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Visit our website for more information about CS, our family support forum, as well as Share and Care events, donations and fundraising opportunities.

Share & Care Cockayne Syndrome Network is a 501(c)3 non-profit organization

www.cockaynesyndrome.org
Cockayne syndrome (CS) is a rare genetic disorder characterized by poor growth, microcephaly, progeria (premature aging), sensitivity to sunlight, moderate to profound developmental and neurological delays, and a shortened lifespan. CS is inherited in an autosomal recessive pattern. In order for a child to be affected by CS, he or she must inherit a mutation (−) in the same CS gene from both parents. The parents and other “carriers” of a single CS gene mutation remain healthy. Once two parents are known to be carriers, they have a 1 in 4 (25%) chance of having a child with CS.

The symptoms of Cockayne syndrome vary significantly, especially with regard to the age of onset and the rate of progression. The resulting spectrum of severity can be imperfectly divided into three “types” of CS:

- **CS type I** is characterized by normal prenatal growth with the onset of growth and developmental abnormalities around one year of age. The typical lifespan is ten to twenty years of age.
- **CS type II** is characterized by growth failure and other abnormalities at birth, with little or no postnatal neurologic development. The typical lifespan is up to seven years.
- **CS type III** is characterized by a later onset, lesser symptoms, and/or a slower rate of progression. The expected lifespan is unclear, but can extend to forty or fifty years of age.

Some individuals have combined features of both Cockayne syndrome and xeroderma pigmentosum, which is characterized by a wide range of skin changes, from mild freckling to skin cancer on areas exposed to sunlight.

Mutations in the ERCC6 (CSB) or ERCC8 (CSA) genes cause Cockayne syndrome. The ERCC6 and ERCC8 genes provide instructions for making two proteins, called CSB and CSA, which are involved in repairing DNA. If either gene is altered, DNA damage is not as rapidly repaired. As a result, damaged DNA accumulates, which probably leads to impaired cell functions and eventually, cell death. Increased cell death likely contributes to features of Cockayne syndrome such as growth failure and premature aging.

Research and genetic testing for CS is being conducted by Dr. Edward G. Neilan, M.D., Ph.D., Staff Physician, Division of Genetics, Children’s Hospital Boston. Email edward.neilan@childrens.harvard.edu or phone (617) 919-2671.

No specific treatment currently exists for CS. Patients should be treated according to the symptoms they have. Physical, occupational, speech, vision, and hearing therapy are most often beneficial. Visit our website for more information at [www.cockaynesyndrome.org](http://www.cockaynesyndrome.org).