you want to do?" the counselor replied.

"Cry," she said quietly.

Her best friend, Colleen Elio, seated next to her, had already begun.

Ms. Moser was 23. It had taken her months to convince the clinic at NewYork-Presbyterian Hospital/Columbia University Medical Center in Manhattan that she wanted, at such a young age, to find out whether she carried the gene for Huntington's disease.

Huntington's, the incurable brain disorder that possessed her grandfather's body and ravaged his mind for three decades, typically strikes in middle age. But most young adults who know the disease runs in their family have avoided the [DNA](#) test that can tell whether they will get it, preferring the torture — and hope — of not knowing.

Ms. Moser is part of a vanguard of people at risk for Huntington's who are choosing to learn early what their future holds. Facing their genetic heritage, they say, will help them decide how to live their lives.

Yet even as a raft of new DNA tests are revealing predispositions to all kinds of conditions, including [breast cancer](#), depression and dementia, little is known about what it is like to live with such knowledge.

"What runs in your own family, and would you want to know?" said Nancy Wexler, a neuropsychologist at Columbia and the president of the Hereditary Disease Foundation, which has pioneered Huntington's research. "Soon everyone is going to have an option like this. You make the decision to test, you have to live with the consequences."

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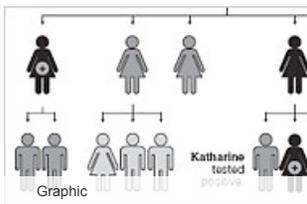
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Suzanne DeChillo/The New York Times

Colleen Elio, Ms. Moser's best friend, with her son Rory. In the background is James Dowd, Ms. Moser's cousin, who had come to live in the Huntington's unit several years earlier. [More Photos >](#)

On that drizzly spring morning two years ago, Ms. Moser was feeling her way, with perhaps the most definitive and disturbing verdict genetic testing has to offer. Anyone who carries the gene will inevitably develop Huntington's.

She fought her tears. She tried for humor.

Don't let yourself get too thin, said the clinic's social worker. Not a problem, Ms. Moser responded, gesturing to her curvy frame. No more than two drinks at a time. Perhaps, Ms. Moser suggested to Ms. Elio, she meant one in each hand.

Then came anger.

"Why me?" she remembers thinking, in a refrain she found hard to shake in the coming months. "I'm the good one. It's not like I'm sick because I have [emphysema](#) from [smoking](#) or I did something dangerous."

The gene that will kill Ms. Moser sits on the short arm of everyone's fourth chromosome, where the letters of the genetic alphabet normally repeat C-A-G as many as 35 times in a row. In people who develop Huntington's, however, there are more than 35 repeats.

No one quite knows why this DNA hiccup causes cell death in the brain, leading Huntington's patients to jerk and twitch uncontrollably and rendering them progressively unable to walk, talk, think and swallow. But the greater the number of repeats, the earlier symptoms tend to appear and the faster they progress.

Ms. Moser's "CAG number" was 45, the counselor said. She had more repeats than her grandfather, whose first symptoms — loss of short-term memory, mood swings and a constant ticking noise he made with his mouth — surfaced when he turned 50. But it was another year before Ms. Moser would realize that she could have less than 12 years until she showed symptoms.

Immediately after getting her results, Ms. Moser was too busy making plans.

"I'm going to become super-strong and super-balanced," she vowed over lunch with Ms. Elio, her straight brown hair pulled into a determined bun. "So when I start to lose it I'll be a little closer to normal."

In the tumultuous months that followed, Ms. Moser often found herself unable to remember what normal had once been. She forced herself to renounce the crush she had long nursed on a certain firefighter, sure that marriage was no longer an option for her. She threw herself into fund-raising in the hopes that someone would find a cure. Sometimes, she raged.

She never, she said, regretted being tested. But at night, crying herself to sleep in the dark of her lavender bedroom, she would go over and over it. She was the same, but she was also different. And there was nothing she could do.

