Running a marathon to restore sight and sound to Jews affected by genetic disorder

by **Steve Marantz** — April 28, 2022 in **News**

Jaime Recht, who is deaf and partially blind, will run the Boston Marathon on April 18. Here, she smiles during the 2019 Berlin Marathon.

Jaime Recht is racing against time, in more ways than one.

Deaf and partially blind, the 56-year-old from Chevy Chase, Md., is running the Boston Marathon April 18, against a field of 30,000, and against her best time of 4:52.30.

She's also running against darkness, a gradual loss of vision due to a genetic disorder called Usher Syndrome Type 1F, the leading cause of inherited deaf-blindness among Jews.

"Usher Syndrome is a deteriorating condition, which eventually will lead to full blindness ... I am aware my vision is slowly dwindling," said Recht, who is of Eastern European Jewish descent. "I am quite hopeful that a cure or treatment will be found in time to help save my vision loss."

Recht is running Boston to raise awareness of the disease and support for the Usher 1F Collaborative, a Massachusetts nonprofit that funds research. The collaborative was founded by Melissa Chaikof, a Newton mother of two daughters with Usher 1F, who will be among Recht's cheering fans.

"Jaime is amazing," said Chaikof. "She's lost significant vision but has more than is typical because aerobic exercise can slow vision loss. She'll run Boston with sighted guides, but she runs by herself in her home area. Limited peripheral vision just doesn't stop her."

Usher is one of several genetic mutations – Gaucher disease, Tay-Sachs, and cystic fibrosis the best-known – common among Ashkenazi Jews. Three subtype Usher mutations (1, 2, and 3) disable the inner ear and retina. About 400,000 people worldwide suffer from Usher, the vast majority with types 1 or 2. Most severe is Type 1: children are born profoundly deaf with balance problems and begin to lose vision in early childhood. Cochlear implants can mitigate deafness, but no cure yet exists for Usher-caused blindness or loss of balance.

Chaikof and her husband, Dr. Elliot Chaikof, founded Usher 1F Collaborative in 2013 to fill a research void for the 1F mutation. Their first child, Rachel, and third, Jessica, were deaf as infants, and got cochlear implants. The Usher 1F diagnosis came as Rachel's vision declined just after she graduated high school in 2006. "It was devastating, she fell to pieces initially, the most painful thing I ever lived through," Chaikof recalled.

About the same time Jessica, then 11, started to lose her night vision.

"I got involved in the Usher Syndrome Coalition and began to realize there was no gene therapy research for 1F," Chaikof recalled.

The Chaikof family: (Top) Jessica, Adam, Rachel. (Bottom) Melissa and Elliot.

The Chaikofs were well-qualified to organize Usher 1F research. Melissa holds a master's degree in engineering from Johns Hopkins and worked as a donor research analyst for Nonprofit Leadership LLC. Elliot is chair of the surgery department and surgeon-in-chief at the Beth Israel Deaconess Medical Center. Both Melissa and Elliot are of Ashkenazi Jewish descent and carriers of the Usher 1F mutation.

The Chaikofs initially raised money from friends and family and scouted for potential 1F researchers. In 2015, they were joined by a 1F family from New Jersey with extensive business contacts, and fund-raising picked up. They connected with two researchers exploring 1F: Monte Westerfield at the University of Oregon and Zubair Ahmed at the University of Maryland. In 2017, they organized the first 1F Research Conference in Boston and opened it to the Harvard Stem Cell Institute. One attendee was Dr. David Corey, a neurobiologist at Harvard Medical School.

By this time, both Chaikof daughters had adapted to their 1F limitations and were thriving as adults – even as they faced futures with loss of vision.

“Both of my girls were there and David was moved when he spoke to them,” Chaikof recalled. “He came to my husband a week later and said he wanted to take on 1F.”

Today, Corey is among eight researchers, including Westerfield and Ahmed, funded by the Usher 1F Collaborative. In an email message, Corey said that hearing loss in mice – caused by the same gene mutation found in 1F – has been restored with gene therapy, and that his lab is about to extend the treatment to vision.

“If everything goes well, we might be able to start a clinical trial in 4-5 years, and the FDA might approve a treatment 2-3 years after that,” Corey wrote. “So often in science, research does not go as planned, so it is really hard to say how close a cure actually is. But our success in restoring hearing in mice makes us optimistic that a cure for blindness in Usher 1F patients will come.

