

Werner Syndrome

Subjects: **Genetics & Heredity**

Contributor: Bruce Ren

Werner syndrome is characterized by the dramatic, rapid appearance of features associated with normal aging.

genetic conditions

1. Introduction

Werner syndrome is characterized by the dramatic, rapid appearance of features associated with normal aging. Individuals with this disorder typically grow and develop normally until they reach puberty. Affected teenagers usually do not have a growth spurt, resulting in short stature. The characteristic aged appearance of individuals with Werner syndrome typically begins to develop when they are in their twenties and includes graying and loss of hair; a hoarse voice; and thin, hardened skin. They may also have a facial appearance described as "bird-like." Many people with Werner syndrome have thin arms and legs and a thick trunk due to abnormal fat deposition.

As Werner syndrome progresses, affected individuals may develop disorders of aging early in life, such as cloudy lenses (cataracts) in both eyes, skin ulcers, type 2 diabetes, diminished fertility, severe hardening of the arteries (atherosclerosis), thinning of the bones (osteoporosis), and some types of cancer. It is not uncommon for affected individuals to develop multiple, rare cancers during their lifetime. People with Werner syndrome usually live into their late forties or early fifties. The most common causes of death are cancer and atherosclerosis.

2. Frequency

Werner syndrome is estimated to affect 1 in 200,000 individuals in the United States. This syndrome occurs more often in Japan, affecting 1 in 20,000 to 1 in 40,000 people.

3. Causes

Mutations in the *WRN* gene cause Werner syndrome. The *WRN* gene provides instructions for producing the Werner protein, which is thought to perform several tasks related to the maintenance and repair of DNA. This protein also assists in the process of copying (replicating) DNA in preparation for cell division. Mutations in the *WRN* gene often lead to the production of an abnormally short, nonfunctional Werner protein. Research suggests that this shortened protein is not transported to the cell's nucleus, where it normally interacts with DNA. Evidence also suggests that the altered protein is broken down more quickly in the cell than the normal Werner protein. Researchers do not fully understand how *WRN* mutations cause the signs and symptoms of Werner syndrome.

Cells with an altered Werner protein may divide more slowly or stop dividing earlier than normal, causing growth problems. Also, the altered protein may allow DNA damage to accumulate, which could impair normal cell activities and cause the health problems associated with this condition.

3.1 The gene associated with Werner syndrome

- WRN

4. Inheritance

Werner syndrome is inherited in an autosomal recessive pattern, which means both copies of the *WRN* gene in each cell have mutations. The parents of an individual with Werner syndrome each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

5. Other Names for This Condition

- Adult premature aging syndrome
- Adult Progeria
- Werner's Syndrome
- Werners Syndrome
- WS

References

1. Huang S, Lee L, Hanson NB, Lenaerts C, Hoehn H, Poot M, Rubin CD, Chen DF, Yang CC, Juch H, Dorn T, Spiegel R, Oral EA, Abid M, Battisti C, Lucci-Cordisco E, Neri G, Steed EH, Kidd A, Isley W, Showalter D, Vittone JL, Konstantinow A, Ring J, Meyer P, Wenger SL, von Herbay A, Wollina U, Schuelke M, Huizenga CR, Leistritz DF, Martin GM, Mian IS, Oshima J. The spectrum of WRN mutations in Werner syndrome patients. *Hum Mutat.* 2006 Jun;27(6):558-67.
2. Kudlow BA, Kennedy BK, Monnat RJ Jr. Werner and Hutchinson-Gilford progeria syndromes: mechanistic basis of human progeroid diseases. *Nat Rev Mol Cell Biol.* 2007 May;8(5):394-404. Review.
3. Lee JW, Harrigan J, Opresko PL, Bohr VA. Pathways and functions of the Werner syndrome protein. *Mech Ageing Dev.* 2005 Jan;126(1):79-86. Review.
4. Monnat RJ Jr, Saintigny Y. Werner syndrome protein--unwinding function to explain disease. *Sci Aging Knowledge Environ.* 2004 Mar 31;2004(13):re3. Review.
5. Opresko PL, Calvo JP, von Kobbe C. Role for the Werner syndrome protein in the promotion of tumor cell growth. *Mech Ageing Dev.* 2007 Jul-Aug;128(7-8):423-36.

6. Opresko PL, Cheng WH, von Kobbe C, Harrigan JA, Bohr VA. Werner syndrome and the function of the Werner protein; what they can teach us about the molecular aging process. *Carcinogenesis*. 2003 May;24(5):791-802. Review.
7. Oshima J, Martin GM, Hisama FM. Werner Syndrome. 2002 Dec 2 [updated 2016 Sep 29]. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJH, Stephens K, Amemiya A, editors. *GeneReviews*® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2020. Available from <http://www.ncbi.nlm.nih.gov/books/NBK1514/>
8. Puzianowska-Kuznicka M, Kuznicki J. Genetic alterations in accelerated ageing syndromes. Do they play a role in natural ageing? *Int J Biochem Cell Biol*. 2005 May;37(5):947-60.

Retrieved from <https://encyclopedia.pub/entry/history/show/12114>