

# Cockayne syndrome in a seven year old girl

A M S D Kumari<sup>1</sup>, H M P Gunathilake<sup>1</sup>, K L P D Seneviwickrama<sup>2</sup>

*Sri Lanka Journal of Dermatology*, 2021, **22**: 62-63

## Abstract

Photosensitivity is common in dermatological practice but photosensitive syndromes are much less common. Herein we report a case of Cockayne syndrome (CS) in a seven years old girl for its rarity.

## Introduction

Cockayne syndrome (CS) is a rare multi system genetic disorder with autosomal recessive inheritance which occurs in about one in 500,000 babies<sup>1</sup>. There are three different types of CS. Type 1 is the classic form and it appears early in life and by the end of second year it is obvious that the baby is very small, thin and developmentally stunted. Type 2 is also called cerebro-oculo-facio-skeletal syndrome (COFS), the most severe form and noticeable at birth. Type 3 is the mildest and rarest type which manifest later in life<sup>2</sup>.

## Case report

A seven year old girl was presented with a rash on face and forearms for six months and tendency to fall when walking for four months duration. She was the first child born to a non-consanguineous parents following an uneventful antenatal history. Her birth weight was normal with uneventful birth and postnatal period. Her developmental milestones were normal. She was blamed to be a slow learner with poor school performances. There were no significant medical problems among family members. When she was seven years old, she developed sun intolerance and erythematous scaly rash over face and forearms. Few months later mother noticed that the child had frequent falls without associated loss of consciousness. She was evaluated by the paediatric neurologist and referred for the dermatological evaluation. On examination she was emaciated with triangular face. Her height and weight were below third centile and she had microcephaly (Figure 1). There was no pallor or icterus. Dermatological examination revealed dry skin, erythema over malar region associated with scaly rash suggestive of photosensitivity. Same was seen in forearms. She had dental caries. Her hair, nail and mucosae were normal. Ear, throat and nose were normal. Oph-

thalmic examination showed reduced visual acuity bilaterally (Right eye 3/4.5, Left eye 3/4.5) with normal fundi. Examination of cardiovascular, respiratory systems and abdomen were normal. Musculo-skeletal system and neurological examination revealed hypotonia, over reactive reflexes with proximal muscle weakness manifest as Gower's sign.



**Figure 1.**

She had dragging of left foot with unsteady gait. At the end of history and examination possibility of photosensitive syndrome with a neurological involvement was raised. Photosensitive syndromes present in childhood include Porphyrias, Xeroderma pigmentosa, Bloom syndrome, Rothmund Thomson syndrome, Tricothiodystrophy and Cockayne syndrome<sup>3</sup>. Investigations ordered to come into a definitive diagnosis. Her routine biochemistry tests were normal. The patient was subjected to computed tomography (CT) of brain. It showed symmetrical basal ganglia and frontal subcortical calcification (Figure 3). We arrived at the diagnosis of CS in the

<sup>1</sup>Medical Officer of Dermatology, <sup>2</sup>Consultant Dermatologist, Sirimavo Bandaranaike Specialized Children's Hospital, Peradeniya, Sri Lanka.

presence of photosensitivity in a cachectic child with the calcifications of basal ganglia and sub cortical calcifications in the CT scan. Parents of the patients have been explained the prognosis of the disorder, natural cause of the disease, life expectancy and 25% possibility of inheritance to off springs<sup>4</sup>.

They were advised for the general care of patient according to the symptoms. Her four years old younger brother who accompanied her also found to have evidence of photosensitivity and he is currently under investigation (Figure 2).



Figure 2.

## Discussion

The CS is an autosomal recessive, multisystem disorder with male to female ratio of 3:1. Classical clinical features include abnormal photosensitivity, sensorineural deafness, retinitis pigmentosa, cachectic dwarfism, microcephaly and dental caries with progressive encephalopathy<sup>5</sup>. This syndrome often undetected in infancy and the progressive degene-



Figure 3. CT with basal ganglia calcification.

ration is manifested by the second or third year of life. Most of the classical features were noted in our patient except pigmentary retinopathy and sensorineural deafness.

The basic pathology is deficient DNA repair mechanism due to mutation in gene CSA/ERCC8 and CSB/ERCC6 located on the chromosome five and ten respectively<sup>6</sup>.

The neuropathologic changes include deposits of calcium in the vessels of the basal ganglia and the cerebrum. The CT scan of the brain in our patient showed symmetrical basal ganglia and frontal subcortical calcification.

There's no definitive treatment available for CS and our patient was treated symptomatically. Expected life expectancy may be up to 3<sup>rd</sup> decade of life which could not be prolonged by treatment<sup>6</sup>.

## References

1. <http://ghr.nlm.nih.gov/condition/cockayne-syndrome>
2. <http://www.ncbi.nlm.nih.gov/books/NBK1342/>
3. Spivak G. The many faces of Cockayne Syndrome. *Proc Natl Acad Sci USA* 2004; **101**: 15273-74.
4. Cleaver JE, Lan ET, Revet I. *Nat Rev Genet.* 2009; **10** (11): 756-68, Epub 2009 Oct 7.
5. Harper J, Oranje A, Prose N. *Textbook of Paediatric Dermatology* 2000 Ed.
6. <http://www.ncbi.nlm.nih.gov/books/NBK22190/>