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Cockayne Syndrome – A Rare Genetic Disorder

Shi Wen Jiang*

Department of Medicine, Center for Genomics, USA

Introduction

Cockayne syndrome (CS) is a disorder marked by cachectic dwarfism, severe neurological manifestations such as microcephaly and cognitive abnormalities, pigmentary retinopathy, cataracts, sensorineural deafness, and mobility and eating difficulties, with death occurring at an average age of 12 years. It's an autosomal recessive condition with a prevalence of 2.5 per million people. There are three phenotypes (1, 2 and 3) as well as complementation groups (CSA and CSB) that overlap with xeroderma pigmentosum (XP). It's been classified as a progeria, and many of the clinical symptoms are similar to those of rapid ageing. CS allows researchers to have a deeper understanding of the fundamental mechanics of ageing. The molecular foundation of CS has long been thought to be transcriptional and transcription-coupled nucleotide excision abnormalities. Edward Cockayne initially reported Cockayne syndrome, also known as Neill-Dingwall syndrome, in a paper titled "Dwarfism with retinal degeneration and deafness" in 1936. He followed up on the same patients ten years later, reporting new clinical characteristics such as increasing hearing loss, visual impairment, and joint contractures. Many clinical aspects of CS are similar to those of normal human ageing. In both CS and normal ageing, cognitive impairment, sensorineural deafness, and high tone hearing loss are common. Furthermore, the advanced atherosclerosis and vasculopathy seen in CS. are similar to those seen in typical elderly people. Premature development of hypertension, which frequently leads to strokes, as well as renal and cardiac failure, are also shared by CS and the ageing phenotype [1-2].

Description

CS is a recessive autosomal condition. CS is divided into two complementation groups: 1) CSA, which is caused by a mutation on ERCC8 on chromosome 5q12–q31, and 2) CSB, which is caused by a mutation on ERCC6 on chromosome 10q11. People with this disease have smaller-than-normal heads (microcephaly), are short in stature (dwarfism), have sunken eyes, and have a "aged" appearance. They frequently have lengthy limbs with joint contractures (inability to relax a muscle at a joint), a bent back (kyphosis), and may be extremely thin (cachetic) due to a loss of subcutaneous fat. Their old appearance is generally due to their small chin, huge ears, and sharp, thin nose. Even in people who do not have XP-CS, hyperpigmentation, varicose or spider veins (telangiectasia), and severe sensitivity to sunlight are typical in people who have Cockayne syndrome. Patients with Cockayne Syndrome frequently burn or blister severely even when exposed to very little heat. Patients' eyes may be affected.

Cataracts and corneal opacity (cloudiness of the cornea) are prevalent. Optic atrophy can result from the loss or injury of the optic nerve's nerves. Involuntary eye movement, or nystagmus, and pupils that do not dilate show a loss of control over voluntary and involuntary muscle action. A salt and pepper pigmentation of the retina is also a common symptom. A specialized test for DNA repair, which evaluates the recovery of RNA following exposure to UV light, is used to make the diagnosis. CS, unlike xeroderma pigmentosum, is not connected with an increased risk of cancer despite being linked to genes involved in nucleotide excision repair (NER).

Conclusion

Although patients can be treated symptomatically, there is no permanent cure for this disease. Physical therapy and minor procedures to the affected organs, such as cataract removal, are commonly used to treat the condition. Because Cockayne Syndrome patients are extremely vulnerable to UV radiation, it is also recommended that they use high-factor sunscreen and protective clothes. Nutritional supplements can also help. Because the condition has a 25% probability of being handed down to future children, genetic counselling for the parents is recommended, as is prenatal testing. Another crucial issue is preventing CS from reoccurring in additional siblings. Because the gene abnormalities involved have been identified, parents who already have one afflicted child can receive genetic counselling and antenatal diagnostic testing [3-5].

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***Address for Correspondence:** Shi Wen Jiang, Department of Medicine, Center for Genomics, USA, E-mail: jiang_s@mercer.edu*

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