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Case report on Cockayene Syndrome

Zahra Fadaeian

Rajiv Gandhi University of Health and Sciences

[1] Department of pharmacy practice, Acharya BM Reddy college of pharmacy

Abstract: **Cockayne syndrome is related to defective DNA transcription and/or repair and belongs to the family of Nucleotide Excision Repair. It is an autosomal recessive multisystemic disorder 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, premature aging (progeria) 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.

Keywords: **cockayne syndrome. Cerebto-Oculo-Facio-Skeletal (COFS), Progeria, Metronidazole.**

1. INTRODUCTION

Cockayne syndrome is a rare inherited disorder in which people are sensitive to sunlight, have short stature, and have the appearance of premature aging. Edward Alfred **Cockayne** (1880-1956), after whom this disease is named, was a London physician who concentrated particularly on hereditary diseases of children. The course and clinical signs of Cockayne's syndrome are distinctive After initial seemingly normal progress the children begin slowly to regress mentally and physically during the second or third year of life. Microcephaly becomes manifest and they remain dwarfed, having disproportionately long limbs and large hands and feet. The skin of the face is wrinkled, suggesting progeria. It may be pigmented and show scars from solar burns. The eyes are deeply sunken, the nose and chin are often prominent, and the ears large. There is loss of subcutaneous fat in the face. Hearing and sight gradually deteriorate. The eyes show optic atrophy and retinal degeneration of the 'pepper and salt' type. The patients become ataxic and may show a 'tottering' gait, athetoid movements, and coarse intention-type tremor. Ankylosis with flexion deformities and kyphosis may set in and the patients are ultimately bed- or chair-ridden. Radiography may show thickened skull bones and scattered cerebral calcification. Bone age remains within normal limits but there may be evidence of osteoporosis. All laboratory tests, and these have been numerous in some of the published cases, have yielded no significant information. In a recently described case Fujimoto, Greene, and Seegmiller (1969) observed hyperlipoproteinaemia, hyperinsulinaemia, and renal insufficiency with acidosis.

2. CASE REPORT

A 12 years old female patient, a product of consanguineous marriage, presented with generalized weakness, short stature, a characteristic facial appearance, premature aging, photosensitivity, progressive neurological dysfunction, and intellectual deficit. inability to do normal and daily work, sensorineural hearing loss and dental anomalies (presence of caries), typical facial appearance includes microcephaly, large ears, butterfly rashes on the face a thin nose, and enophthalmia. Cataracts and cutaneous photosensitivity are observed in this patient Subcutaneous lipoatrophy is present which has led to signs of premature aging of the skin in this patient

As per the information which has been extract from this case the sign of retardation has been started in the first year of age, it was progressive and slow in onset which can protect diagnosis of type one cockayne syndrome or classic Cockayne syndrome, presents in childhood with characteristic facies and somatic features that occur late in the first decade of life.

Child also present with the sever photosensitivity and abnormal sign of aging, urinary incontinence, tremor, progressive hearing lost and difficulty to understand the simple words. Kid also shows difficulty in eating, getting dress and doing simple social and personal activities. the symptoms such as microcephalus and short stature has started to appearing since3 years back and progressed gradually to present extend. On physical examination the blue tint has been observed to the skin (cyanosis) on the arms mostly left arm.

3. DISCUSSION

In summary, a 12-year old girl, a product of consanguineous marriage was diagnosed clinically as a case of Cockayne syndrome because of delayed milestones, spastic paraplegia, dwarfism, ocular fundus changes, typical facies and a photosensitive rash on the butterfly area of the face. A diagnosis of CS was suspected based on the clinical features, neurological, ocular and dermatological signs. CT Scan assisted in the diagnosis, but the definitive diagnosis of CS is by molecular genetic testing or a specific DNA repair assay on fibroblasts [6,8]. Cockayne syndrome has to be differentiated from other conditions having similar clinical features. The treatment for CS is essentially supportive care. Despite the lack of effective treatment and progressive course of the disease, a correct diagnosis is very important to assist the family with the caretaking of the child and genetic counselling should be done to prevent reccurrence of the condition in the family.

According to the general guidelines and standard practical's treatment of cockayene syndrome is mostly symptomatic and supportive includes photoprotection with sunscreen and clothing.

Physiotherapy can help patient with cockayene syndrome to overcome physical disability

4. CONCLUSION

Cockayne's Syndrome is a rare autosomal recessive disorder, first described in 1936. It is characterized by growth retardation, skeletal and retinal abnormalities, neurological defects and mental retardation. A prenatal diagnostic test is available. It has been classified into three clinical subtypes: classical (Type I), severe (Type II) and mild. Classical Cockayne's Syndrome involves growth failure and neurodevelopmental and neurological dysfunction. Type II is early onset severe disease with death usually occurring by the age of six or seven years. In mild Cockayne's Syndrome, early death does not occur and intelligence is normal. The other features of the disease are mild and later in onset.

Affected individuals show postnatal growth failure and few adults exceed 20 kg in weight and 115 cm in height. The term 'Cachectic dwarfism' has been used to convey the greater decrease in weight compared with height. The neurological features include microcephaly, mental retardation, unsteady gait and tremor. Other clinical features include photosensitivity, cataract, optic atrophy, pigmented retinopathy, nystagmus and deafness. Cryptorchidism occurs in 30% of males and oligomenorrhoea, underdeveloped breasts, a small square pelvis and hypoplastic alae of the iliac bones in women.

The diagnosis is made on the clinical features and by fibroblasts showing decreased recovery of RNA synthesis following exposure to ultraviolet light. In normal individuals, ultraviolet light depresses both DNA and RNA synthesis, which returns to normal soon after the exposure stops. In contrast, fibroblast and lymphoblastoid cells from patients with Cockayne's Syndrome show greatly increased sensitivity to the killing affects of ultraviolet radiation, deficient recovery of RNA synthesis and defective repair of transcriptionally active genes. This forms the basis of the laboratory diagnosis and has been attributed to a specific deficiency in the ability to carry out preferential repair of damage in actively transcribed regions of DNA. The primary defect involves components of the excision repair pathway: mutation detection, repair or repair coordination.

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