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Cockayne Syndrome: Molecular Mechanism and its Treatment Options

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Perspective

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DESCRIPTION

Cockayne Syndrome is a rare genetic disorder that affects the development and aging of children. It is caused by mutations in genes that control DNA repair, leading to the accumulation of DNA damage over time. This disorder is characterized by growth failure, intellectual disability, and premature aging. Children with Cockayne Syndrome have a small head size, a sunken appearance to their eyes, and a thin nose and lips. They may also have hearing loss, vision problems, and dental abnormalities. Cognitive and motor developments are also significantly impaired, and many children with this disorder are unable to walk or speak.

Genetics

Type A: OMIM 216400 -Gene ERCC8 also called Cockayne Syndrome A (CSA)

Type B: OMIM 133540 -Gene ERCC6 also called Cockayne Syndrome B (CSB)

Type C: OMIM 216411 - Gene none known

Cockayne syndrome is brought on by mutations in the ERCC8 also known as CSA or ERCC6 also known as CSB genes. About 70% of cases involve mutations in the ERCC6 gene. Through the transcription-coupled repair mechanism, the proteins produced by these genes help to repair damaged DNA, particularly the DNA in active genes. UV rays from the sun, radiation, or free radicals in the body all damage DNA. A healthy cell can fix DNA damage before it builds up. DNA damage sustained during transcription isn't repaired if either the ERCC6 or ERCC8 gene is mutated as in Cockayne syndrome, which causes RNA polymerase to stall at that place and interfere with gene expression. Unrepaired DNA damage causes ever more active genes to have their expression blocked, which causes dysfunctional cells or cell death. This may be a factor in the development of the Cockayne Syndrome symptoms of early ageing and neuronal hypomyelination.

Types and mechanism

Cockayne Syndrome is typically classified into three types, with varying degrees of severity. Type I, also known as the classical type, is the most common and severe form. Type II, also known as the severe form, has an earlier onset and more rapid progression than Type I. Type III, also known as the mild form, has a later onset and milder symptoms than the other two types. The underlying mechanism of Cockayne Syndrome involves mutations in genes that control DNA repair, which leads to the accumulation of DNA damage in cells. This results in impaired cellular function and premature aging. Specifically, Cockayne Syndrome is associated with mutations in the ERCC6 and ERCC8 genes, which encode proteins involved in Transcription-Coupled Nucleotide Excision Repair (TC-NER). TC-NER is a mechanism that repairs DNA damage in actively transcribed genes. When this mechanism is impaired, cells are unable to effectively repair DNA damage, leading to the symptoms of Cockayne Syndrome.

Treatment options

Cockayne Syndrome affects the development and aging process of the body. There is no cure for Cockayne Syndrome, and treatment is aimed at managing symptoms. Therapeutic actions include physical therapy, occupational therapy, speech therapy and hearing aids. In addition, some medications may be used to manage specific symptoms, such as seizures. Genetic counseling may also be offered to families affected by Cockayne Syndrome to discuss the risk of passing the condition to future generations. Early detection and management of symptoms can improve the quality of life for individuals with Cockayne Syndrome. With proper care and support, they can lead happy and fulfilling lives. While Cockayne Syndrome highlights the importance of genetic research and the need for continued efforts to better understand and treat genetic disorders.