

Progeria: Old Before Their Time

Hayley Okines is like any other five-year old little girl; she enjoys school, likes playing with jewelry and costumes, and loves the fact that she has her very own television in her room, wrote Rachael Murphy in *The Daily Mirror*. Although Hayley is like her friends in so many ways, there are many physical features that set her aside from her friends: she suffers from hair loss, joint stiffness, thin and wrinkled skin, dwarfism, a small, skinny nose, many dislocated hips, and late formation of teeth. In addition, she has to worry about heart attacks, strokes, and even death (Murphy 16). Yes, these conditions sound like they should be facing sixty or seventy year olds; but instead, a little five-year-old girl and other children throughout the world are suffering from these conditions every day. This rare, but very fatal disease, is known as Hutchinson-Gilford Progeria Syndrome, or Progeria.

According to Erica Blake from *The Blade*, progeria is known as the "accelerated aging" disease because it results in children aging eight to ten times faster than the normal rate (A.1). Progeria patients typically die of heart attacks or strokes by age thirteen because of cardiovascular disease, reports Jennifer Bails in the *Pittsburgh Tribune*. Progeria is a devastating disease because it is always fatal to its victims. However, it is quite fortunate that it only affects one in eight million babies worldwide. That means that there are only about 40-43 children with progeria in the world; and in the United States alone, there are only twelve reported cases of the disease to date (Blake A.1). Although records of progeria have only existed for about one hundred years, researchers have a pretty good understanding of the disease (Murphy 16). Scientists are aware of the characteristics associated with progeria, the gene that may cause it, and also the future for progeria patients.

Many terrible physical characteristics go along with progeria. Usually before age two, parents begin to notice that something is wrong with their child because the odd characteristics of progeria begin to appear. For example, parents notice strange looking skin on their child, usually on the stomach, and also little hair growth. However, the most obvious warning sign maybe the lack of weight and height growth. Most progeria patients barely even double their birth weight by age one. Even as the progeria victims grow older, they continue to struggle with growth and are actually considered to have dwarfism. A few other physical characteristics progeria patients have include, a pinched nose, a large head with a small face and jaw, and late formation of teeth. Not only are these physical traits a part of progeria, but there are also some physical disabilities involved, such as joint stiffness, hip dislocation, and heart complications (Murphy 16).

Researchers have not only identified the physical character traits of progeria, but they have also done many studies on what actually causes the disease.

After extensive research and several studies, scientists believe to have found what exactly causes progeria. A gene called lamin A was discovered by scientists in 2003 to be the cause of this terrible disease. The lamin A gene is responsible for building proteins in the lamina of a cell. The lamina is located right inside the cell membrane near the nucleus. When there is a mutation in this gene, protein builds up in the lamina. As a result of the lamin A mutation, scientists first believed the network of proteins became more fragile. However, later, scientists at the University of Pennsylvania and Johns Hopkins University in Baltimore found that the network of proteins did not become more fragile, but actually stiffer. The stiffness results from all of the extra proteins trying to fit into the lamina shell. The stiff shell isn't good because there is a greater chance of the lamina breaking when it's under a lot of pressure. The researchers then concluded that the change in the stiffness of the lamina shell may affect how it controls gene function and also the way the nucleus reacts when mechanical forces are present (Bails). These resulting changes are believed to be what leads to the accelerated aging process.

Another astounding find relating to the cause of progeria is that when scientists studied the structure of the nucleus in older people, it was very similar to the structure of the nucleus in progeria patients. This find leads researchers to believe that progeria is closely linked to the normal aging process. Professor Kris Noel Dahl said that she hopes to learn more about other "nuclear-related" disorders and also the normal aging process through all of the studies being conducted on progeria. (qtd. in Bails).

After discovering the gene that caused progeria, scientists began searching for cures and treatments that would aid progeria patients.

One young medical student discovered the first possible treatment that may help to better the future for progeria patients. Craig Spychalla declared in the Wisconsin State Journal that in July of 2004, less than one year after the lamin A gene was discovered to be the cause of progeria, a New York University medical student, named Brian Capell, began to study the lamin A gene. Immediately, Capell noticed the significant likeness between progeria and normal aging. Using a drug that was first used to kill off cancer-causing proteins, Capell tried to treat the cells infected by progeria. First, the drug forced the progeria cells to stay the same shape, whereas without the drug, the cells usually change shape. After discovering that the anti-cancer drug helped to stop some of the stages of progeria, such as the cell shape changing, Capell posted his findings on an online publication. The next thing scientists planned to do was to try the drug on mice, and then hopefully do a clinical trial on children (Spychalla). Dr. Leslie Gordon, a clinical researcher, and her husband, Dr.

Scott Berns, a pediatrician, helped to jump start the process of organizing the first clinical trial on children with progeria.