A Lesson in Stigma

Ms. Moser grew up in Connecticut, part of a large Irish Catholic family. Like many families affected by Huntington's, Ms. Moser's regarded the disease as a curse, not to be mentioned even as it

dominated their lives in the form of her grandfather's writhing body and unpredictable rages.

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Once, staying in Ms. Moser's room on a visit, he broke her trundle bed with his violent, involuntary jerking. Another time, he came into the kitchen naked, his underpants on his head. When the children giggled, Ms. Moser's mother defended her father: "If you don't like it, get out of my house and go."

But no one explained what had happened to their grandfather, Thomas Dowd, a former New York City police officer who once had dreams of retiring to Florida.

In 1990, Mr. Dowd's older brother, living in a veteran's hospital in an advanced stage of the disease, was strangled in his own restraints. But a year or so later, when Ms. Moser wanted to do her sixth-grade science project on Huntington's, her mother recoiled.

"Why," she demanded, "would you want to do it on this disease that is killing your grandfather?"

Ms. Moser was left to confirm for herself, through library books and a CD-ROM encyclopedia, that she and her brothers, her mother, her aunts, an uncle and cousins could all face the same fate.

Any child who has a parent with Huntington's has a 50 percent chance of having inherited the gene that causes it, Ms. Moser learned.

Her mother, who asked not to be identified by name for fear of discrimination, had not always been so guarded. At one point, she drove around with a "Cure HD" sign in the window of her van. She told people that her father had "[Woody Guthrie's](#) disease," invoking the folk icon who died of Huntington's in 1967.

But her efforts to raise awareness soon foundered. Huntington's is a rare genetic disease, affecting about 30,000 people in the United States, with about 250,000 more at risk. Few people know what it is. Strangers assumed her father's unsteady walk, a frequent early symptom, meant he was drunk.

"Nobody has compassion," Ms. Moser's mother concluded. "People look at you like you're strange, and 'What's wrong with you?'"

Shortly after a simple DNA test became available for Huntington's in 1993, one of Ms. Moser's aunts tested positive. Another, driven to find out if her own medical problems were related to Huntington's, tested negative. But when Ms. Moser announced as a teenager that she wanted to get tested one day, her mother insisted that she should not. If her daughter carried the gene, that meant she did, too. And she did not want to know.

"You don't want to know stuff like that," Ms. Moser's mother said in an interview. "You want to enjoy life."

Ms. Moser's father, who met and married his wife six years before Ms. Moser's grandfather received his Huntington's diagnosis, said he had managed not to think much about her at-risk status.

"So she was at risk," he said. "Everyone's at risk for everything."

The test, Ms. Moser remembers her mother suggesting, would cost thousands of dollars. Still, in college, Ms. Moser often trolled the Web for information about it. Mostly, she imagined how sweet it would be to know she did not have the gene. But increasingly she was haunted, too, by the suspicion that her mother did.

As awful as it was, she admitted to Ms. Elio, her freshman-year neighbor at Elizabethtown College in Pennsylvania, she almost hoped it was true. It would explain her mother's strokes of meanness, her unpredictable flashes of anger.

Ms. Moser's mother said she had never considered the conflicts with her daughter out of the ordinary. "All my friends who had daughters said that was all normal, and when she's 25 she'll be your best friend," she said. "I was waiting for that to happen, but I guess it's not happening."

When Ms. Moser graduated in 2003 with a degree in occupational therapy, their relationship, never peaceful, was getting worse. She moved to Queens without giving her mother her new address.

Wanting to Know

Out of school, Ms. Moser soon spotted a listing for a job at Terence Cardinal Cooke Health Care Center, a nursing home on the Upper East Side of Manhattan. She knew it was meant for her.

Her grandfather had died there in 2002 after living for a decade at the home, one of only a handful in the country with a unit devoted entirely to Huntington's.

