Hutchinson – Gilford Progeria

Student Name

Medical Careers Institute, School of Health Science, ECPI College of Technology
HUTCHINSON – GUILFORD PROGERIA

Hutchinson – Gilford Progeria

DESCRIPTION

Progeria is a rare genetic disease characterized by dramatic premature aging disorder. The disorder derives its name from the Greek word "geras," and means “prematurely old.” Hutchinson-Gilford Progeria Syndrome (HGPS), the most severe form of Progeria, was described by two different British doctors; Dr. Jonathan Hutchinson in 1886 and at a later time, Dr. Hastings Gilford. The condition is estimated to affect one in four to eight million male and female newborns worldwide (Progeria Research Foundation, 2004).

SYMPTOMS AND SIGNS

According to the Progeria Research Foundation (2004), “although they are born looking healthy, children with HGPS begin to display many characteristics of accelerated aging at around 18-24 months of age.” Their growth rate begins to decrease around this time and they will be shorter and weigh much less than others their age. W. Ted Brown, M.D., Ph.D., regarded as the world's leading clinical expert on Progeria said (as cited by Spencer, 2003), "Many people consider Progeria to be the most dramatic example of a genetic disease that clearly resembles accelerated aging. The children appear to have an aging rate that is five to ten times what is normal" (Spencer, 2003). For example, a child around the age of ten years is living in a body approximately equivalent to that of an octogenarian (K. Okines & M. Okines, 2008).

Children with Progeria exhibit no signs of mental deficiency and, relative to the average population, are of normal intelligence. Affected children all have a similar appearance without regard to ethnicity. Characteristic signs and symptoms include: loss of body fat and hair (including eyelids and eyebrows), aged-looking skin, pinched nose, a small lower jaw (micrognathia), high-
HUTCHINSON – GUILFORD PROGERIA

pitched voice, prominent scalp veins, prominent eyes, large head for size of face (macrocephaly) with open soft spot (fontanelle), limited range of motion, and delayed or absent tooth formation (Kirmse, 2007).

Commonalities with aged people are “stiffness of joints, hip dislocation, generalized atherosclerosis, cardiovascular (heart) disease and stroke” (Progeria Research Foundation, 2008). It is common for children with Progeria to have hypertension, angina, enlarged heart, and heart failure; all conditions associated with aging. Atherosclerosis, leading to heart attack or stroke, is the leading cause of death in children with Progeria. The average patient survives to the early teens, and most will die within eight to twenty-one years (Progeria Research Foundation, 2004).

ETIOLOGY

As stated in the National Institutes of Health Progeria Fact Sheet, “thirty years ago, virtually nothing was known about Progeria, and due to the rarity of the disease, little research was done until the 1990s.” It is now known that HGPS is “caused by mutations in the gene which encodes the nuclear matrix protein lamina A” (Decker, Chavez, Vulto & Lansdorp, 2009), the protein necessary to hold the nucleus of a cell together. The abnormal protein was found to be “missing a stretch of 50 amino acids near one of its ends [and] destabilizing the nuclear membrane accelerating the aging process in a way that may be particularly harmful to tissues routinely subjected to intense physical force, such as the cardiovascular and musculoskeletal systems” (Spencer, 2003). Progeria is not a hereditary or communicable disease “rather the gene change is a chance occurrence that researchers believe affects a single sperm or egg just before conception. Neither parent is a carrier, so the mutations in the children's genes are new” (Mayo Clinic Staff, 2009a).
HUTCHINSON – GUILFORD PROGERIA

DIAGNOSIS

“A genetic test for Hutchinson-Gilford Progeria syndrome is currently available” (NIH-Progeria, p.2). After an initial clinical evaluation, a sample of the child’s blood will be tested for the Progeria gene. This test can detect mutations in Lamin A. “This genetic test now enables doctors to diagnose a child at a younger age and initiate treatment early in the disease process” (NIH-Progeria, p.2).

TREATMENT

At this time, there is no treatment for HGPS, however, hope may be on the horizon with the recently discovered Farnesyltransferase inhibitors or FTIs (*see Prognosis). FTIs were originally developed for treating cancer, but laboratory studies have shown promise in correcting the cell defects that cause Progeria. The best that can be done until the drug is approved is to treat the complications of Progeria (Purdy, 2007). Like aging adults, tendon problems can cause joint contracture in the ankles, knees, and fingers, limiting range of motion. Hydrotherapy (swimming in warm water) can promote relaxation, relieve pain, assist movement and enable exercise. “It can also help prevent arthritis from getting worse” (K. Okines & M. Okines, 2008). In order to guarantee the highest range of motion and physical healthiness possible, children with Progeria need physical and occupational therapy up to three times per week. Examples for strengthening the hips and abdominal muscles are activities such as sit-ups, bridges, and leg lifts. It is encouraged for those with Progeria to participate in Physical Education class and swimming activities (Progeria Research Foundation, 2004).
“Some children with Progeria have undergone coronary artery bypass surgery and/or angioplasty in attempts to ease the life-threatening cardiovascular complications caused by progressive atherosclerosis” (Spencer, 2003).

PROGNOSIS

At this time there is no cure for Progeria. The average patient survives to the early teens, and most will die due to atherosclerosis leading to heart attack or stroke. There is, however, a drug in the final stages of testing which may provide some hope for Progeria sufferers. According to reports from the Progeria Research Foundation (2004):

“Researchers have now identified a potential drug treatment for children with Progeria, called farnesyltransferase inhibitors (FTIs). [which show promise in correcting the cell defects that cause Progeria]. The Progeria clinical drug trial began on May 7th, 2007 with two children arriving in Boston, MA for their first of seven visits over a 2-year period. At this first visit, they were given extensive tests and their first doses of the drug. An average of two families have been flying to Boston each week since then, and in October 2007, the trial became fully enrolled. The trial is expected to end in October 2009, with results published in 2010” (Progeria Research Foundation, 2004).

PATIENT TEACHING

Families with Progeria children, as well as the children themselves, quickly come face to face with complex challenges. Support groups consisting of other families going through similar predicaments are available to assist in coping with the ordeals surrounding Progeria. These groups share the information needed to understand and manage the disease as well as offering much needed emotional support. The internet and local health departments are good sources of information to locate self-help groups or therapists offering counseling. Reunions for Progeria
children, as well, are helpful and therapeutic (Mayo Clinic Staff, 2009b). Some children with Progeria have their own web pages to assist in educating others about their disease. One such page is Hayley’s Progeria Page which offers not only the details of the life of a child with Hutchinson-Gilford Progeria, but information for all those searching for hope.
References