“The support of the Usher 1F Collaborative has been invaluable, providing funding for the research when federal agencies thought it could not work.”

Corey is among Recht’s cadre of fans, saying, “That she can run without an intrinsic sense of balance – relying just on her vision and the feeling in her feet – is extraordinary.”

For Recht, her history of Usher 1F combined with her love of running made Boston irresistible. She and a younger sister were born deaf, and her vision receded throughout her childhood in New York City, where she attended deaf schools and mastered sign language. She was 16 when she learned she had Usher 1F, at about the time she took up running. Recht enjoyed an elective running course at Gallaudet University in Washington. D.C., where she earned a business degree and launched her career as a program analyst for the Office of Railroad Safety in the U.S. Department of Transportation. Recht continued to run through adulthood, as she married and raised two sons (now 28 and 24), and as her vision further declined.

“Right now I have approximately 40 percent of tunnel vision, which still enables me to see as long as there’s enough light and contrast. I don’t have any depth perception nor am I able to see in dim light,” Recht said.

“I often train alone on a familiar, flat paved trail during daytime. I need assistance on unfamiliar, curving, and changing terrains, especially uneven surfaces. I’ve managed with the vision I have, though there were a few incidences of bumping into people and cyclists, and tripping.

“The first time I used a tether with a running guide was just five years ago, which has helped me tremendously running in a group in a busy race. It also made running unfamiliar places less stressful for me.” Recht is halfway toward her goal of running the six major world marathons. She already has run Chicago, Berlin, and New York City. London and Tokyo are planned. Boston, she says, is “the most legendary” of the majors. Running Boston has the added incentive of helping fund a cure to save her vision.

“I love to travel, cook unique foods, bake goodies, and spend time with my sons,” said Recht. “It’s my utmost desire to continue enjoying these things with my vision, especially with my future grandchildren.”

To donate to Jaime Recht’s run for the Usher 1F Collaborative, visit www.usher1f.org/usher-1f-stories/jaime-recht.html

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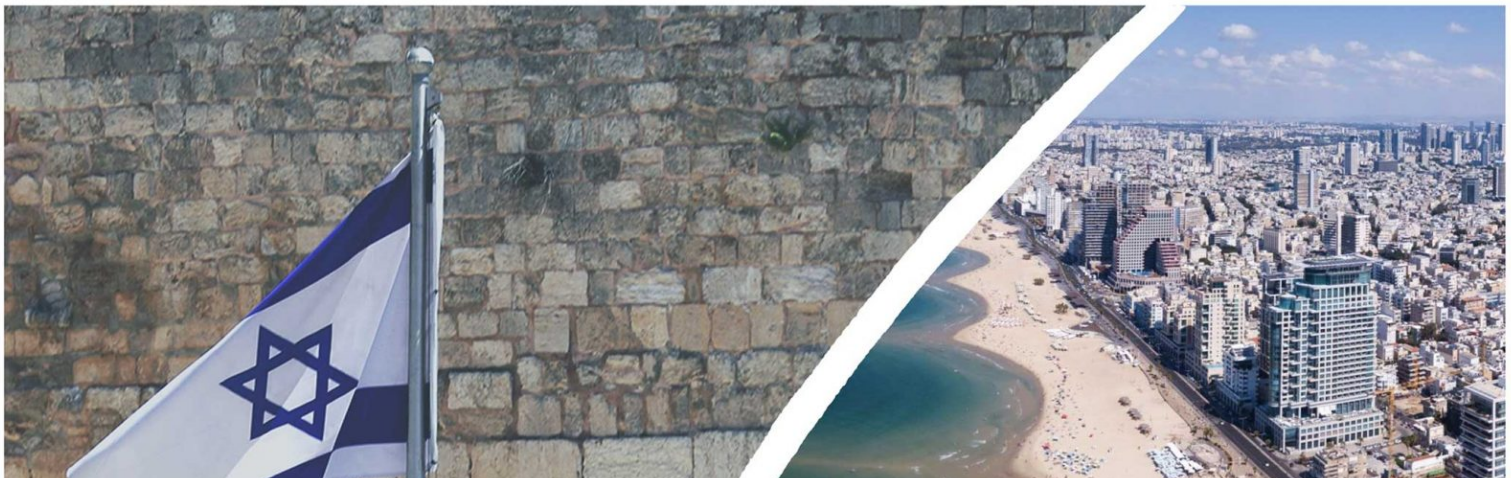

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