"I hated visiting him growing up," Ms. Moser said. "It was scary."

Now, though, she was drawn to see the disease up close.

On breaks from her duties elsewhere, she visited her cousin James Dowd, the son of her grandfather's brother who had come to live in the Huntington's unit several years earlier. It was there, in a conversation with another staff member, that she learned she could be tested for only a few hundred dollars at the Columbia clinic across town. She scheduled an appointment for the next week.

The staff at Columbia urged Ms. Moser to consider the downside of genetic testing. Some people battle depression after they test positive. And the information, she was cautioned, could make it harder for her to get a job or health insurance.

But Ms. Moser bristled at the idea that she should have to remain ignorant about her genetic status to avoid discrimination. "I didn't do anything wrong," she said. "It's not like telling people I'm a drug addict."

She also recalls rejecting a counselor's suggestion that she might have asked to be tested as a way of crying for help.

"I'm like, 'No,'" Ms. Moser recalls replying. "I've come to be tested because I want to know."

No one routinely collects demographic information about who gets tested for Huntington's. At the [Huntington's Disease](#) Center at Columbia, staff members say they have seen few young people taking the test.

Ms. Moser is still part of a distinct minority. But some researchers say her attitude is increasingly common among young people who know they may develop Huntington's.

More informed about the genetics of the disease than any previous generation, they are convinced that they would rather know how many healthy years they have left than wake up one day to find the illness upon them. They are confident that [new reproductive technologies](#) can allow them to have children without transmitting the disease and are eager to be first in line should a treatment become available.

"We're seeing a shift," said Dr. Michael Hayden, a professor of human genetics at the University of British Columbia in Vancouver who has been providing various tests for Huntington's for 20 years. "Younger people are coming for testing now, people in their 20s and early 30s; before, that was very rare. I've counseled some of them. They feel it is part of their heritage and that it is possible to lead a life that's not defined by this gene."

Before the test, Ms. Moser made two lists of life goals. Under "if negative," she wrote married, children and Ireland. Under "if positive" was exercise, [vitamins](#) and ballroom dancing. Balance, in that case, would be important. Opening a bed-and-breakfast, a goal since childhood, made both lists.

In the weeks before getting the test results, Ms. Moser gave Ms. Elio explicit instructions about acceptable responses. If she was negative, flowers were O.K. If positive, they were not. In either case, drinking was acceptable. Crying was not.

But it was Ms. Elio's husband, Chris Elio, who first broached the subject of taking care of Ms. Moser, whom their young children called "my Katie," as in "this is my mom, this is my dad, this is my Katie." They should address it before the results were in, Mr. Elio told his wife, so that she would not feel, later, that they had done it out of a sense of obligation.

The next day, in an e-mail note that was unusually formal for friends who sent text messages constantly and watched "Desperate Housewives" while on the phone together, Ms. Elio told Ms. Moser that she and her husband wanted her to move in with them if she got sick. Ms. Moser set the note aside. She did not expect to need it.

'It's Too Hard to Look'

The results had come a week early, and Ms. Moser assured her friends that the "Sex and the City" trivia party she had planned for that night was still on. After all, she was not sick, not dying. And she had

already made the dips.

“I’m the same person I’ve always been,” she insisted that night as her guests gamely dipped strawberries in her chocolate fountain. “It’s been in me from the beginning.”

But when she went to work the next day, she lingered outside the door of the occupational therapy gym, not wanting to face her colleagues. She avoided the Huntington’s floor entirely, choosing to attend to patients ailing of just about anything else. “It’s too hard to look at them,” she told her friends.

In those first months, Ms. Moser summoned all her strength to pretend that nothing cataclysmic had happened. At times, it seemed easy enough. In the mirror, the same green eyes looked back at her. She was still tall, a devoted Julia Roberts fan, a prolific baker.

She dropped the news of her genetic status into some conversations like small talk, but kept it from her family. She made light of her newfound fate, though often friends were not sure how to take the jokes.

