

The Boston Globe

A pediatric researcher whose son died of progeria sees promise in new treatment



LEAH FASTEN

Dr. Leslie Gordon with her son, Sam Berns, who died in 2014.

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APRIL 24, 2018

The year before Sam Berns died at 17 of progeria, an ultra-rare fatal genetic disease that causes premature aging, the Foxborough High School student said he hoped his mother, a physician, would one day no longer have to research a cure for the disorder.

“I kind of just want my mom to be done with progeria for her sake, because my mom will keep working forever until progeria is cured,” Berns, the subject of the critically acclaimed 2013 HBO documentary “Life According to Sam,” said in an interview on ABC that year.

A professor of pediatric medicine, Dr. Leslie Gordon hasn’t found a cure, but she’s convinced that she and fellow researchers have hit on a promising treatment.

A study published Tuesday in the Journal of the American Medical Association found that children with progeria who received an experimental drug called lonafarnib orally twice a day had a lower mortality rate than those who got no treatment.

One in 27 kids who took the drug died after slightly more than two years of follow-up, compared with nine in 27 who didn’t get the medicine, according to the study. Gordon was lead author of the article, written by a team of researchers from Boston Children’s Hospital and Brown University.

The children who received the medicine were compared with untreated children of roughly the same age, sex, and continent of residency. All were born in 1991 or later.

Lonafarnib isn’t a cure, Gordon stressed. But the difference in mortality rates — 3.7 percent for children who received the medicine at Boston Children’s Hospital, compared with about 33 percent for children who didn’t — was so stark that she wants the government to approve the drug early, even though it was just a mid-stage clinical trial.



ARAM BOGHOSIAN FOR THE BOSTON GLOBE

Dr. Leslie Gordon.

“The key here is that lonafarnib has pushed this disease in the right direction,” said Gordon, a 53-year-old professor at Brown University Alpert Medical School and medical director and cofounder of the nonprofit Progeria Research Foundation, which funded the study. “It shows us something we haven’t been able to see before.”

Lonafarnib was originally developed by the pharmaceutical giant Merck & Co. as a potential treatment for cancer, but has since been the subject of multiple studies on progeria funded by the foundation. Merck has licensed the drug to the small company Eiger BioPharmaceuticals Inc., of Palo Alto, Calif.

Typically, an experimental drug must pass three stages of clinical trials, the last one relatively large, before the Food and Drug Administration will consider approval. But Gordon said that progeria is so rare — the foundation estimates no more than 400 children in the world have it — that it would be impossible to do a late-stage trial of any size.

Merck isn’t interested in pursuing FDA approval of the drug for progeria, she said, but the foundation is talking to other drug makers about that possibility. Eiger did not respond to a request for comment.

Progeria occurs in 1 in 20 million children. The disease, which some people believe inspired F. Scott Fitzgerald’s 1922 short story “The Curious Case of

Benjamin Button” and the 2008 movie of the same name, is caused by a genetic mutation. The mutation, discovered in 2003 by a research team that included Gordon, results in an overabundance of the protein progerin.

A buildup of progerin occurs within a cell in normal aging. But the rate of accumulation is highly accelerated in children with progeria, resulting in heart disease, stiff joints, stunted growth, wrinkled skin, and loss of body fat and hair. Children with progeria live an average of 14 years.

Some scientists believe that a breakthrough in progeria research could have implications for the treatment of heart disease and other infirmities of aging in the general population.

“While it’s a long way off, it’s possible that some of the same approaches that aim to treat children with progeria could slow the aging process for the rest of us,” said the National Institutes of Health’s director, Francis S. Collins, a physician and geneticist who did early research on a drug similar to lonafarnib in mice with progeria.

Gordon had never heard of progeria when Sam, her only child, was diagnosed with it at 22 months. She has devoted her life to finding a treatment. She gave up a residency as a pediatric ophthalmologist and helped to found the Progeria Research Foundation with her husband, Dr. Scott Berns, president and chief executive of the National Institute for Children’s Health Quality, a nonprofit group in Boston.

Sam’s determination not to let progeria define his life inspired his parents and countless others.

An avid sports fan, he played the snare drum in the Foxborough High School marching band. He was befriended by New England Patriots owner Robert Kraft, who said he was so impressed with the teenager that he helped raise over \$1 million for progeria research. Sam gained renown as a motivational speaker with a widely circulated TEDx talk and won the hearts of many in the documentary “Life According to Sam.”

“I didn’t put myself in front of you to have you feel bad for me,” he says at the beginning of the film. “I put myself in front of you to let you know you don’t need to feel bad for me. I want you to know me. This is my life, and progeria is part of it. It’s not a major part of it, but it is part of it.”

Gordon said, “He dealt with it so graciously and beautifully, I think he taught us how to deal with it, to be honest.”

Gordon, whose foundation is holding a fund-raising event Saturday evening at the Renaissance Boston Waterfront Hotel, said she always told her son she didn’t need a break from researching the disease.

“It’s really not a burden,” she recalled saying. “It’s a privilege.”

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