INCREASING AWARENESS ABOUT PROGERIA

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Because of its rarity, I am sure that not too many people have heard of the condition Progeria. It is a very uncommon syndrome (condition involving multiple bodily systems) affecting one in every four million children. Not only is this disease rare, but it is also fatal, as the affected person ages very prematurely and excessively fast. One year of life with Progeria is equivalent to about seven to eight years of normal aging, so an eight year old child with this condition actually possesses the physical appearance and physiological functioning, especially that of the cardiovascular system, of a fifty six year old.

The different parts of the body that manifest rapid aging are the skin (including its appendages such as the hair and nails), the bones, the ears/hearing, and the blood vessels of the heart and brain. When they are born, children with Progeria look like any normal babies, with no obvious signs of aging. But around one and a half years, they start showing changes not usually found in toddlers. The veins on the forehead/scalp and abdomen suddenly become very prominent, lips can appear bluish, **and their skin covered with a mixture of over or under pigmentation.** They start losing their hair but with distinctive preservation of the mid scalp. They also experience loss of their eyebrows and eyelashes. **Their growth is stunted at a certain point and they start losing weight incessantly.** They are usually lagging behind their peers by 3-4 years in height and the average weight of the known affected children is about 10.6 kilograms only.

Most Progeria patients share many common physical characteristics that make them easy to distinguish from other syndromes. Their skin becomes extremely wrinkly, dry, thick and tight. Their eyes also suffer from severe dryness necessitating the use of "artificial eye drops." Their bones become osteoporotic or brittle making them prone to fractures. Aside from that, they also have bony abnormalities and deformities such as big heads coupled with narrow jaws and small chins, so their teeth tend to crowd and overlap in their small mouths. Their noses get thin and usually with a beak-like deformity. The ends of their fingers somehow get fixed at a bent position with shortened nails. Their shoulders are very narrow, and hips are deformed resulting in bow-leggedness. Very early on, they develop arthritis with complaints of joint pains making activities of daily living like walking and enjoying sports more difficult. **Their voices are characteristically high pitched and shrill (I believe an advantage if they love to sing because this enables them reach the highest notes).** Lastly, their hearing diminishes just like our **grandparents',** sometimes even requiring them to use hearing aids.

The most common causes of deaths among Progeria kids are either heart attack (myocardial infarction) or brain attack (stroke/cardiovascular accident). These are brought about by the hardening (atherosclerosis) of the blood vessels that bring oxygen to the heart and brain tissues. They become so stiff to the point at which the heart and

brain are no longer able to receive oxygen, causing all functions to cease. On average, the life span of a child with Progeria is between 13 and15 years old.

It seems like every organ gets old too fast, but this is certainly not the case with their intellect. God has blessed these kids with normal thinking appropriate to their chronological age. They are able to attend regular school, achieve high marks, establish friendship, and even have colorful love lives. Their emotions are just as similar to the kids their age. They can feel sad, happy, angry, ecstatic, depressed or hopeful. **Just like other kids, the girls love to accessorize to look pretty and fashionable, and the boys groom up to appear handsome to their crushes.**

So, what really went wrong with them? Through the leadership of Dr. Leslie Gordon and several other doctors, and their unparalleled dedication of several years of studies, the genetic mutation that leads to the development of this syndrome has been found. It is the PROGERIN, a defective structural nuclear membrane protein, that causes the collapse of the nucleus, that essentially leads to cellular death/dysfunction. Progerin is present in almost all cell types in the body, which is the reason why different body systems/functions are affected. This gene mutation is something not inherited or familial, but rather sporadic, meaning that it happens randomly, although the risk of having another child with Progeria increases slightly.

Knowing the earliest symptoms of Progeria is very important, as it will aid medical treatment geared towards extending the lifespan of these Progeria children by at least 1.6 years. The Progeria Research Foundation at Boston, MA, joined by other hospitals/learning institutions (Hasbro Children's Hospital, Dana-Farber Cancer Institute, Beth Israel Deaconess Medical Center, Harvard Medical School, Warren Alpert Medical School of Brown University) has proven the positive effects of the medicine Lonafarnib, of the farnesyltransferase inhibitor group, and Everolimus (an antineoplastic chemotherapy medicine) **in slowing down or retarding the rate of the aging process especially in the areas of weight gain and cardiovascular system.** Further studies, especially the Everolimus, will be necessary to potentiate the effects of these medications, which can also possibly be used as one of the anti-aging treatments for regular adults. Wouldn't that be exciting?

Our foundation has partnered with Progeria Research Foundation in trying to locate possibly 150 more Progeria children around the world. From the Philippines, Kim (13), Jess (12) and Nezha (6) (siblings), and Osang (8), our honorary angels, are able to avail of this medication which can add at least a couple of years to their lives. (The first ever Filipino child officially diagnosed with progeria, Rochelle, had her life prolonged up to 18 years with the help of Lonafarnib, until she succumbed to the cardiovascular complications of the advanced aging. Our president Jojo Sayson, and yours truly, accompany these tiny blessed angels to Boston Children's Hospital when they come to the USA. Every two years, they have to come back to Boston for re-evaluation and documentation of the effects of the trial medications (Lorafarnib and Everolimus) on their condition. We are encouraging you to help spread awareness about Progeria to enable these kids benefit from these medications through the generosity of PRF and its donors.