“That’s my Huntington’s kicking in,” she told Rachel Markan, a co-worker, after knocking a patient’s folder on the floor.

Other times, Ms. Moser abruptly dropped any pretense of routine banter. On a trip to Florida, she and Ms. Elio saw a man in a wheelchair being tube-fed, a method often used to keep Huntington’s patients alive for years after they can no longer swallow.

“I don’t want a feeding tube,” she announced flatly.

In those early days, she calculated that she had at least until 50 before symptoms set in. That was enough time to open a bed-and-breakfast, if she acted fast. Enough time to repay \$70,000 in student loans under her 30-year term.

Doing the math on the loans, though, could send her into a tailspin.

“I’ll be repaying them and then I’ll start getting sick,” she said. “I mean, there’s no time in there.”

Finding New Purpose

At the end of the summer, as the weather grew colder, Ms. Moser forced herself to return to the Huntington’s unit.

In each patient, she saw her future: the biophysicist slumped in his wheelchair, the refrigerator repairman inert in his bed, the onetime professional tennis player who floated through the common room, arms undulating in the startlingly graceful movements that had earned the disease its original name, “Huntington’s chorea,” from the Greek “to dance.”

Then there was her cousin Jimmy, who had wrapped papers for The New York Post for 19 years until suddenly he could no longer tie the knots. When she greeted him, his bright blue eyes darted to her face, then away. If he knew her, it was impossible to tell.

She did what she could for them. She customized their wheelchairs with padding to fit each one’s unique tics. She doled out special silverware, oversized or bent in just the right angles to prolong their ability to feed themselves.

Fending off despair, Ms. Moser was also filled with new purpose. Someone, somewhere, she told friends, had to find a cure.

It has been over a century since the disease was identified by George Huntington, a doctor in Amagansett, N.Y., and over a decade since researchers first found the gene responsible for it.

To raise money for research, Ms. Moser volunteered for walks and dinners and golf outings sponsored by the [Huntington’s Disease Society of America](#). She organized a Hula-Hoop-a-thon on the roof of Cardinal Cooke, then a bowl-a-thon at the Port Authority. But at many of the events, attendance was sparse.

It is hard to get people to turn out for Huntington’s benefits, she learned from the society’s professional fund-raisers. Even families affected by the disease, the most obvious constituents, often will not help publicize events.

“They don’t want people to know they’re connected to Huntington’s,” Ms. Moser said, with a mix of anger and recognition. “It’s like in my family — it’s not a good thing.”

Her first session with a therapist brought a chilling glimpse of how the disorder is viewed even by some who know plenty about it. “She told me it was my moral and ethical obligation not to have children,” Ms. Moser told Ms. Elio by cellphone as soon as she left the office, her voice breaking.

In lulls between fund-raisers, Ms. Moser raced to educate her own world about Huntington’s. She added links about the disease to her MySpace page. She plastered her desk at work with “Cure HD” stickers and starred in a video about the Huntington’s unit for her union’s Web site.

Ms. Moser gave blood for one study and spoke into a microphone for researchers trying to detect subtle speech differences in people who have extra CAG repeats before more noticeable disease symptoms emerge.

When researchers found a way to cure mice bred to replicate features of the disease in humans, Ms. Moser sent the news to friends and acquaintances.

But it was hard to celebrate. “Thank God,” the joke went around on the Huntington’s National Youth Alliance e-mail list Ms. Moser subscribed to, “at least there won’t be any more poor mice wandering around with Huntington’s disease.”

In October, one of Ms. Moser’s aunts lost her balance while walking and broke her nose. It was the latest in a series of falls. “The cure needs to be soon for me,” Ms. Moser said. “Sooner for everybody else.”

A Confrontation in Court

In the waiting room of the Dutchess County family courthouse on a crisp morning in the fall of 2005, Ms. Moser approached her mother, who turned away.