Wasting no time, Dr. Gordon quickly got started on planning a future clinical trial. Amy Dockser Marcus explains in The Wall Street Journal that Dr. Leslie Gordon and her husband were so eager to start the process of a clinical trial with progeria patients because their own son, Sampson, was infected with progeria. Dr. Gordon began to learn about all the other children in the world with progeria by studying each child's case. She continued to push for a clinical trial to be done, and finally doctors said that they hoped to start enrolling children with progeria in the trial in March of 2007. The trial would consist of children flying from all over the world to a hospital in Boston every four months to see if an experimental drug would help with their disease. To prepare for the trial, Dr. Gordon answered many questions of the other families that would participate in the trial. She also translated many medical documents into the language of each participant and then asked parents for blood samples or baby teeth of their children, so researchers could begin studying them. However, as Dr. Gordon worked on finding enough children with progeria to participate in the trial, she knew she was short on time and also the money needed for the trial. It was also difficult to do the clinical trial because there are so few children with progeria in the world. Dr. Gordon figured that 25 of the 42 children with progeria would need to participate in the clinical trial in order for it to work appropriately. One other concern was that the trial would take at least two years, and most of the children participating were between eight and ten years of age, so they would have to work fast. Also, the last concern was raising the two million dollars needed to complete the trial.

Dr. Gordon worked long and hard on raising funds for the trial because she knew it would brighten the future for progeria patients. She also ordered thirty scales, one for each family, to allow the parents of each child in the trial to chart the weight growth of their child while taking the experiment drug (Marcus A.1) Although the trial is not quite ready to begin because of too few participants, Dr. Leslie Gordon continues to be involved with the families dealing with progeria because she is the founder of the Progeria Research Foundation.

As the founder of the Progeria Research Foundation, Dr. Gordon acts as a major support system for the families by receiving many e-mails and letters and also answering any questions they might have. According to Marcus' Wall Street Journal article, the Progeria Research Foundation was founded in

1999 by Dr. Gordon, her husband, and also her sister, Audrey Gordon. Their goal was to somehow unite the families dealing with progeria. Because the families were located all over the world, the foundation created a website with an online message system for the families to communicate on. It also gave links to personal web pages that some families had made for their child with progeria (A.1). The Progeria Research Foundation is a great source for families dealing with progeria; however, it is not the only way to receive help and support.

There are other organizations out there that deal with children with chronic illnesses or diseases, and there are also many books to help educate families and progeria patients about their future. One great book is titled *How to Get Quality Care for a Child with Special Health Needs*, written by Georgianna Larson and Judith A. Kahn. The book gives tips on talking to health care teams involved with a child's case, on paying for all of the health care needs, and on finding additional support. One book they suggest is actually written by Georgianna Larson and is titled *Managing the School Age Child with a Chronic Health Condition*. Another place doctors and therapists suggest to go for further information on the disease is the National Organization for Rare Diseases located in New Fairfield, Connecticut (69). All of these sources offer great help, guidance, support, and information needed to deal with everything that comes with progeria.

Progeria is a tough disease to live with because children with progeria, like Hayley Okines, are just like every other child their age, except they have to suffer from physical traits and health issues that people ten times their age have to deal with. While progeria patients suffer from lack of growth, hair, and teeth, they also have to worry about their health. They are usually struck by death at very young ages. Although progeria is a tragic disease, it is fortunate that there are only about 43 known cases of it in the world (Blake A.1). Also, through extensive research and many studies, scientists have found the gene that causes progeria and also some drugs that may help to slow down the disease by inhibiting certain proteins.

Though there has not yet been a clinical trial done on children, families of progeria children and scientists have continued to make advances toward one. Through help from the Progeria Research Foundation, other families dealing with the disease, and many books and organizations, parents of progeria patients can hope to one day find a cure that will allow their child to live like a child.

Works Cited

- Bails, Jennifer. "Findings May Help Treat Aging Disease." Pittsburgh Tribune 1 July 2006. ProQuest. Western Technical College Library Web Database Access. 22 Mar. 2007 <<http://proquest.umi.com/>>.
- Blake, Erica. "Tot with Rare Disease Races Clock." The Blade 25 Sept. 2006. ProQuest. Western Technical College Library Web Database Access. 20 Mar. 2007 <<http://proquest.umi.com/>>.
- Marcus, Amy Dockser. "Aging Fast." The Wall Street Journal 29 Jan. 2007. ProQuest. Western Technical College Library Web Database Access. 20 Mar. 2007 <<http://proquest.umi.com/>>.
- Murphy, Rachael. "Hayley's Heartbreak." The Daily Mirror 22 Mar. 2003. ProQuest. Western Technical College Library Web Database Access. 22 Mar. 2007 <<http://proquest.umi.com/>>.

Spychalla, Craig. "Anti-cancer Drug May be Key to Progeria Cure." Wisconsin State Journal 21 Sept. 2005.

ProQuest. Western Technical College Library Web Database Access. 22 Mar. 2007

<<http://proquest.umi.com/>>.

April 25, 2007