Cockayne syndrome

Cockayne syndrome is a rare disorder characterized by an abnormally small head size (microcephaly), a failure to gain weight and grow at the expected rate (failure to thrive) leading to very short stature, and delayed development. The signs and symptoms of this condition are usually apparent from infancy, and they worsen over time. Most affected individuals have an increased sensitivity to sunlight (photosensitivity), and in some cases even a small amount of sun exposure can cause a sunburn or blistering of the skin. Other signs and symptoms often include hearing loss, vision loss, severe tooth decay, bone abnormalities, hands and feet that are cold all the time, and changes in the brain that can be seen on brain scans.

People with Cockayne syndrome have a serious reaction to an antibiotic medication called metronidazole. If affected individuals take this medication, it can cause life-threatening liver failure.

Cockayne syndrome is sometimes divided into types I, II, and III based on the severity and age of onset of symptoms. However, the differences between the types are not always clear-cut, and some researchers believe the signs and symptoms reflect a spectrum instead of distinct types. Cockayne syndrome type II is also known as cerebro-oculo-facio-skeletal (COFS) syndrome, and while some researchers consider it to be a separate but similar condition, others classify it as part of the Cockayne syndrome disease spectrum.

Frequency

Cockayne syndrome is estimated to occur in 2 to 3 per million newborns in the United States and Europe.

Causes

Cockayne syndrome can result from mutations in either the *ERCC6* gene (also known as *CSB*) or the *ERCC8* gene (also known as *CSA*). These genes provide instructions for making proteins that are involved in repairing damaged DNA. DNA can be damaged by ultraviolet (UV) rays from the sun and by toxic chemicals, radiation, and unstable molecules called free radicals. Cells are usually able to fix DNA damage before it causes problems. However, in people with Cockayne syndrome, DNA damage is not repaired normally. As errors build up in DNA, cells malfunction and eventually die. The faulty DNA repair underlies photosensitivity in affected individuals, and researchers suspect that it also contributes to the other features of Cockayne syndrome. It is unclear how *ERCC6* or *ERCC8* gene mutations cause all of the varied features of this condition.

Inheritance Pattern

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

Other Names for This Condition

- CS
- dwarfism-retinal atrophy-deafness syndrome

Diagnosis & Management

Genetic Testing Information

- What is genetic testing?
  /primer/testing/genetictesting
- Genetic Testing Registry: Cockayne syndrome
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0009207/
- Genetic Testing Registry: Cockayne syndrome type A
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0751039/
- Genetic Testing Registry: Cockayne syndrome type II
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C2931277/
- Genetic Testing Registry: Cockayne syndrome, type III
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0751037/

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22cockayne+syndrome%22

Other Diagnosis and Management Resources

- GeneReview: Cockayne Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1342
- MedlinePlus Encyclopedia: Failure to Thrive
  https://medlineplus.gov/ency/article/000991.htm

Additional Information & Resources

Health Information from MedlinePlus

- Drug: Metronidazole Oral
  https://medlineplus.gov/druginfo/meds/a689011.html
- Encyclopedia: Failure to Thrive
  https://medlineplus.gov/ency/article/000991.htm

- Encyclopedia: Microcephaly
  https://medlineplus.gov/ency/article/003272.htm
- Health Topic: Growth Disorders
  https://medlineplus.gov/growthdisorders.html

Genetic and Rare Diseases Information Center

- Cockayne syndrome
  https://rarediseases.info.nih.gov/diseases/6122/cockayne-syndrome

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke: Cerebro-Oculo-Facio-Skeletal Syndrome
  https://www.ninds.nih.gov/Disorders/All-Disorders/Cerebro-Oculo-Facio-Skeletal-Syndrome-COFS-Information-Page

Educational Resources

- MalaCards: cockayne syndrome
  https://www.malacards.org/card/cockayne_syndrome
- Orphanet: Cockayne syndrome
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=191
- University of Arizona Health Sciences: Cockayne Syndrome Type A
  https://disorders.eyes.arizona.edu/disorders/cockayne-syndrome-type
- University of Arizona Health Sciences: Cockayne Syndrome Type B
  https://disorders.eyes.arizona.edu/handouts/cockayne-syndrome-type-b

Patient Support and Advocacy Resources

- Amy and Friends: Cockayne Syndrome Support
  https://www.amyandfriends.org/
- Human Growth Foundation
  https://www.hgfound.org/
- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/cockayne-syndrome/
- Share and Care Cockayne Syndrome Network
  http://cockaynesyndrome.org/

Clinical Information from GeneReviews

- Cockayne Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1342

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Cockayne+Syndrome%5BMAJR%5D%29+AND+%28Cockayne+syndrome%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- CEREBROOCULOFACIOSKELETAL SYNDROME 1
  http://omim.org/entry/214150
- COCKAYNE SYNDROME A
  http://omim.org/entry/216400
- COCKAYNE SYNDROME B
  http://omim.org/entry/133540

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