“I need to tell her something important,” Ms. Moser told a family member who had accompanied her mother to the hearing.

He conveyed the message and brought one in return: Unless she was dying, her mother did not have anything to say to her.

That Ms. Moser had tested positive meant that her mother would develop Huntington’s, if she had not already. A year earlier, Ms. Moser’s mother had convinced a judge that her sister, Nora Maldonado, was neglecting her daughter. She was given guardianship of the daughter, 4-year-old Jillian.

Ms. Moser had been skeptical of her mother’s accusations that Ms. Maldonado was not feeding or bathing Jillian properly, and she wondered whether her effort to claim Jillian had been induced by the psychological symptoms of the disease.

Her testimony about her mother’s genetic status, Ms. Moser knew, could help persuade the judge to return Jillian. Ms. Maldonado had found out years earlier that she did not have the Huntington’s gene.

Ms. Moser did not believe that someone in the early stages of Huntington’s should automatically be disqualified from taking care of a child. But her own rocky childhood had convinced her that Jillian would be better off with Ms. Maldonado.

She told her aunt’s lawyer about her test results and agreed to testify.

In the courtroom, Ms. Moser took the witness stand. Her mother’s lawyer jumped up as soon as the topic of Huntington’s arose. It was irrelevant, he said. But by the time the judge had sustained his objections, Ms. Moser’s mother, stricken, had understood.

The next day, in the bathroom, Ms. Maldonado approached Ms. Moser’s mother.

“I’m sorry,” she said. Ms. Moser’s mother said nothing.

The court has continued to let Ms. Moser’s mother retain guardianship of Jillian. But she has not spoken to her daughter again.

“It’s a horrible illness,” Ms. Moser’s mother said, months later, gesturing to her husband. “Now he has a

wife who has it. Did she think of him? Did she think of me? Who's going to marry her?"

Facing the Future

Before the test, it was as if Ms. Moser had been balanced between parallel universes, one in which she would never get the disease and one in which she would. The test had made her whole.

She began to prepare the Elio children and Jillian for her illness, determined that they would not be scared, as she had been with her grandfather. When Jillian wanted to know how people got Huntington's disease "in their pants," Ms. Moser wrote the text of a children's book that explained what these other kinds of "genes" were and why they would make her sick.

But over the winter, Ms. Elio complained gently that her friend had become "Ms. H.D." And an impromptu note that arrived for the children in the early spring convinced her that Ms. Moser was dwelling too much on her own death.

"You all make me so happy, and I am so proud of who you are and who you will be," read the note, on rainbow scratch-and-write paper. "I will always remember the fun things we do together."

Taking matters into her own hands, Ms. Elio created a profile for Ms. Moser on an online dating service. Ms. Moser was skeptical but supplied a picture. Dating, she said, was the worst thing about knowing she had the Huntington's gene. It was hard to imagine someone falling enough in love with her to take on Huntington's knowingly, or asking it of someone she loved. At the same time, she said, knowing her status could help her find the right person, if he was out there.

"Either way, I was going to get sick," she said. "And I'd want someone who could handle it. If, by some twist of fate, I do get married and have children, at least we know what we're getting into."

After much debate, the friends settled on the third date as the right time to mention Huntington's. But when the first date came, Ms. Moser wished she could just blurt it out.

"It kind of just lingers there," she said. "I really just want to be able to tell people, 'Someday, I'm going to have Huntington's disease.'"

'A Part of My Life'

Last May 6, a year to the day after she had received her test results, the subject line "CAG Count" caught Ms. Moser's attention as she was scrolling through the online discussion forums of the [Huntington's Disease Advocacy Center](#). She knew she had 45 CAG repeats, but she had never investigated it further.

She clicked on the message.

"My mother's CAG was 43," it read. "She started forgetting the punch line to jokes at 39/40." Another woman whose husband's CAG count was 47 had just sold his car. "He's 39 years old," she wrote. "It was time for him to quit driving."

