Progeria: Aging in Fast Forward

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Key Points:

- Overview of Progeria
- Symptoms and Characteristics
- Genetic Causes and challenges
- Treatment and Management

Overview

Progeria also called Hutchinson-Gilford progeria syndrome and Hutchinson–Gilford progeria syndrome (HGPS) is a rare genetic disorder characterized by accelerated aging, impaired growth, disrupted lipid metabolism and reduced lifespan.[1]

Progeria is one of several progeroid syndromes. The word progeria comes from the Greek words "pro" meaning "before" or "premature" and "gēras" meaning "old age". Dr. Jonathan Hutchinson and Dr. Hastings Gilford originally described the disease in the late 1800s.

Symptoms

Newborns with the disorder appear to be healthy at birth but usually start to show signs of premature aging during their first one to two years of life. Their growth rate slows and they don't gain weight as expected. Children with the condition have typical intelligence. However, their physical characteristics include:

>Baldness [Hair loss]
>Aged, wrinkled skin
>Disproportionately small face compared to head size
>Prominent Eyes
>A thin beaked nose

Genetic Causes

HGPS is an ultrarare Autosomal dominant Disorder. This means one copy of the mutated gene in each cell is enough to cause the condition. It's a genetic disease caused by progerin, a broadly expressed mutant variant of lamin A protein that accelerates aging and leads to premature death typically in adolescence.

Progerin affects many organs and reproduces many characteristics of physiological aging, with the main cause of death in HGPS being atherosclerotic cardiovascular disease (CVD).[2]

Treatment and Cure

Zokinvy(Lonafarnib) is an approved medication for HGPS; benefits include improved low-tone hearing, decreased headaches, decreased carotid-femoral pulse wave velocity, decreased arterial wall stiffness, and increased life span.[3]

One possible treatment for progeria is farnesyltransferase inhibitors (FTIs). These are currently used for treating cancer, but scientists believe they might reverse the nuclear structure abnormalities that are thought to cause progeria.

Under the partnership of Progeria Research Foundation, National Institutes of Health, Children's Hospital Boston

and Dana-Farber Cancer Institute, the progeria clinical drug trial was initiated in 2010 to test the effectiveness of three 'drugs of hope' – a statin drug called Pravastatin, a bisphosphonate drug called Zoledronic acid and a farnesyltransferase inhibitor called Lonafarnib.[4]

Global Cases

The Progeria Research Foundation (PRF) has identified 203 individuals in 50 countries as of December 31, 2024, including 149 with HGPS and 54 with progeroid laminopathies. Despite these efforts, there are still an estimated 150 to 250 undiagnosed cases worldwide, highlighting the challenges in identifying and diagnosing this rare disease.

Conclusion

Though the condition presents numerous challenges, both physical and emotional, the progress in research provides optimism for future advancements, and ongoing efforts in social awareness and advocacy continue to foster global support. With the support of organizations like the Progeria Research Foundation, a brighter future for those with progeria is on the horizon, making the once bleak prognosis a little more hopeful. Progeria reminds us how precious life is. Science and compassion must go hand in hand to fight this condition.

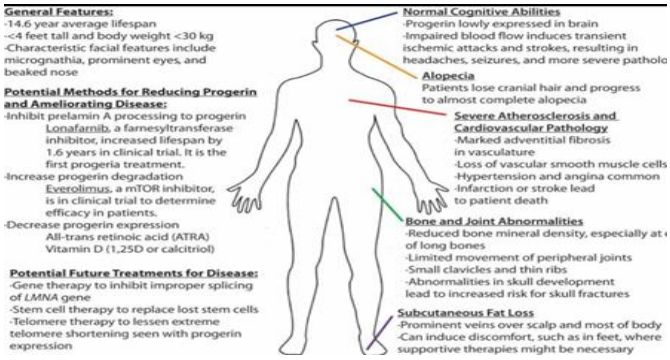

Figure 1 *: Pathophysiology of HGPS*[5]

References

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