National Human Genome Research Institute (NHGRI)

Patented & Patent-pending Technologies Available for Licensing

LMNA Gene and Its Involvement in Hutchinson-Gilford Progeria Syndrome (HPGS) and Arteriosclerosis

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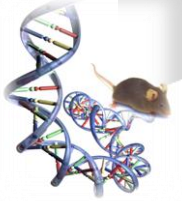

Summary

Children with Hutchinson-Gilford progeria syndrome (HGPS) suffer from dramatic acceleration of certain aging symptoms, mainly cardiovascular disease that generally leads to death from myocardial infarction and/or stroke. In addition to arteriosclerosis, the children exhibit bone deformations and osteoporosis.

Researchers at NHGRI have discovered the genetic basis for HGPS, namely a *de novo* point mutation in the LMNA gene, which encodes Lamin A/C. Lamin is a component of the nuclear lamina, and the discovered mutation results in a shortened form of lamin A, called progerin. Although believed to be a genetic disorder, until the present discovery the mode of inheritance, molecular basis, and pathogenic mechanism of HGPS were unknown.

Potential Commercial Applications

The identification of the mutation and the phenotype of HGPS can lead to further breakthroughs in the detection, diagnosis, prognosis, and treatment of this disease and related conditions. For example, research into arteriosclerosis (e.g., atherosclerosis and vascular calcification) and cellular aging could be further advanced.

Related Article

Eriksson, M. et al., *Recurrent De Novo Point Mutations on Lamin A Cause Hutchinson-Gilford Progeria Syndrome*, 423 NATURE 293 (2003).
http://www.nature.com/nature/journal/v423/n6937/pdf/nature01629.pdf

Key Words

Hutchinson-Gilford Progeria Syndrome, Arteriosclerosis, Aging, LMNA Gene, Lamin A