Quickly, Ms. Moser scanned a chart that accompanied the messages for her number, 45. The median age of onset to which it corresponded was 37.

Ms. Elio got drunk with her husband the night Ms. Moser finally told her.

"That's 12 years away," Ms. Moser said.

The statistic, they knew, meant that half of those with her CAG number started showing symptoms after age 37. But it also meant that the other half started showing symptoms earlier.

Ms. Moser, meanwhile, flew to the annual convention of the Huntington's Disease Society, which she had decided at the last minute to attend.

"Mother or father?" one woman, 23, from Chicago, asked a few minutes after meeting Ms. Moser in the elevator of the Milwaukee Hilton. "Have you tested? What's your CAG?"

She was close to getting herself tested, the woman confided. How did it feel to know?

"It's hard to think the other way anymore of not knowing," Ms. Moser replied. "It's become a part of my life."

After years of trying to wring conversation from her family about Huntington's, Ms. Moser suddenly found herself bathing in it. But for the first time in a long time, her mind was on other things. At a youth support group meeting in the hotel hallway, she took her place in the misshapen circle. Later, on the dance floor, the spasms of the symptomatic seemed as natural as the gyrations of the normal.

"I'm not alone in this," Ms. Moser remembers thinking. "This affects other people, too, and we all just have to live our lives."

Seizing the Day

July 15, the day of Ms. Moser's 25th birthday party, was sunny, with a hint of moisture in the air. At her aunt's house in Long Beach, N.Y., Ms. Moser wore a dress with pictures of cocktails on it. It was, she and Ms. Elio told anyone who would listen, her "cocktail dress." They drew the quotation marks in the air.

A bowl of "Cure HD" pins sat on the table. Over burgers from the barbecue, Ms. Moser mentioned to family members from her father's side that she had tested positive for the Huntington's gene.

"What's that?" one cousin asked.

"It will affect my ability to walk, talk and think," Ms. Moser said. "Sometime before I'm 50."

"That's soon," an uncle said matter-of-factly.

"So do you have to take medication?" her cousin asked.

"There's nothing really to take," Ms. Moser said.

She and the Elios put on bathing suits, loaded the children in a wagon and walked to the beach.

More than anything now, Ms. Moser said, she is filled with a sense of urgency.

"I have a lot to do," she said. "And I don't have a lot of time."

Over the next months, Ms. Moser took tennis lessons every Sunday morning and went to church in the evening.

When a planned vacation with the Elio family fell through at the last minute, she went anyway, packing Disney World, Universal Studios, Wet 'n Wild and Sea World into 36 hours with a high school friend who lives in Orlando. She was honored at a dinner by the New York chapter of the Huntington's society for her outreach efforts and managed a brief thank-you speech despite her discomfort with public speaking.

Having made a New Year's resolution to learn to ride a unicycle, she bought a used one. "My legs are tired, my arms are tired, and I definitely need protection," she reported to Ms. Elio. On Super Bowl Sunday, she waded into the freezing Atlantic Ocean for a Polar Bear swim to raise money for the Make-a-Wish Foundation.

Ms. Elio complained that she hardly got to see her friend. But one recent weekend, they packed up the Elio children and drove to the house the Elios were renovating in eastern Pennsylvania. The kitchen floor needed grouting, and, rejecting the home improvement gospel that calls for a special tool designed for the purpose, Ms. Moser and Ms. Elio had decided to use pastry bags.

As they turned into the driveway, Ms. Moser studied the semi-attached house next door. Maybe she would move in one day, as the Elios had proposed. Then, when she could no longer care for herself, they could put in a door.

First, though, she wanted to travel. She had heard of a job that would place her in different occupational therapy positions across the country every few months and was planning to apply.

"I'm thinking Hawaii first," she said.

Then they donned gloves, mixed grout in a large bucket of water and began the job.

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