

EXCERPTS FROM *STILL ALICE*, BY LISA GENOVA

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Page 58, Diagnosis: Neurologist and Alice

Neurologist: “I have the results of all of your tests now, so we can go over everything. I don’t see anything abnormal in your MRI. No cerebral vascular disease, no evidence of any small, silent strokes, no hydrocephalus or masses. Everything there looks fine. And your blood work and lumbar puncture all came back negative as well. I was as aggressive here as we can be and looked for every condition that could sensibly account for the kinds of symptoms you’re experiencing. So we know you don’t have HIV, cancer, a vitamin deficiency, mitochondrial disease, or a number of other rare conditions.”

His speech was well constructed, obviously not his first delivery of its kind. The ‘what she did have’ would come at the end. She nodded, letting him know that she followed him and that he should continue.

Neurologist: “You scored in the ninety-ninth percentile in your ability to attend, in things like abstract reasoning, spatial skills, and language fluency. But unfortunately, here’s what I do see. You have a recent memory impairment that is out of proportion to your age and is a significant decline in your previous level of functioning. I know this from your own account of the problems you’ve been having and from your description of the degree to which they’ve been interfering with your professional life. I also personally witnessed it when you couldn’t retrieve the address I’d asked you to remember the last time you were here. And although you were perfect in most of the cognitive domains today, you showed a lot of variability in two of the tasks that were related to recent memory. In fact, you were down to the sixtieth percentile in one. When I put all of this information together, Alice, what it tells me is that you fit the criteria of having probable Alzheimer’s disease.”

Alzheimer’s disease. The words knocked the wind out of her. What exactly did he just tell her? She repeated his words in her head. Probable. It gave her the will to inhale, the ability to speak.

Alice: “So ‘probable’ means that I might not fit the criteria.”

Neurologist: “No, we use the word ‘probable’ because the only definitive diagnosis for Alzheimer’s right now is by examining the histology of the brain tissue, which requires

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either an autopsy or a biopsy, neither of which is a good option for you. It's a clinical diagnosis. There's no dementia protein in your blood that can tell us you have it, and we wouldn't expect to see any brain atrophy on an MRI until much later stages in the disease."

Brain atrophy.

Alice: "But this can't be possible, I'm only fifty."

Neurologist: "You have early-onset Alzheimer's. You're right, we typically think of Alzheimer's as a disease that affects the elderly, but ten percent of people with Alzheimer's have this early-onset form and are under the age of sixty-five."

Alice: "How is that different from the older form?"

Neurologist: "It's not, except that its cause usually has a strong genetic linkage, and it manifests much earlier."

Strong genetic linkage. Anna, Tom, Lydia.

Alice: "But if you only know for sure what I don't have, how can you say with any certainty that this is Alzheimer's?"

Neurologist: "After listening to you describe what's been happening and to your medical history, after testing your orientation, registration, attention, language, and recall, I was ninety-five percent sure. With no other explanation turning up in your neurological exam, blood cerebral spinal fluid, or MRI, the other five percent goes away. I'm sure, Alice."

Page 67-68, Genetic counseling: Genetic counselor, John, Alice

Stephanie Aaron was the genetic counselor affiliated with Mass General Hospital's Memory Disorders Unit... She greeted them with a warm smile.

Stephanie: "So, tell me why you're here today."

John: "My wife was recently told she has Alzheimer's disease, and we want her screened for the APP, PS1, and PS2 mutations."

John had done his homework. He'd spent the last several weeks buried in literature on the molecular etiology of Alzheimer's. Errant proteins born from any of these three mutated genes were the known villains for the early-onset cases.

Stephanie: "Alice, tell me, what are you hoping to learn from the testing?"

Alice: "Well, it seems like a reasonable way to try to confirm my diagnosis. Certainly

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more so than a brain biopsy or an autopsy.”

Stephanie: “Are you concerned that your diagnosis might be inaccurate?”

John: “We think it’s a real possibility.”

