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Cockayne Syndrome as a Rare Cause of Hemiplegia: Review of the Literature Accompanied by a Case Report

Nadir Bir Hemipleji Nedeni Olarak Cockayne Sendromu: Bir Olgu Sunumu Eşliğinde Literatürün Gözden Geçirilmesi

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Abstract

Cockayne syndrome is a rare genetic disease that presents with growth retardation, premature aging, retinal and generalized neurologic abnormalities. The presented case is a 29-year-old male patient, who was previously diagnosed as having Cockayne syndrome and who was admitted to the physical medicine and rehabilitation outpatient clinic with loss of strength in the right upper and lower extremity. The patient was detected to have acute-onset, non-traumatic right-sided muscle weakness secondary to acute subdural hematoma diagnosed in cranial computed tomography imaging at admission to emergency care department 8 months ago. When the patient was evaluated by us, he had a chronic stage subdural hematoma. Chronic subdural hematoma is a disease of elderly and aging is a well-known risk factor. Our case demonstrates that the disease of old age, chronic spontaneous subdural hematoma, can be seen in a very young age secondary to premature aging syndrome known as Cockayne syndrome.

Keywords: Cockayne syndrome, hemiparesis, subdural hemorrhage

Öz

Cockayne sendromu, karakteristik klinik özellikleri gelişme geriliği, erken yaşlanma, retinal ve yaygın nörolojik anormallikler olan genetik bir hastalıktır. Olgumuz, fizik tedavi ve rehabilitasyon polikliniğine sağ üst ve alt ekstremitelerinde güçsüzlük ile başvuran, başvurusundan uzun zaman önce Cockayne sendromu tanısı almış 29 yaşında bir erkek hastadır. Sekiz ay önce, travmaya bağlı olmayan akut başlangıçlı sağ taraf güçsüzlüğü ve yürüme zorluğu ile acil servise başvuran hastanın kraniyal bilgisayarlı tomografisinde akut subdural hematom saptanmıştır. Olgu tarafımızca değerlendirildiğinde kronik evre subdural hematomu mevcuttu. Kronik subdural hematom esas olarak yaşlı bireylerde görülen bir hastalık olup, ileri yaş majör risk faktörlerinden birisidir. Olgumuz, esasen yaşlı bireylerin bir hastalığı olan kronik spontan subdural hematomun, erken yaşlanma sendromu olan genç bir bireyde de olabileceğini göstermektedir.

Anahtar Kelimeler: Cockayne sendromu, hemiparezi, subdural hemoraji

Introduction

Cockayne syndrome was first described by Cockayne (1) in 1936. Kleijer et al. (2) detected the incidence of the syndrome as 2.7 per million live births in the Western European population. Characteristic clinical findings are growth retardation and neurologic abnormalities with other common clinical findings like sensorineural hearing loss, cataract, pigmentary retinopathy, cutaneous photosensitivity, and early tooth decay (3). Moreover, as it is a premature aging syndrome, other neurologic abnormalities, mental retardation secondary to developmental microcephaly, progressive brain volume loss, and dementia are very common (4). The most commonly encountered neurodiagnostic findings are calcifications in basal ganglia detected in cranial computed tomography (CT) and white matter abnormalities secondary to dysmyelination detected in cranial magnetic resonance imaging (5).

Case Report

A 29-year-old male patient who was diagnosed as Cockayne syndrome in childhood was admitted to physical medicine and rehabilitation outpatient clinic with symptoms of right upper and lower extremity muscle weakness. His past medical

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history included term, uncomplicated birth, and motor growth retardation beginning at the ages of 3 to 4 years. He was diagnosed as Cockayne syndrome with characteristic facial appearance, photosensitivity, dwarfism, muscle rigidity, joint contractures, and, xerodema pigmentosum in skin biopsy. He also had a past medical history of hypertension, loss of vision for the last 4 years, hearing loss for the last 3 years, and 50 pack-year smoking. Eight months ago, he was admitted to the emergency care department with the symptoms of headache, difficulty in walking, right-sided extremity weakness, and then acute subdural hemorrhage was detected in cranial CT. He had no history of trauma and had increased blood pressure (180/110 mmHg) at the time of admission. The patient was admitted to intensive care unit and was followed up without surgery.

Physical examination revealed height of 126 cm, weight of 25 kg, and characteristic findings of Cockayne syndrome including microcephaly, short stature, and xeroderma pigmentosum. In the neurologic evaluation he was conscious and anxious in mood, his speech was slow, slurred, and difficult to understand, and his comprehension level was normal. Cranial nerve examination was normal with muscle strength of 4/5 in the right upper and lower extremity, with accompanying spasticity on the same side of the body. The patient was evaluated as stage 2 according to functional ambulation scale and Brunstrom staging was 3 for all parts (right upper extremity, right hand, and right lower extremity). Stage 3 spasticity was detected in the right shoulder adductor muscles, right pronator teres, right finger flexors, right ankle plantar flexor muscles according to the Modified Ashworth scale. The Barthel index was calculated as 30 points. The first cranial CT performed 8 months ago revealed early subacute subdural hematoma around 36 mm in width leading to a midline shift (Figure 1). The second cranial CT performed 6 months ago showed bilateral subdural hematoma, and calcifications in the cerebellum, and occipital regions of the brain (Figure 2, 3). Cardiological examination, cardiac echocardiography, carotid, and vertebral artery Doppler ultrasound revealed no abnormality. Nerve conduction studies were within normal limits for right upper extremity nerves, whereas sural responses could not be detected, and nerve conduction

