Werner Syndrome

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Overview

What is Werner syndrome

Werner syndrome (also called progeria) is a hereditary condition associated with premature aging and an increased risk of cancer and other diseases. Signs of Werner syndrome usually develop in the teenage years. A person with Werner syndrome does not have the usual growth spurt typical of a teenager and is shorter on average. Signs of aging, including gray hair and hair loss, may appear in the 20s. Cataracts (clouding of the eye?s lens), type 2 diabetes, and osteoporosis (decrease in bone mineral density) may develop in the 30s. One of the most significant health problems faced by people with Werner syndrome is the early development of atherosclerosis, commonly known as hardening of the arteries, which can lead to a heart attack.

What causes Werner syndrome

Werner syndrome is a genetic condition. This means that the risk of Werner syndrome can be passed from generation to generation in a family. Mutations (alterations) in the *WRN* gene are known to cause Werner syndrome. Research is ongoing to learn more about Werner syndrome.

How is Werner syndrome inherited?

Normally, every cell has two copies of each gene: one inherited from the mother and one inherited from the father. Werner syndrome follows an autosomal recessive inheritance pattern, which means that a mutation must be present in both copies of the gene for a person to be affected. This means that both parents must pass on a gene mutation for a child to be affected. A person who has only one copy of the gene mutation is called a carrier. When both parents are carriers of a recessive gene mutation, there is a 25% chance that a child will inherit two mutations and be affected.

How common is Werner syndrome?

Werner syndrome is considered to be very rare. It is estimated that one in 200,000 people in the United States may have Werner syndrome. Werner syndrome is somewhat more common in Japan, where it is estimated that one in 30,000 people may have the condition. This is because a genetic mutation was known to occur many generations ago, when Japan?s population was smaller in number, and over time the mutation has been passed down repeatedly, affecting a higher number of people; this is called a founder mutation.

How is Werner syndrome diagnosed?

Currently, the diagnosis of Werner syndrome is suspected if someone has several of the features listed below.

Common features of Werner syndrome (diagnosed after age 10):

- Cataracts in both eyes (bilateral)
- Skin changes associated with aging, including tight skin, pigments changes, and skin ulcers
- Characteristic facial features, including wrinkling and loss of muscle tone, described as ?bird like? due to loss of fat in underlying skin.
- Short stature (height)
- Early graying or thinning of the hair
- Family history of Werner syndrome

Other features seen in Werner syndrome:

- Type 2 diabetes
- Decreased fertility
- Osteoporosis
- Cancer
- Bone changes in the fingers and toes
- Tissue changes
- Early atherosclerosis (plaque build up in arteries)
- Hoarse or high-pitched voice
- Flat feet

Guidelines for the diagnosis of Werner syndrome have been proposed by the International Registry of Werner Syndrome, which uses clinical and

radiologic findings to establish the diagnosis. Genetic testing for mutations in the *WRN* gene is now available. Mutations in the *WRN* gene are found in about 90% of people with Werner syndrome.

What are the estimated cancer risks associated with Werner syndrome?

The risk of cancer is increased in people who have Werner syndrome, but the specific risk of cancer is unknown. Types of cancers reported in people with Werner syndrome include thyroid cancer [2], melanoma [3], soft tissue sarcoma [4], and osteosarcoma [5] (bone cancer).

What are the screening options for Werner syndrome?

Suggested screenings for people diagnosed with Werner syndrome include:

- Yearly screening for type 2 diabetes
- Yearly lipid profile (blood test)
- Yearly eye examination to look for signs of cataracts (clouding of the eye?s lens)
- Yearly physical and dermatological (skin) examinations to screen for cancers associated with Werner syndrome

People with Werner syndrome should also avoid smoking, be physically active, and maintain a healthy weight to help reduce the risk of heart disease. Any chest pains (also known as angina) should be carefully evaluated by a doctor. Due to the risk of skin cancer, people with Werner syndrome should also limit sun exposure and use skin protection when outside [6].

Screening recommendations may change over time as new technologies are developed and more is learned about Werner syndrome. It is important to talk with your doctor about appropriate screening tests.

Find out more about what to expect when having common tests, procedures, and scans [7].

Questions to ask the doctor

If you are concerned about your risk of cancer, talk with your doctor. Consider asking the following questions:

- What is my risk of cancer?
- What can I do to reduce my risk of cancer?
- What are my options for cancer screening?

If you are concerned about your family history and think you or other family members may have Werner syndrome, consider asking the following questions:

- Does my family history increase my risk of cancer?
- Should I meet with a genetic counselor?
- Should I consider genetic testing [8]?

Additional resources

Guide to Melanoma [3]

Guide to Osteosarcoma [5]

Guide to Skin Cancer [9]

Guide to Soft Tissue Sarcoma [4]

Guide to Thyroid Cancer [2]

What to Expect When Meeting With a Genetic Counselor [10]

National Cancer Institute
www.cancer.gov [11]

American Cancer Society
www.cancer.org [12]

To find a genetic counselor in your area, ask your doctor or visit these websites:

National Society of Genetic Counselors
www.nsgc.org [13]

National Cancer Institute: Cancer Genetics Services Directory
www.cancer.gov/cancertopics/genetics/directory

[14]

Links:
[1] http://www.cancer.net/about-us
[2] http://www.cancer.net/node/19293
[3] http://www.cancer.net/node/19251
[4] http://www.cancer.net/node/19604
[5] http://www.cancer.net/node/19467
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