Stephanie: “Okay, first, let’s walk through what a positive versus a negative mutation screen would mean for you. Three mutations are fully penetrant. If you’re mutation positive for APP, PS1, or PS2, I would say that’s a solid confirmation of your diagnosis. Things get a bit tricky, though, if your results come back negative. We can’t really interpret with any certain what that would mean. About fifty percent of people with early-onset Alzheimer’s don’t show a mutation in any of these three genes. This isn’t to say that they don’t actually have Alzheimer’s or that their disease isn’t genetically based, it’s just that we don’t yet know the gene in which their mutation resides.”

John: “Isn’t that number more like ten percent for someone her age?”

Stephanie: “The numbers are a bit more skewed for someone her age, that’s true. But if Alice’s screen comes back negative, we unfortunately can’t say for sure that she doesn’t have the disease. She may just happen to fall in the smaller perfect of people that age with Alzheimer’s who have a mutation in a gene not yet identified.”

Stephanie (to Alice): “I also want to make it clear that if your screening comes back with a positive mutation, a genetic diagnosis isn’t going to change anything about your treatment or prognosis.... Okay, a nurse is going to come in and draw a sample of blood. We’ll send it off to be sequenced and should have the results within a couple of weeks.”

Page 70, Diagnosis: Genetic counselor, Alice, John

Stephanie was sitting behind her desk when they came in, but this time, she didn’t smile.

Stephanie: “Before we talk about your results, is there anything you’d like to review about any of the information we went over last time?”

Alice: “No.”

Stephanie: “Do you still want the results?”

Alice: “Yes.”

Stephanie: “I’m sorry to tell you, Alice, you’re positive for the PS1 mutation.... As we talked about, this mutation is autosomal dominant; it’s associated with certain development Alzheimer’s, so this result fits with the diagnosis you’ve already received.”

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John: “What’s the lab’s false positive rate? What’s the name of the lab?”

Stephanie: “It’s Athena Diagnostics, and they cite a greater than ninety-nine-percent accuracy level of detection for this mutation.”

Alice: “John, it’s positive.”

Stephanie: “I’m sorry, I know you were both searching for a way out of this diagnosis.”

Alice: “What does this mean for our children?”

Stephanie: “Yes, there’s a lot to think about there. How old are they?”

Alice: “They’re all in their twenties.”

Stephanie: “So we wouldn’t expect any of them to be symptomatic yet. Each of your children has a fifty percent chance of inheriting this mutation, which has a one hundred percent chance of causing the disease. Presymptomatic genetic testing is possible, but there’s a lot to consider. Is this something they’ll want to live with knowing? How would it change their lives? What if one of them is positive and one is negative, how will that affect their relationship with each other? Alice, do they even know about your diagnosis?”

Alice: “No.”

Stephanie: “You might want to think about telling them soon. I know it’s a lot to unload at once, especially since I know you’re still absorbing it yourselves. But with a progressive illness like this, you can lay out

plans to tell them later, but then you may not be able to in the way you originally wanted. Or maybe this is something you’re going to leave to John to do?”

Alice: “No, we’ll tell them.”

Stephanie: “Do any of your children have children?”

Alice: “Not yet.”

Stephanie: “If they’re planning to, this might be really important information for them to have. Here’s some written information I gathered that you can give them if you want. Also, here’s my card and the card of a therapist who’s wonderful with talking to families who’ve gone through genetic screening and diagnosis. Are there any other questions that I can answer for you now?”

Alice: “No, none that I can think of.”

Stephanie: “I’m sorry I couldn’t give you the results you were hoping for.”

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Alice: “Me, too.”

Page 82-84, Telling the Children: Alice, John, Anna, Tom, Lydia

[Anna and Charlie (her husband) are having trouble getting pregnant and are planning to have in vitro fertilization.]

Alice: “Your dad and I have something important we need to talk to you about, and we wanted to wait until we had all three of you together. I’ve been experiencing some difficulties with my memory for some time now, and in January, I was diagnosed with early-onset Alzheimer’s disease.”

Tom: “Are they sure it’s Alzheimer’s? Did you get a second opinion?”

John: “She had genetic screening. She has the presenilin-1 mutation.”