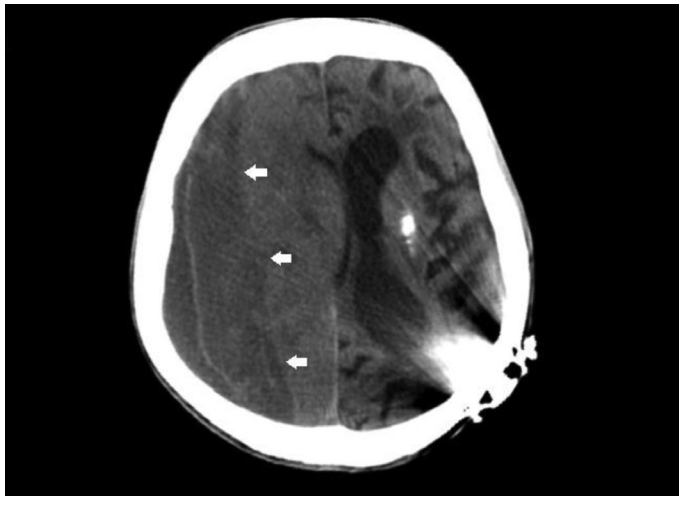

Figure 1. Early subacute subdural hemorrhage and midline shifting in brain computed tomography at the first hospital admission

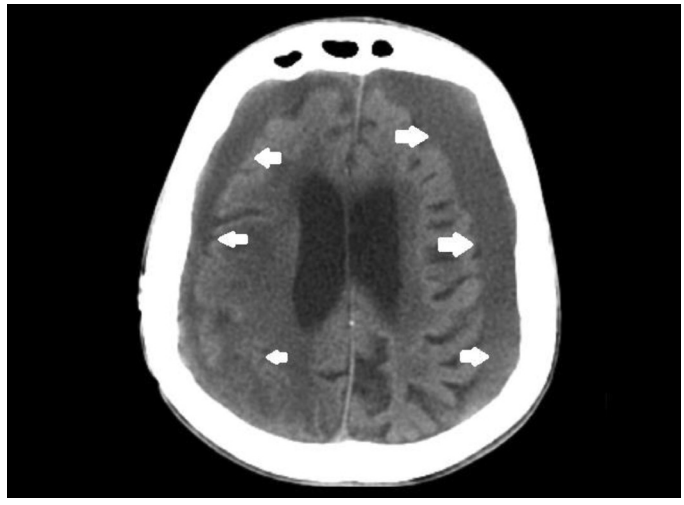

Figure 2. Bilateral chronic subdural hemorrhage 2 months after the first admission

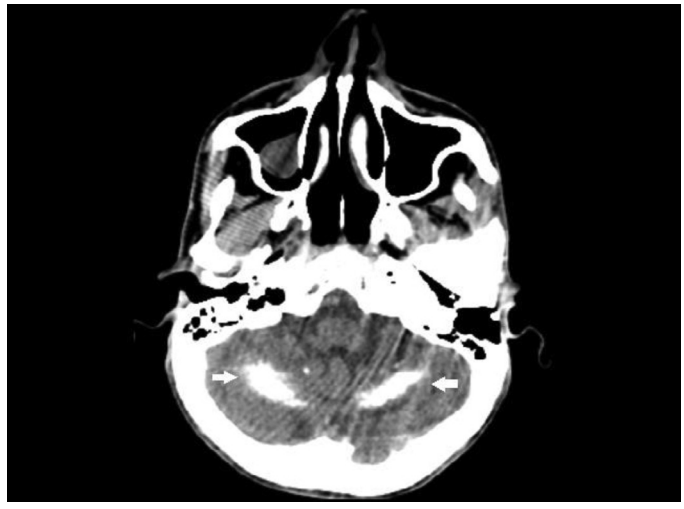

Figure 3. Bilateral cerebellar calcifications

blocks and decreased conduction velocities were detected in the tibial and peroneal nerve evaluations. Bilateral pes equinovarus deformity was detected. All clinical findings supported the diagnosis of hereditary demyelinating neuropathy, but there was no family history for polyneuropathy and foot deformity. The patient was treated through a rehabilitation program.

Discussion

We searched for "Cockayne syndrome" and "Cockayne syndrome-associated hemiplegia" in PubMed, EMBASE, the Cochrane Database, and Google Scholar without time limitations. We analyzed all publications about the relationship between Cockayne syndrome and hemiplegia. We found only two case reports. The first described a patient who presented with acute gait difficulty, cerebellar ataxia, rigidity, hyperreflexia and cranial imaging showed bilateral subdural hematoma, calcifications and atrophy of the brain, cerebellum, and brain stem (6). The second

described a patient who presented with right hemiparesis, which resolved in 4 days following thrombolytic treatment. Brain imaging did not show any abnormalites but stenotic plaques were detected in cerebral angiography (7). Many different vascular changes can be encountered in patients with Cockayne syndrome. Pathologic studies showed increased numbers of small arteries and arterioles filled with fibrotic tissue without atherosclerosis in the subarachnoid space, and a decrease in brain mass, which may also lead to increased amount of vasculature (8). Chronic subdural hematoma is a disease of the elderly and aging is one of its major risk factors (9). Recently, it was postulated that formation of new vessels (angiogenesis) could be a source of bleeding in chronic subdural hematomas (10). As a result, our case report describes how chronic subdural hematoma, which is known as a disease of elderly, can also be encountered in young patients with premature aging syndromes.

Ethics

Informed Consent: Consent form was filled out by all participants.

Peer-review: Internally peer-reviewed.

Authorship Contributions

Surgical and Medical Practices B.Ş.İ., O.B., Concept B.Ş.İ., O.B., Design B.Ş.İ., O.B., Y.S., Data Collection or Processing: B.Ş.İ., Analysis or Interpretation: B.Ş.İ., O.B., Y.S., Literature Search: B.Ş.İ., O.B., Y.S., Writing: B.Ş.İ., Y.S.

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