Cockayne Syndrome

Arun Neupane, MD
Department of Pediatrics
Shree Birendra Hospital (Chhauni)
Kathmandu,Nepal

Shiv B. Basneyt, MD
Department of Radiology
Shree Birendra Hospital (Chhauni)
Kathmandu,Nepal

Jamuna Manandhar, MBBS
National Academy of Medical Sciences
Bir Hospital, Mahabauddha
Kathmandu, Nepal

Address for correspondence:
Arun Neupane, MD
Department of Pediatrics
Shree Birendra Hospital (Chhauni)
Kathmandu, Nepal
Email: arunneopane@hotmail.com

Received, June 12, 2005
Accepted, June 23, 2005

A case of Cockayne syndrome in a nine-year-old boy is reported. The clinical course of the case is described and pertinent literature is reviewed.

Key Words: Cockayne syndrome, congenital

Cockayne syndrome is a rare autosomal recessive, heterogeneous, multisystem disorder characterized by dwarfism, progressive pigmentary retinopathy, birdlike facies, and photosensitivity. This is the first case report of cockayne syndrome from Nepal

Case Report

This- nine-year-old male child presented to our outpatient department with a history of facial erythema in butterfly distribution exaggerated by sun exposure since two months of age and tremor of hands with unsteady gait since the age of seven years. He was seen elsewhere and was on triphenhexidil for one year with some improvement. The reminder of his history was non-contributory.

General physical examination revealed a thin built child with unusual progeroid facies. Face was characteristic with unusually large ears ("Mickey Mouse ears"); small chin with prominent, pinched nose ("birdlike"), and had sunken eyes (**Figure 1**). Face on general inspection looked aged or wizened, and there was associated microcephaly.

His musculoskeletal examination revealed a dorsal kyphosis. Ophthalmological and dental examinations revealed no abnormality except for the caries tooth.

A working diagnosis of Cockayne syndrome was made. The patient subsequently had his imaging studies. X-ray of his hands was normal. A computed tomography (CT) scan of brain was obtained which showed bilateral symmetrical basal ganglia calcifications (**Figure 2**). Magnetic resonance imaging (MRI) (not shown here) of his head revealed a large cisterna magna.

Discussion

This syndrome was first reported by Edward Alfred Cockayne,[1] a London- based physician in 1946. Since than more than 150 cases have been reported. However no such case has been reported from Nepal. The basic disorder in Cockayne syndrome has been localized to the nucleotide excision repair pathway, a process that is responsible for the removal from DNA of a variety of lesions, including those induced by ultraviolet light.[2]

The syndrome is divided into two subtypes; Cockayne syndrome I (CS-I), or classic Cockayne Syndrome which presents in childhood with characteristic facies and somatic features that occur late in the first decade of life. Cockayne syndrome II (CS-II), or severe Cockayne Syndrome, presents at birth with accelerated facial and somatic features. Individuals who are affected with CS-I typically have progressive neurological degeneration with death occurring by the second or third decade of life, whereas patients with CS-II typically die by age six or seven years. Abnormalities associated with this syndrome are: [3, 4]

Growth Retardation

Weight and height of patient is significantly less than the 5th percentile for age. This is true to our patient.

Characteristic Facies (Facial Appearances)

The patient has unusually large ears for head size ("Mickey Mouse ears"); there is small chin with prominent, pointed nose ("birdlike"); deficiency in adipose tissue ("fat") under skin, giving sunken eyes and skeletal appearance. Face overall may appear aged or wizened, and head size is likely to be unusually small for age and sex of the patient (microcephaly). These entire features were present in our patient.

Ocular Findings

These include retinitis pigmentosa, optic atrophy, strabismus, hyperopia, decreased lacrimation, nystagmus, corneal opacity, and cataract which were absent in our patient.

Dental Examination

Abnormalities included are caries teeth, delayed eruption of deciduous teeth, malocclusion, and absent/hypoplastic teeth.