Tom: “Is it autosomal dominant?”

John: “Yes.”

Anna: “What does that mean? Dad, what did you just tell him?”

Tom: “It means we have a fifty percent chance of getting Alzheimer’s disease.”

Anna: “What about my baby?”

Lydia: “You’re not even pregnant.”

Alice: “Anna, if you have the mutation, it’s the same for your children. Each child you have would have a fifty percent chance of inheriting it, too.”

Anna: “Oh my God, what if I have it? And then my baby could have it.”

Tom: “There’ll probably be a cure by the time any of our kids would need it.”

Anna: “But not in time for us, is that what you’re saying? So my kids will be fine, but I’ll be a mindless zombie?”

John snaps: “Anna, that’s enough!”

Anna: “Sorry.”

Alice: “It’s very likely that there’ll be a preventative treatment by the time you’re my age. That’s one of the reasons to know if you have the mutation. If you do, you might be able to go on a medication well before you’re symptomatic and, hopefully, you never will be.”

Anna: “I have to know if I have this. I want to get tested. Don’t you guys want to get tested?”

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Tom: “I think living with the anxiety of not knowing would be worse for me than knowing, even if I have it.”

Lydia closed her eyes. Everyone waited. Alice entertained the absurd idea that she had either resumed memorizing her lines or fallen asleep. After an uncomfortable silence, she opened her eyes and took her turn.

Lydia: “I don’t want to know.”

Lydia always did things differently.

Page 86–87, Children find out who has the mutation: Alice, Tom, Anna

The odds of them both being negative for the mutation descended from unlikely to remote when they still weren’t home an hour after Alice had anticipated their arrival. If they were both negative, it would have been a quick appointment, a ‘you’re both fine,’ ‘thank you very much,’ and out-the-door appointment... The odds crashed from remote to infinitesimal when they finally walked through the front door. If they were both negative, they would have just blurted it out or it would have sprung, will and jubilant, from their facial expressions. Instead, they muscled what they knew beneath the surface as they moved into the living room, stretching out the time of Life Before This Happened as long as possible, the time before they’d have to unleash the hideous information they so obviously held.

They sat side by side on the couch, Tom on the left and Anna on the right...

Tom: “I don’t have the mutation.”

Anna: “But I do.”

After Tom was born, Alice remembered feeling so blessed, that she had the ideal—one of each. It took twenty-six years for that blessing to deform into a curse. Alice’s façade of stoic parental strength crumbled, and she started to cry.

Alice: “I’m sorry.”

Anna: “It’s going to be okay, Mom. Like you said, they’re going to find a preventative treatment.”

When Alice thought about it later, the irony was striking. Outwardly, at least, Anna appeared to be the strongest. She did most of the consoling. And yet, it didn’t surprise her. Anna was the child who most mirrored their mother. She had Alice’s hair, coloring, and temperament. And her mother’s presenilin-1.

Anna: “I’m going to go ahead with the in vitro. I already talked with my doctor, and

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they're going to do a preimplantation genetic diagnosis on the embryos. They're going to test a single cell from each of the embryos for the mutation and only implant ones that are mutation-free. So we'll know for sure that my kids won't ever get this."

It was a solid piece of good news. But while everyone else continued to savor it, the taste turned slightly bitter for Alice. Despite her self-reproach, she envied Anna, that she could do what Alice couldn't—keep her children safe from harm. Anna would never have to sit opposite her daughter, her firstborn, and watch her struggle to comprehend the news that she would someday develop Alzheimer's. She wished that these kinds of advances in reproductive medicine had been available to her. But then the embryo that had developed into Anna would've been discarded.

According to Stephanie Aaron, Tom was okay, but he didn't look it. He looked pale, shaken, fragile. Alice had imagined that a negative result for any of them would be a relief, clean and simple. But they were a family, yoked by history and DNA and love. Anna was his older sister. She'd taught him how to snap and blow gum bubbles, and she always gave him her Halloween candy.

Tom: "Who's going to tell Lydia?"

Anna: "I will."