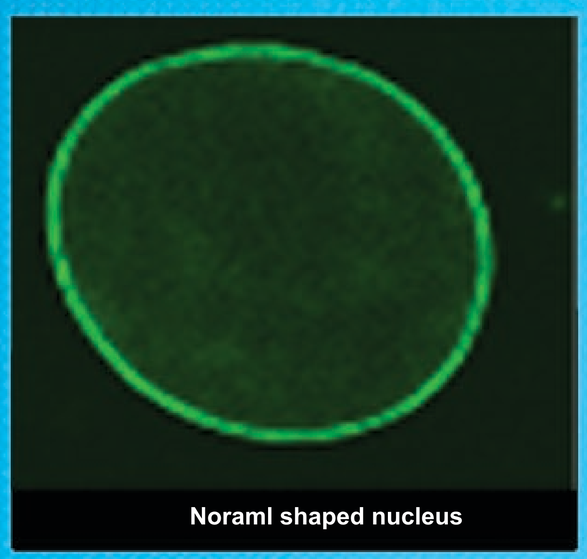
Young But Old

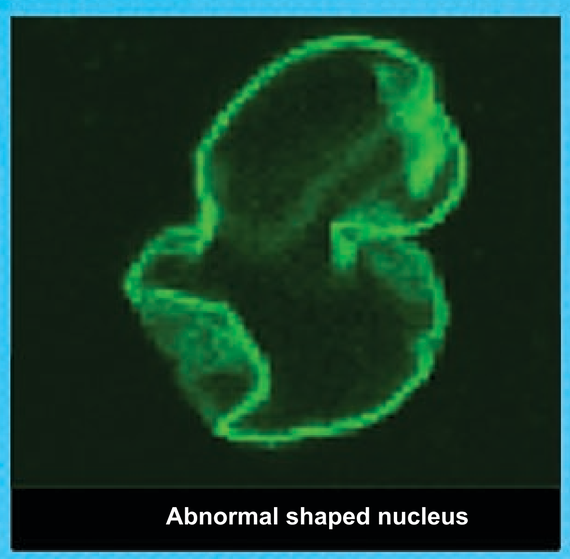
Sadeem Jamal Etolhi, 2nd year medical student
Libyan International Medical University

Introduction

Hutchinson-Gilford Progeria Syndrome also known as "Benjamin Button disease" is an extremely rare genetic disorder affecting around 1 in every 4 million children.(1) It is caused by LMNA gene mutations and expression of “progerin” protein that causes cellular and organismal decline.(3) symptoms resembling aspects of aging are manifested at a very early age. Progeria is one of several progeroid syndromes.(1)



Noraml shaped nucleus

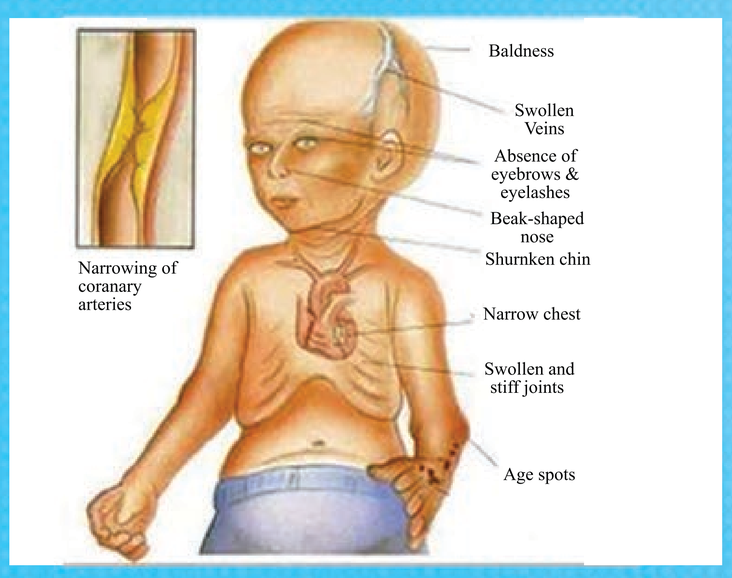


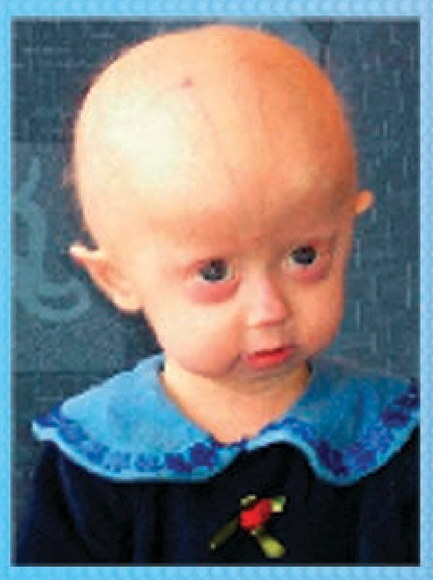
Abnormal shaped nucleus

Clinical features

- Limited growth and short stature.
- Lack of body fat and muscle.
- Loss of hair, including eyelashes and eyebrows.
- Early signs of skin aging, including thin skin.
- Visible veins.
- Narrow, wrinkled, or shrunken face.
- A head that is large compared with the body.
- A small jaw bone.
- Slow and abnormal tooth development.
- Limited range of motion and possible hip dislocation.

In children with progeria, genetic factors increase the risk of developing progressive heart disease from an early age Children with progeria commonly experience cardiovascular events, such as hypertension, or high blood pressure, stroke, angina, an enlarged heart, and heart failure. These conditions are linked to aging.(1)





Discussion

Progeria is a rare genetic condition. Most children with progeria have a mutation on the gene that encodes for lamina.(1) The lamina is a filamentous meshwork beneath the inner nuclear membrane that confers mechanical stability to nuclei. The defective protein is thought to affect the lamin filament assembly and make the nucleus unstable.(2) A newborn with progeria looks healthy, but by the age of between 10 months and 24 months, features of accelerated aging start to appear. Progeria is incurable. But a drug called lonafarnib can extend the average 14 year life expectancy by 1.6 years.(1)

Conclusion

- Progeria refers to a genetic condition in which a child ages rapidly
- Its due to a mutation on the gene that encodes for lamin A
- The disease can lead to fatal heart complications and a heightened risk of stroke Progeria is incurable, but symptoms can be managed.

References:

1.Nordqvist, Christian. "Causes, symptoms, and treatment of progeria." Medical News Today. MediLexicon, Intl., 19 Dec. 2017. Web 27 Jan. 2018. https://www.medicalnewstoday.com/articles/146746.php
2.Apte, Ketaki, Reimer Stick, and Manfred Radmacher. "Mechanics in human fibroblasts and progeria: Lamin A mutation E145K results in stiffening of nuclei." Journal of Molecular Recognition 30.2 (2017)
3.Gonzalo, Susana, Ray Kreienkamp, and Peter Askjaer. "Hutchinson-Gilford Progeria Syndrome: A premature aging disease caused by LMNA gene mutations." Ageing research reviews 33 (2017): 18-29