A. Title

Katie Rogers

BIO 141; Dr. Saiful Islam

“Racing With Sam”

Robin Marantz Heinig

The New York Times
B. Introduction

The article, “Racing With Sam,” is about a boy with a disease called progeria. Progeria is a very rare accelerated aging disease that causes children’s bodies to become aged and frail at very young ages, and eventually causes their death by heart attack or stroke in their early teens. As sad as that is, this article highlights a family that is attempting to do something about the disease by bringing attention to progeria. Doctors Leslie Gordon and Scott Berns, Sam’s parents, have dedicated their lives to the research of progeria. Dr. Gordon even founded the Progeria Research Foundation to provide information for parents, as well as keep the public abreast of any new research about progeria.

There have been large advances in progeria research, such as the mapping of the progeria gene. It was found on Chromosome 1, and involves the switching of one nitrogenous base with another. There has even been evidence that the research for progeria could be vital in decoding the secrets in normal human aging. Even with all of the promising advances and research, progeria is still one hundred percent fatal. Gordon continues to push for testing with new technologies on these children and remains involved with the Progeria Research Foundation.

C. Library Research

Progeria (specifically Hutchinson-Gilford progeria syndrome) is a disease characterized by accelerated aging in children. It is exceedingly rare, only occurring in about 1 in 4 million children. These children are normal in appearance at birth, and it is not until symptoms appear at about 6 months of age to a year that a diagnosis is made (Travis, 2003).
Symptoms of progeria include slow growth; large face with protruding eyes; translucent, thin skin; hair loss; osteoporosis; and low body fat, along with other common signs of aging (Hegele, 2003). The most important symptom associated with progeria is atherosclerosis, which is often the cause of death for these children (Juncosa, 2008). Something important to note about progeria is that it does not affect the mental capacity of the children, only their bodies (Travis, 2003).

As recently as 2003, important discoveries have been made about the cause of progeria. Researchers have found that in children with progeria there is a mutation in the LMNA gene on Chromosome 1. This protein is the structural element that forms the nuclear envelope (Travis, 2003). The mutation is a single point mutation (a single base substitution) in one base pair of the approximately 25,000 that make up the LMNA gene, and it occurs in the sperm before conception (National Institutes of Health, 2009). A mutation of the lamin a protein causes a disruption in the nuclear lamina and affects nuclear shape and structure, DNA replication, and gene expression (Rodriguez, et al., 2009). The misshapen nucleus cannot replicate properly causing cell death and accelerated aging. The LMNA mutation in progeria is not inherited; in recent studies, both parents of children affected by progeria are free of the mutation (Hegele, 2003).

Finally, there is no cure for progeria, and it is always fatal, usually by age 13. Previously, the only treatment for the disease was increased caloric intake to increase body fat and low-dose aspirin, angioplasty, and bypass surgery for cardiovascular disease (Juncosa, 2008). Now, researchers are working with a new drug called a farnesyltransferase inhibitor (FTI). FTIs have been effective in reversing cardiovascular disease in old mice and also in reshaping the deformed nucleus (Juncosa, 2008). This is
very promising, as it could lead to the prevention of cell aging and death not only in children with progeria, but also in normal aging adults.

In conclusion, Hutchinson-Gilford progeria syndrome is caused by defective protein that causes a misshapen nucleus. This accounts for cell death and the inability for the cells to replicate properly, thus causing accelerated aging. There is no cure, but a new treatment has shown some promise in treating the cardiovascular disease and repairing the damaged nuclei, and these drugs may someday help slow or reverse the normal process of aging.

D. Conclusions and Impact

I chose this article because when I was younger I saw a special on a child with progeria on The Learning Channel, and I have been keeping up with new research ever since. Also, I find it fascinating that these children could help scientists learn more about aging, and perhaps discover how to slow or reverse the process.

