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Review Article

Hutchinson-Gilford Syndrome/Progeria

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ABSTRACT

Progeria is a rare autosomal dominant genetic disorder, Condition of childhood with striking features resembling premature aging. It was first described in 1886 by Jonathan Hutchinson. It was also first described independently in 1897 by Hastings Gilford, The condition was later named by their discoverer - Hutchinson-Gilford Progeria Syndrome (HGPS). The condition is extremely rare affecting around 1 in every 4 million children. Children with progeria usually die of heart attacks or strokes. Children with progeria appear normal at birth but, during the first year, signs and symptoms, such as slow growth and hair loss, begin to appear. Disease is incurable, but symptoms can be managed.

Keywords: Autosomal, Premature, HGPS, Strokes.

INTRODUCTION

The word *progeria* comes from the Greek words "pro" (πρό), meaning "before" or "premature", and "gēras", meaning "old age" Progeria is a rare genetic condition that causes a person to age prematurely. Children with progeria appear healthy, but by the age of 2 years, they look as if they have become old too fast. It is caused by a mutation in the lamin A (LMNA) gene, and it involves severe hardening of the arteries from a young age. This condition greatly increases the chances of having a heart attack or stroke at a young age. These serious complications can worsen over time and are life-threatening for affected individuals. Death occurs at a mean age of 14.6 years.

CAUSES

The mutation occurs in the LMNA gene lamin A, a protein known as progerin that holds the nucleus of the cell together. The defective protein is thought to make the nucleus unstable. This instability makes cells more likely to die younger, leading to the symptoms of progeria.

Person with Progeria might have Symptoms of:

Babies with progeria do not grow or gain weight normally, they develop physical traits including:

- A bigger head
- Large eyes
- A thin nose with a "beaked" tip
- Ears that stick out
- Slow and abnormal tooth growth
- A high-pitched voice
- Loss of body fat and muscle
- Hair loss, including eyelashes and eyebrows
- Limited growth and short stature
- Loss of hair, including eyelashes and eyebrows
- Early signs of skin aging, including thin skin
- Stiffness in the joints
- Visible veins
- Stroke
- Narrow, wrinkled, or shrunken face
- A small jaw bone
- Limited range of motion and possible

How is progeria diagnosed?

Genetic test can be done to confirm if a child's symptoms are caused by progeria.

TREATMENT

There is no specific treatment for progeria. Aspirin and statin medicines may be used to protect against a heart attack or stroke. There are certain precautions we will take to improve the conditions:

- Eating healthy and regular exercise.
- Joint Stiffness-occupational and physical therapy can help the child keep moving.
- Cardiac surgery to slow the progression of heart disease.
- **Regular monitoring should be done for certain conditions:**
- **Monitoring for heart disease**: This includes regular tests such as echocardiograms and blood pressure.
- **Imaging studies (such as magnetic resonance imaging, or MRI)**: These can be used to watch for strokes, or to check on headaches or seizures.
- **Regular eye exams**: Eye problems, including farsightedness or dry eyes (because their eyelids may not close completely). As the condition advances, they develop cataracts. Some children have a strong sensitivity to light and may be advised to wear sunglasses in some settings.
- **Hearing tests**: Hearing loss that can be improved with hearing aids.
- **Regular dental exams**: Dental problems such as cavities, severe crowding, delayed arrival of teeth and recessed gums.
- **Monitoring for skin problems**: Include dark spots or bulges on the skin, hair loss, itching and skin tightness that restricts motion and can make it difficult to breathe or digest food.
- **Monitoring of bone health**.
- Lifestyle and home remedies
- **Make sure your child stays well-hydrated.** Dehydration can be more serious in children with Progeria.
- **Provide frequent, small meals.**
- **Provide opportunities for regular physical activity. .**
- **Get cushioned shoes or shoe inserts for your child.** The loss of body fat in the feet can cause discomfort.
- **Use sunscreen.** Use a broad-spectrum sunscreen with an SPF of at least 15..
- **Make sure your child is up to date on childhood immunizations.** A child with progeria isn't at increased risk of infection, but like all children, is at risk if exposed to infectious diseases.
- **Provide learning and social opportunities.**
- **Make adaptations.** You may need to make some changes at home that enable your child to have some independence and to be comfortable. These can include household changes so that your child can reach items such as faucets or light switches, clothes with special closures or in special sizes, and extra padding for chairs and beds.

Support Groups

- Progeria Research Foundation, Inc. -- www.progeriaresearch.org.
- Genetic and Rare Diseases (GARD) Information Center

Prognosis

Progeria causes early death. People with the condition most often only live to their teenage years (average lifespan of 14 years). However, some can live into their early 20s. The cause of death is very often related to the heart or a stroke.

CONCLUSION

Hutchinson-Gilford progeria syndrome (HGPS) is an extremely rare, fatal, autosomal dominant segmental premature aging disease, it has no sex, ethnic, or regional predisposition. Morbidity includes failure to thrive, generalized lipodystrophy, alopecia, bone dysplasia, and progressive atherosclerosis leading to cardiac disease and stroke. Neither serum cholesterol nor high-sensitivity C-reactive protein levels are elevated in this

disease. Mortality is caused primarily by heart failure at a mean age of 14 years.

Hutchinson-Gilford progeria syndrome is caused by single base pathogenic variants in the *LMNA* gene that activate a cryptic splice site and result in the production of a farnesylated mutant lamin A protein called progerin Lamin A, Persistent farnesylation of the mutant protein causes it to intercalate into the inner nuclear membrane, where it accumulates and exerts damage to cells as they age.

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