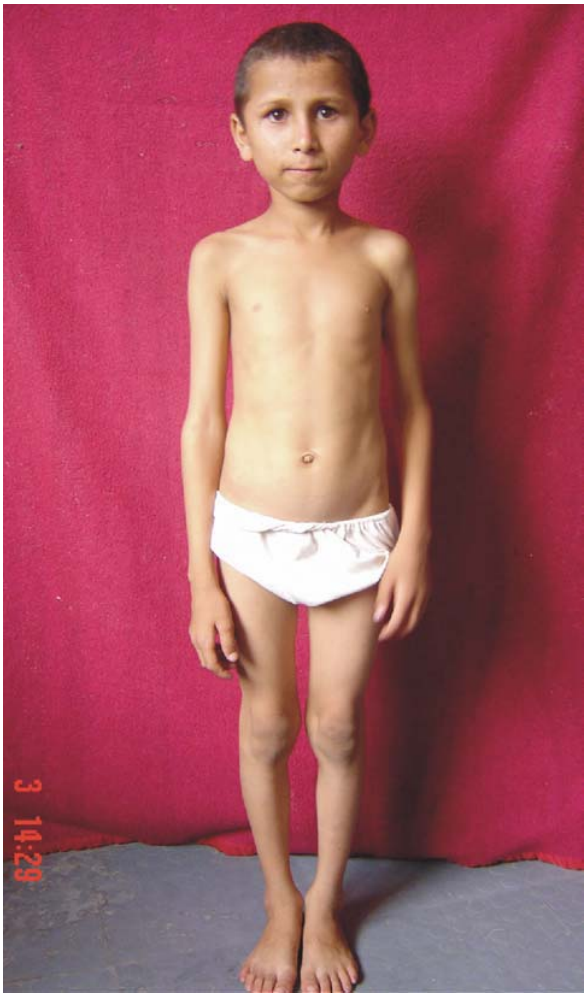

Figure 1. Patient with Cockayne syndrome.

Musculoskeletal System

Dorsal kyphoscoliosis which was present in our patient.

Skin Manifestations

These include photosensitive dermatitis which was there in our patient since two months of age.

Central and Peripheral Nervous System

Unsteady gait, ataxia, tremor, incoordination, and dysarthria are common features noted. Seizure is present in 5-10% of such patients.

Others

Presence of thin, dry hair is common. Sclerotic ivory epiphyses are most obviously seen in the fingers.

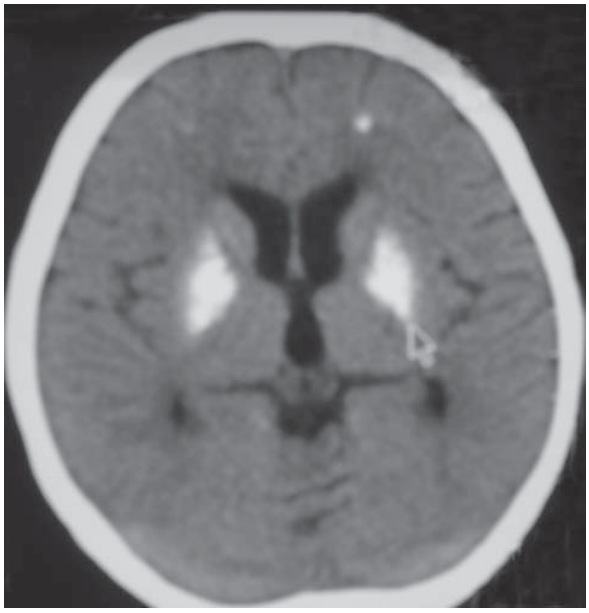

Figure 2. CT scan of head of the patient showing bilateral basal ganglia calcifications.

Histopathological examination of the brain generally shows patchy demyelination and atrophy of white matter and cerebellum. The brain can undergo perivascular calcification in the basal ganglia and cerebellum; these changes are termed a calcifying vasopathy.

The disease is usually progressive with time and treatment is generally supportive. Eventually a diffuse extensive demyelination of the peripheral and central nervous systems ensues, and patients generally die of atheromatous vascular disease before the third decade.

References

1. Cockayne E A: Dwarfism with retinal atrophy and deafness. **Arch Dis Child 21:**52,1946
2. Jones KL: **Smith's Recognizable Pattern of Human Malformation ed 5**. New York, WB Saunders Company, ed 5, 1997, pp 144-145
3. Nance MA, Berry SA: Cockayne syndrome: review of 140 cases. **Am J Med Genet 42:**68-84, 1992
4. Ozdirim E, Topcu M, Ozon A, et al: Cockayne syndrome: review of 25 cases. **Pediatr Neurol 15:**312-316, 1996