When the Human Genome was successfully mapped in 2003, new and exciting information about progeria began to appear. Since taking Anatomy and Physiology courses I have had a much easier time reading scientific journals. When the journal talks about nuclei being deformed and this causing cell death and difficulty with replication, I understand. Also, since taking science classes, it is clear how important DNA is to the human body, and I know that even a small “mix up” can cause a huge problem, for example, the mutated protein that causes progeria. An understanding of DNA, cell replication, cell death, and basic genetics is essential for understanding progeria, and after having this class and more like it, I feel that I understand the disease much better than I did when I first became interested as a child.
E. Literature Cited


Newspaper Attachment from Internet
Racing With Sam

By ROBIN MARANTZ HENIG

Published: January 30, 2005

At first, Sam Berns looked as odd as all the kids whose pictures I had been studying: big bald head, beaky nose, strangely undersize chin and mouth, blue veins twisting beneath a translucent scalp. He was very short and very skinny, and he had a stiff-hipped waddle when he walked. But after I spent a few minutes with him, the oddness melted away, and I was in the vivid vortex of Sam's personality -- noisy, jokey, smart, imaginative and just a little bit full of himself. The articles I'd read about Sam's disease didn't quite jibe with the bossy little kid who barked, "No shoes in the playroom!" the minute I
walked in the door of his home in Foxboro, Mass., southwest of Boston. Sam has progeria, which is usually described as a grossly accelerated form of aging that turns children into wizened old men and women before their teens. But this child was no wizened old man.

My shoes off, I was allowed to enter Sam's world: a large area just off the kitchen, dominated by a Lego confabulation, as big as a piano, that was part Harry Potter magic kingdom, part Spider-Man lair. He babbled nonstop, his voice permanently high-pitched, as if a 45 r.p.m. record were being played at 78. In every single way, except the way his face and body looked, he was pure 7-year-old boy.

Sam showed me his favorite things, moving quickly from the Yu-Gi-Oh! cards to the Legos because we had only an hour together. In that time, he beat me soundly at air hockey, Lego soccer and a board game. His mother threw some cinnamon muffins in the oven because she knew Sam would need to eat something soon. And, as she predicted, Sam started to flag. But once he gobbled down a muffin, he was all bluster again, attacking the Sorry! game board with spirit and, in between moves, doing his imitation of the Cowardly Lion from "The Wizard of Oz." "If I/were king/of the for-est!" he sang at the top of his lungs, doing a credible Bert Lahr.

Keeping up with Sam Berns is exhausting. He operates at warp speed, cramming three times the normal action into any single hour. It's almost as if Sam knows that he is, quite literally, running out of time.
That familiar sense of life zooming by too fast is what makes progeria so compelling for the rest of us. But a disease that affects no more than 15 or 20 Americans at any one time would barely get any research attention if not for an advocate to promote it. Scientists make choices, and politicians make financing decisions, based on the likely payoff; the more common the disease, the greater the chance that there will be government grants available to support it and scientists who want to do the work. How, then, does a disease this rare become the object of study?

That's where Sam Berns's mother comes in. The woman who offered those warm cinnamon muffins for Sam's breakfast is herself a physician-scientist, and she has devoted the last six and a half years of her life to searching for a cure for her son's disease. Since the day in 1998 when Sam's condition was diagnosed, his mother, Dr. Leslie Gordon, has marched to the cruel allegro of her son's illness, which she knew could kill him by the time he reached his teens. She redirected her career, quitting her internship in pediatrics in order to devote herself full time to progeria. She created a nonprofit organization to promote research into the cause and possible treatments and cures. After spending months reading scientific literature, she eventually talked her way into the laboratory of a cell biologist at Tufts University, soon learning enough from him to start her own progeria-research lab. She co-sponsored three major conferences with the help of the National Institutes of Health. She shuttled to Washington to lobby on Capitol Hill, resulting in a legislative coup -- specific mention in the Children's Health Act of 2000 -- that forced N.I.H. to conduct and support progeria research. And in the spring of 2003, she stood alongside two of the nation's leading geneticists to announce the discovery of the progeria gene.
Hands (November 17, 2003)

DISEASE VICTIM DIES AT GATHERING (June 21, 1983)

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